Textbook of Psychiatry for Intellectual Disability and Autism Spectrum Disorder

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Foreword

People with Intellectual Disability (ID) are a diverse group of people with individual needs. Aetiological factors associated with ID have an impact on the origin and course of mental health problems and on the type of therapeutic interventions that would be most appropriate for them.

Historically, it was believed that people with ID were being incapable of having mental health problems. However, epidemiological studies over the last few decades consistently disproved this, showing that people with ID have a higher frequency of mental health problems than the general population. The occurrence of mental health problems in people with ID varies according to methodology used for their estimation. An increased vulnerability to mental health problems for people with ID is likely to be due to an increased incidence of brain abnormalities including associated epilepsy, physical and sensory problems, social and psychological hardships, and misfortunes. The severity of ID, adaptive behaviour and social skills deficits are also related to the presence of mental health problems for people with Autism Spectrum Disorders (ASD).

The current textbook of "Psychiatry for Intellectual Disability and Autism Spectrum Disorder" is a welcome addition to the few existing textbooks related to mental health problems for people with ID. Written by a large group of international multi-professional contributors and edited expertly by experienced editors, led by the recognised expertise and proficiency of Dr Marco Bertelli, under the auspices of the World Psychiatric Association (WPA), presents the up-to-date evidence base in the field. Dr Marco Bertelli is credited with his long-standing systematic contribution for the enhanced involvement of the World Psychiatric Association to mental health problems of people with Intellectual Disability. He was fellow at the 12th (Yokohama 2002) and 13th (El Cairo 2005) WPA World Congress, chair of the WPA Section on Psychiatry of Intellectual Disability and WPA Lifetime Honorary Member since the 15th World Congress (Buenos Aires 2015).

The authors of the different chapters provide arguably a worldwide perspective of the research summarising the key topics in the mental health of ID field. A distinct difference of the current textbook includes several contributions on conceptual, cultural, ethical and spiritual issues as well as human rights and broad health issues relating to people with ID. There are also important contributions relating to people with ASD who are known having a high prevalence of comorbid anxiety, obsessive compulsive and attention deficit hyperactivity disorders (ADHD). In reviewing each area, the authors have included recent advances, and many have offered pointers to further progress. In addition, this book is a comprehensive resource for clinical practice including those underpinning assessment, management and service delivery.

The process of deinstutionalisation and the development of community care of ID services, which took place in the latter part of the 20th century, brought up the complexities of meeting the mental health needs

of people with ID. The introductory chapters on definition, new terminology of Intellectual Developmental Disorders, nosology, epidemiology and prevalence offer insight into the underline debate and conceptual concerns. Considerable improvement has been documented for the assessment and diagnosis of mental health problems for people with ID. This is highlighted in this volume by leading experts of international recognition. Reliable tools and methods that have been developed and are used in clinical practice and research for the assessment and diagnosis of mental health problems in both adults and children with ID are lucidly described.

The aetiology, symptomatology and treatment methods of the core psychiatric conditions are presented on an updated evidence and practice base. The controversy on psychiatric versus behavioural problems or challenging behaviour remains a thorny issue that is further explored. Challenging behaviour, such as aggression, is often the primary reason people are referred to ID and mental health services, and they can often fluctuate over years and be chronic in duration. The overlap between psychiatric conditions and challenging behaviours in people with ID and low-functioning autism should neither be over nor under-played because it has significant clinical and service implications. In clinical practice, it is not always possible to precisely separate psychiatric conditions from behavioural problems in people with ID. It is likely that there is a combination of biological, social and environmental factors interacting with cognitive and adaptive deficits to make people with ID vulnerable to mental health and behavioural problems.

As the population of ID survives longer into old age, aspects of mental health previously not considered relevant have become more prominent. For example, in adults with Down syndrome who now are surviving well into their sixties, a considerable percentage at the age of 50 years have a clinical diagnosis of Alzheimer's disease. This issue is also discussed from research and clinical points of view.

The need for a bio-psychosocial approach, including coordinated multidisciplinary input, is of paramount importance. After decades of community care, unanswered question remains about the appropriateness of generic mental health services versus specialists services for meeting the mental health needs of people with ID. The arguments and the existing evidence base in favour and against mental health systems are rehearsed in this volume and should be of help to policymakers, practitioners and researchers. The number of possible interfaces between services is increasing. Together with existing uneven financing systems, these interfaces are increasingly struggling to manage personalised care pathways adjusted to the needs of people with ID and mental health problems, their careers and families.

The needs of people with ID and offending behaviour, several of whom having also additional mental health problems was overlooked with the deinstitutionalisation. Service provision has lagged developments for local services in countries such as UK, resulting in many people with ID and forensic needs to have been placed in residential facilities a long way from their local communities and at times in unnecessary restrictive environments. The authors of the relevant chapter, having

extensive knowledge and experience, suggest that care for people with ID and forensic needs requires strong interfaces and partnership between ID, mental health and forensic services.

The concluding chapters of the book refer to cultural, spiritual and human rights issues. Cultural aspects vary together with deep-rooted traditions and values that are reflected in the quality of care for people with ID and or ASD. Furthermore, people with ID have the right to spiritual expression and practice of beliefs. The protection of human rights is of utmost importance where prejudices and barriers are removed and everyone can live without being stigmatised.

A strategic approach needs to be adopted to ensure effective person-centred services where there are care pathways that connect health and social care, support and education services as when a person might need these services. Despite progress, there is still a need for clear interfaces with services, joined-up working between different agencies, proactive commissioning strategies, development of academic centres and further research into service models and outcomes.

This volume presents an evidence-based comprehensive publication for the mental health problems of people with ID and or ASD. An obvious question is how the outcomes produced by research can be translated for the benefit of people with ID, their families and their careers. The transformation of care for people with ID over the last half a century includes the movement towards integration, participation and choice as well as civil and human rights movements on a national and international level. There is now a consensus on the need to respond more adequately to mental health needs in this population. The current trends are geared towards community integration and schemes with service users' participation at all levels, including design and implementation by using a person-centred approach. Despite the existence of policies and services for people with ID, recognition of and provision for their mental health needs carry low priority. This gap is greater in low and middle-income countries, where the burden of disability needs to be recognised to plan for and meet the needs of those with developmental and intellectual disabilities across their lifespan. It remains to be seen how people with ID experience the reality of equality and absence of discrimination and whether stigma and negative attitudes by others will be eliminated.

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Preface

Since I entered University to become a medical doctor, I have always loved research. I think research is fundamental for clinicians to the same extent to which clinical challenges represent a base for research. Medical research is also a unique opportunity to identify or operationalise things that can improve humans' health and quality of life.

Halfway through my training in medicine, I felt that most of the medical specialisations would not have met my interest in philosophy and humanities developed during my previous studies at the high school, but fortunately I discovered psychiatry as a discipline implying a complex approach to human suffering and thus able to combine all my interests. I thought that entering the mind of persons with schizophrenia or other major psychiatric disorders and helping them to recover could be the most satisfactory thing I could ever do as a medical doctor. Thus I was very disappointed when in response to a request for a topic for my graduation thesis, the Professor of Psychiatry imposed me a research on intellectual disability, mental retardation at that time, which was largely considered the "Cinderella of psychiatry".

Nevertheless, I was looking forward to working as a psychiatrist in a real clinical way and my main desire was to complete my training and to graduate, so I decided to accept. I can now say that this apparently unlucky obligation was one of the greatest opportunities of my life. In fact by entering the field of mental health of intellectual disability, I increasingly realised that I was getting in touch with the most complex pathogenic and phenomenic aspects within psychiatry and the best chances to help other persons to soothe their mental suffering and bring a contribution in life to make a better world.

In the last century, intellectual disability and autism have been neglected by psychiatry for a long time and still are, although in the previous century psychiatry had a well-established and well-documented role in promoting an appropriate care for what was called mental retardation. According to Frank Menolascino, who can be considered one of the founders of modern psychiatry of intellectual disability, this withdrawal resulted "in a number of stereotyped views or blindspots …, that are: uncritical acceptance of mental age as an adequate description of a person; treatment nihilism … associated to the overriding focus on chronicity and irreversibility; and excessive focus on the severely retarded in contrast to the mildly retarded".

In the last 15 years, the interest in autism has grown a lot in the psychiatric field but not that for people with the greatest need of support, such as those with minimal verbal ability or low IQ, to whom the main attention is given in this textbook. In most countries across the world, persons with intellectual disability and low-functioning autism still face great difficulties in accessing mental health service and in receiving adequate assessment and care. Specific training of mental health professionals is also lacking, at both undergraduate and graduate levels.

This neglect has always been unfair and inappropriate to me. Unfair because persons with intellectual disability and low-functioning autism

would deserve, as persons, at least the same attention as all other persons, but even more for their higher difficulties and risks in daily life and higher vulnerability to the full range of psychopathological features.

Many of the current limits to an appropriate mental health care and more generally to community inclusion are linked to the conceptualisation of intelligence, as a unitary capacity which defines human nature in its most important transcendental component. The persistent IQ reduction confines persons with intellectual disability into a condition of eternal sickness or eternal childhood as well as into a persistent position of inferiority and unworthiness, which many persons with intellectual disability end up identifying with. In reality, it has never been demonstrated that unitary intelligence is an entity of nature and not a mere human construction. Nor has it ever been proven that the mind and the body represent two dichotomous realities.

The study of the human psyche has been characterised for many years by a clear separation between body and mind but also between affectivity/emotionality and cognition/intelligence. In traditional cognitivism, body processes were seen essentially as by-products of cognition, and as too little specific to be able to contribute to the variety of emotional experiences. The embodied vision of the mind has not yet been adopted in theories of emotionality, despite the fact that the body has always occupied an important place in the study and characterisation of emotions, from Darwin onwards. To date, the level of knowledge on the inter-relationship between emotionality, corporeality and cognition absolutely does not allow us to consider people with intellectual disabilities and autism as limited in their humanity.

In addition to being unfair, the scientific community's neglect of the mental health of people with intellectual disabilities and low-functioning autism has also been inappropriate because the development of scientific knowledge in this area has general important implications for the entire neuroscientific field, such as the understanding of the link between early specific cognitive deficits and psychopathological vulnerability, the definition of the grade of adjunctive functional impairment and clinical distress associated with the co-occurrence of psychopathological conditions, the possibility to identify psychiatric symptoms in patients with cognitive and communicative limits, basing on observable and behavioural changes from the baseline, and to distinguish them from the features characteristics of the developmental conditions. Furthermore, psychiatry of intellectual disability and autism is looking for the most useful way to combine developmental/idiographic (narrative) and nomothetic approaches to mental suffering and to implement person-centred outcome measures, such as generic quality of life. Even models of care (residential care, respite care, multidisciplinary approach to care, etc.) and social issues of health (stigma and labelling, self-advocacy, etc.) that were first developed in the intellectual disability field are now widely used in general psychiatry and other neuroscientific disciplines. Many contributions are also being brought on how individual cognitive, emotional, behavioural and relational characteristics emerge and change across early and lifetime development of the brain, and how they can be altered and manifest as mental health conditions or mental health

problems. Recent findings in neuroscience display that many of what have been traditionally considered to be distinctive forms of psychopathology have features in common with one another, co-occur, present in the same person across the life span or represent age-adjusted variations of common underlying dispositions. Genome-wide association studies increasingly show that copy number variants of several syndromes including intellectual disability and autism are present also in many other major psychiatric disorders such as schizophrenia, bipolar disorder and major depressive disorder. Most developmental, psychiatric and neurological disorders have recently been proposed to be part of a unique group of disorders affecting neurodevelopment.

Thus, the present textbook places intellectual disability and autism at the very centre of neuroscience in general and psychiatry in particular, anticipating a position that will soon be embraced by the entire scientific community.

People with intellectual disabilities and autism are an asset to the world community on many levels. On a conceptual level, because they value the diversity and fragility foreseen by nature for the neuropsychic system of the human being, on a concrete level, because in the right context they can make their own contribution to society, and on an evolutionary level, because they help the humankind to overcome its limits and to acquire increasing capacity for attention and balance with the essential aspects of their existence.

This textbook represents for me a major milestone in the long path of research and clinical practice. Its preparation spanned over 4 years, amidst numerous difficulties, unforeseen events, the COVID-19 pandemics (after which we added a chapter focusing on teletraining and teleassistance) and the need to keep updated the chapters already completed. This book was inspired by the will of sharing knowledge and transmitting passion to colleagues, especially young and future colleagues. In fact, it is intended for use by graduate students and trainees of university faculty, practitioners in clinical disciplines or management roles in developmental disabilities services and education, and to a lesser degree, undergraduate students, parents, attorneys and advocacy groups. Faculty will find this book particularly useful as a primary course text at the graduate level. Researchers will find the coverage contained herein useful for a summary of current knowledge about a subarea of psychiatry of intellectual disability and low-functioning autism that is new to them or that intersects their own specialty in the wider field of developmental disabilities. Practitioners and educators can use the textbook as a resource to point to when there is a doubt or a debate regarding diagnostic issues, appropriate treatments or intervention practices.

The overall purpose of this textbook is to provide readers with a complete and up-to-date overview of the state of knowledge in the field of psychiatry for persons with intellectual disability and autism.

I have invited the wide majority of most authoritative authors to contribute to the textbook in order to provide readers with the very best and up-to-date knowledge, and to reflect the growing awareness of the mental health needs of people with intellectual disability and autism across many countries and cultures.

Authors were asked to yield foundational conceptual information on their assigned topic, to identify valuable studies in the related literature, to emphasise the latest research findings and to provide clear conclusions and interpretations.

There is variation across the chapters in terms of length and focus, which is mainly a reflection of the evidence base available on specific issues. I have also allowed some overlaps, as for the numerous and strong links between the different aspects of mental health in this field and an interdisciplinary approach.

Not all the issues concerning the complex field of psychiatry of intellectual disability and autism have been deepened as I would have liked. I hope that I will still be given time and energy to produce increasingly precise and useful updates and insights in the coming years. Nevertheless, as far as my knowledge, the present work represents the most comprehensive and up-to-date textbook worldwide in the field of mental health of neurodevelopmental disorders, with the highest number of chapters and eminent authors from across the world. It has been realised under the aegis of the World Psychiatric Association within its 2021–2023 action plan.

It can be considered an essential reading also for future prospects, as it displays how we started moving into a new century of discovery, evolution, and high-quality care for people with intellectual and developmental disabilities.

Marco O. Bertelli, Leading Editor
Florence, Italy

Acknowledgments

This textbook has been realised thanks to the precious contribution of a stellar group of world-renowned authors, 116 leading experts in the field of mental health of intellectual and developmental disorders from around the world. I am deeply grateful to all of them for their diligence in writing the chapters and for their willingness to accept my recommendations, integrations and various other requests along the way. Special thanks go to Dr. Shaun Gravestock, who worked on his part until a few days before passing away from COVID-19, and his partner, Darren Ward, who took care to find and pass on Dr. Gravestock's work to me in times of extreme grief.

Thanks also to the co-editors, Professors Shoumitro (Shoumi) Deb, Angela Hassiotis, Luis Salvador-Carulla and Kerim Munir, who have generously shared their insights, suggestions and thoughts. Particular appreciation goes to Professor Shoumi Deb, who has been remarkably active and has provided huge unwavering support at the times when it was needed most. The making of this textbook took more than five years, but it would have taken even longer without him.

The present textbook represents an important outcome in my long path through the field of psychiatry of neurodevelopmental disorders, which started with Dr. Giampaolo La Malfa and Professor Pier Luigi Cabras at the Operative Unit of Psychiatry, Department of Neurological and Psychiatric Sciences of the University of Florence, Italy, during which I met many people who have fuelled my passion, informed my knowledge and inspired my research and clinical activities. I want to thank them all.

My career in the World Psychiatric Association (WPA) has begun in 2001 at the Antalya (Turkey) WPA Regional Meeting, where I delivered a speech in substitution of Professor Giovanni Stanghellini and where I met Professor Juan Mezzich who encouraged me to apply for a fellowship for the 2002 Yokohama World Congress (Japan). I am deeply grateful to Professor Mezzich for this opportunity and for his constant presence during my WPA career. During the Yokohama fellowship and the following one at El Cairo, Egypt, I had the extraordinary opportunity to be trained by leading international experts and to share opinions and experiences with young colleagues from all around the world. I could also progress my activities within the WPA Section "Psychiatry of Intellectual Disability" which I had started few years before the Antalya congress. Professor Nick Bouras and Professor Luis Salvador-Carulla have been my guides within the Section and other related assignments. I want to thank them for the knowledge and the constant support they provided to me during my tenure as the Section chair and the other commitments within the WPA, including the creation of this textbook. I received an extraordinarily intense albeit brief contribution from Professor Ludwik Szymanski in a meeting with him that represented a physical contact with the origins of the Section and the history of psychiatry of intellectual disability.

Thanks also to Professor Mario Maj, the WPA President during 2008–2011, and his Executive Committee, who awarded me the WPA lifetime honorary membership at the end of the first of my two terms as Section chair and who helped to sustain my enthusiasm to write this textbook. Other WPA leadership figures who have shown respect for me and whom I want to thank are Professor Norman Sartorius, the WPA President during 1996–1999 who was my mentor all along my WPA training, Professor Levent Kuey, the WPA Secretary General during 2008–2011, and more recently Professor Afzal Javed, the current WPA President, who has included mental health needs of people with neurodevelopmental disorders in the priorities of the 2020–2023 WPA action plan and who entrusted me with the chairing of the working group on autism spectrum disorder, one objective of which is to produce this textbook. I also thank Professor Michel Botbol and Professor Norman Sartorious for their particular interest in the production of this textbook and highlighting its cultural and scientific importance on the world scene.

Another part of the knowledge and passion that I have put into this work has developed thanks to the valuable persons I met along my journey in the European Association for Mental Health in Intellectual Disability (EAMHID) such as Anton Dosen, founding father of the association together with Professor Nick Bouras, Germain Weber, Roger Banks, Herman Woiters (my super-efficient and loyal secretary during my presidency and a person of extraordinary kindness and moral fiber), Jane McCarthy, Johan DeGroef, Raymond Ceccotto and Tanja Sappok. I would like to express further thanks to Tanja for protecting the importance and visibility of this textbook by refusing the invitation of various publishers to write a similar book. Intellectual honesty and corporate spirit of this kind are hard to find these days.

My ideas and arguments have also benefited from enormously helpful feedback I have received in presenting parts of these textbooks at the EAMHID International Congress I organised in Florence in September 2015 and at the last three WPA World Congresses, in Mexico City, Lisbon and Bangkok (virtual). The rich ideas and discussions at these meetings have broadened my scientific and cultural horizons and have given me a new critical perspective on my work.

My understanding of neurodevelopmental disabilities, both as lived experience and as an area of academic and clinical practice, has been greatly enhanced through the dialogues and interactions with persons with intellectual disabilities and/or autism spectrum disorders and their family members as well as through the conversations with professionals and scholars within and beyond the mental health field. I want to acknowledge Professor Giovanni Stanghellini for his extraordinary insights and innovative perspectives in psychopathological phenomenology and philosophy, Dr. Carlo Francescutti, whose views increased my research attention to the interdisciplinary working in daily practice and whose passion for this field continues to encourage me, Dr. Maria Luisa Scattoni for her extraordinary neuroscientific knowledge that ranges from basic research to the multidisciplinary organisation of interventions and services, Dr. Serafino Corti for his invaluable assis-

tance and involvement in both the reform of national scientific societies and the increasing strengthening of the activities that emerged, and Professor Ivan Brown, who introduced me to the deeper dimensions of whole-person quality of life and their importance in evaluating the outcomes of any intervention. I also want to offer special thanks to two fellow psychiatrists who I am honoured to count among my closest friends, Dr. Michele Conte, whose great culture, critical spirit, and "visionary" skills have challenged my convictions and stimulated important improvements on several occasions, and Dr. Michele Mancini, whose ample experience in clinical research has animated many productive discussions on some of the key topics of this work.

I am fortunate to have found in the last decade welcoming intellectual home at "Fondazione San Sebastiano della Misericordia di Firenze", whose General Director, Dr. Leandro Lombardi, and the whole board of directors shared my belief that clinical research, detection of needs and dissemination of new knowledge represent a greater gift to others than material goods. Thanks to Dr. Alessandro Burberi, President of the San Sebastiano Foundation, and all the members of the scientific committee of CREA (Research and Clinical Center), who have always appreciated the production of this textbook and eagerly awaited its realisation.

Almost eight centuries have passed since "Misericordia di Firenze" was founded, during which it has never interrupted its mission of charity and solidarity, constantly trying to keep up with the times without disrespecting the historical and social context in which it was called to operate, blending modernity and tradition. Special thanks go to Dr. Andrea Morino and Dr. Alvise Revedin, respectively Vice-secretary and Secretary of "Misericordia di Firenze", who strongly supported the establishment of CREA and valued its activities, including the realisation of this textbook.

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Thanks to my family, especially to my wise and loving parents, Piera Bonciani and Giancarlo Bertelli, and to my grandmothers, Genny Mori and Laurina Lippi. This book gives voice to ideas and convictions that I learned from them and that I have come to appreciate in sharing my life with them such as the belief in the beauty and dignity of human beings, the importance of empathy, kindness and respect, the value of loving relationships, and the possibility of hope and improvement. Also, their teachings on rising from falls, never complaining and persevering have been particularly helpful in this endeavour. I hope that this and my other publications will at least partially fulfill their wish for me to make a contribution to the world that could go beyond the time length of our lives.

Thanks to my loved ones and friends who have tolerated my absence over the years of working on this book and in the many others dedicated to mental health research in neurodevelopmental disabilities.

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Marco O. Bertelli

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About the Editors

Marco O. Bertelli, MD is the scientific director of the Research and Clinical Centre (CREA) of Fondazione San Sebastiano, Misericordia di Firenze, Florence, Italy. He acted and still acts as a consultant psychiatrist to various service providers for people with neurodevelopmental disabilities and mental health problems. He worked as an adjunct professor at the University of Florence, and taught and still teaches in several Italian and international courses. Dr Bertelli is the current president of SIDiN (Italian Society for Neurodevelopmental Disorders) and past president of both the Psychiatry of Intellectual Disability section of the World Psychiatric Association and the European Association for Mental Health in Intellectual Disability. For the World Psychiatric Association, he is currently chairing the Working Group on Autism Spectrum Disorder. He has been a member of the Working Group on Classification of Intellectual Disabilities of the WHO International Advisory Group for the revision of ICD-11. For the World Health Organization is also worked within the Development Group for Autism Spectrum Disorders to the creation of the WHO Package of Interventions for Rehabilitation. He is an editorial board member of some scientific journals in the field, such as the *Journal of Intellectual Disability Research*, and author of around 200 journal articles and book chapters. Since 2017, Dr Bertelli is member of the scientific committee of the Italian Observatory for Disabilities. In 2011, he was awarded with the Honorary Membership of the World Psychiatric Association for excellence in service, and in 2021, he received the Leon Eisenberg Award of the Harvard Medical School for outstanding leadership and stewardship in the field of neurodevelopmental disorders.

Shoumitro (Shoumi) Deb, MBBS, FRCPsych, MD is Visiting Professor of Neuropsychiatry at the Imperial College London, UK. Prior to that, he was a substantive full time Clinical Professor of Neuropsychiatry at the University of Birmingham, UK, for many years. He led the development of the International (*World Psychiatry,* 2009,8(3),181–186) and the National Guidelines on the use of medication for problem behaviour among adults with intellectual disabilities and produced the first ever freely available online accessible psychotropic medication leaflets (► http://www.ld-medication.bham.ac.uk). He led the development of the first ever European guideline for the assessment and diagnosis of mental disorders among adults with intellectual disabilities which has recently been updated (*European Journal of Psychiatry*, 2022,36,11–25). He was part of the group that developed the first ever evidence-based practice guidelines in intellectual disabilities in the UK NHS. He developed a screening instrument for dementia in people with intellectual disabilities (DSQIID), which has been translated into more than 24 languages and validated for worldwide use. He developed the first-ever online training programme, SPECTROM (► https://spectrom.wixsite.com/project), for support staff to help reduce overmedication of adults with intellectual disabilities. He has around 300 publications (citation index: 7357, h-index: 46, i10 index: 96); done over 250 conference presentations nationally and internationally, including keynote speeches and chairing of sessions; received research grants from the UK NIHR, Wellcome Trust and other charities; led a research/teaching team of over 14 staff; led several masters' courses in UK universities (neuropsychiatry, epilepsy, psychological medicine etc.); and was the co-chair of SPID WPA, a member of the WHO ICD-11 workgroup on intellectual disabilities and a member of several NICE guideline development groups in the UK.

Kerim Munir, MD, MPH, DSc, DFAACAP is Director of Psychiatry at the University Center for Excellence in Developmental Disabilities in the Division of Developmental Medicine, Boston Children's Hospital, and William A. Hinton Society Fellow in Global Health at Harvard Medical School, Boston, Massachusetts, USA. His research focuses on autism spectrum disorder (ASD) and intellectual developmental disorders (IDD) across the lifespan, with an emphasis on understanding determinants of developmental psychopathology and comorbidity. He served as a member of the WHO International Classification of Diseases, 11th edition (ICD-11) Review Group on Intellectual Disabilities, as well as the WHO Development Group on Rehabilitation 2030. He chairs the World Psychiatric Association (WPA) Section on Psychiatry

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Intellectual Disability/ Intellectual Developmental Disorder

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Learning Objectives

This chapter describes the evolution of the construct of intellectual development disorder from its earliest historical descriptions to the current diagnostic criteria. It also includes a detailed treatment of adaptive functioning, intelligence, and specific cognitive functions, as key aspects for an effective dimensional approach to this clinical condition. A specific section will concern the interdisciplinary issues of the multidimensional assessment.

1.1 Introduction

Intellectual disability (ID), intellectual developmental disorder (IDD) or Disorders of Intellectual Development (DID), according to the most recent terminology [1, 2], is a common condition with consequences across the lifespan, including a considerable burden on families and caregivers, and high need for service provision. ID/IDD is the leading condition in health and societal costs, not only in the mental health sector, but in all medicine, at least in the Western countries [3–5]. In spite of its global burden, ID/IDD has a non-prominent position within psychiatry. Training in ID/IDD is not included in the psychiatric curriculum of many countries, and the majority of psychiatrists are not prepared to deal with the specific health needs and demands of persons with this condition [6].

1.2 Historical Evolution

Intelligence and ID/IDD might not be a natural kind but the result of a historically contingent human attempt of self-definition and self-characterization [7].

The earliest descriptions of ID date back to ancient Egypt. Two papyri of the sixteenth century BCE, the Edwin Smith papyrus and the Ebers papyrus, document early treatment of wounds, surgical operations, and identify, very likely for the first time, the brain as the site of mental functions. These papyri also show that, despite innovative thinking about disease, magic and incantations were used to treat ID/IDD and other mental conditions that were of unknown origin, often thought to be caused by supernatural forces such as demons or disgruntled divine beings [8].

However, the first important reflections on innate mental faculties were produced by the civilization of Ancient Greece. During the fifth century BCE, the Greek physician Hippocrates challenged the long-held belief that ID/IDD and mental illnesses were caused by supernatural forces and instead proposed that they stemmed from natural occurrences in the brain. A few decades later the philosopher Plato compared intelligence (nous) to a block of wax, which can vary in size, hardness, humidity, and purity. Intellectual defects would have been caused by alterations in this block of wax, which could be too small, too hard, too soft, too wet, or too impure. He described memories as imprints in this wax block and associated memory impairment with what is currently called ID/IDD. In a famous passage in the Phaedrus, Plato compares the intellect to a charioteer who holds the reins, guides, and directs, while emotion and will are compared to the horses that supply the motivation power to the chariot. In his masterpiece, the Republic, Plato seems to approve the alleged Spartan practice of depositing defective newborns at the foot of Mount Taygetos. However, the text is ambiguous, and it is not clear how the Spartan practice could refer to anything except visible physical malformations. In fact, Plato's conception of intellectual functioning does not seem to be compatible with such a cruel method. He distinguishes three types of impairment: simple-mindedness, civic ignorance, and ultimate ignorance. Simple-mindedness is used for those who judge things by appearances and sense perception rather than by appreciating their "ideal" forms. Civic ignorance is more severe and refers to lack of mental skills that one needs to function as a citizen, including good conduct toward the state, one's family, and oneself. Ultimate ignorance represents the greatest threat to the city-state or *polis* and can be further differentiated in lack of knowledge (agnoia) and abuse of personal power over others (amathia).

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Aristotle interpreted what is now named ID as a natural predisposition to slavery, according to his wider attempt of characterizing human beings in economic and politic terms. Nevertheless, he considered "slaves by nature" (doulos phusei) to be "partners in their masters' lives," without explicit references to an inferiority criterion. Aristotle considerably extended the concept of intellect learned at the Plato's academy by identifying two indivisible components (hylomorphism), a passive component, similar to matter, and an active one, similar to form. Intelligence is associated with logical and knowledge skills, which represent the main aspects of the Aristotelian interpretation of the psyche not defined simply as acts of receiving "contents" (acquiring information) but as points of contact (principles) with the outer world. He contrasted the cognitive or intellectual capacities with the "orectic" ones and simplified Plato's threefold classification by grouping together emotion and will.

Athens continued to be the Western world's main center of knowledge until the second century CE, when Rome and Alexandria started to host a variety of new philosophical and medical schools. At the end of the first century BC, the Roman orator, politician, and scholar Cicero merged the Platonic and Aristotelian concepts of intellect and cognitive functioning into the one of "intelligentia," with the main reference to acquired knowledge rather than natural disposition or innate ability. For these latter aspects, he used the term "ingenium," better explained as the complex of capacities and virtues through which humans "surpass what lies in their sensory awareness" and discern the world around them. For Cicero, the activity of ingenium "consists of catching sight of relationships and likenesses among things" to create meanings that meet our needs in the natural world [9]. Around one century later Avicenna, or Abu Ali al-Hussein ibn ʿAbdallah ibn Sīnā, physician, scientist, statesman, and one of the greatest Islamic philosophers, claimed that each human soul is a simple, independent combination of substance and form, created by God, and immortal by nature. In the second century CE, the physician, surgeon, and philosopher Claudius Galenus, often Anglicized as Galen, criticized the Stoic assumptions on the composition and localization of the psyche and, following Plato's idea, theorized mental functions to be organic facets of the brain, and intellectual alterations not to be ultimately fixed or determined in any individual. The many versions of his masterpiece "The Art of Medicine," also known as the Articella, had an enormous influence on the world medicine for centuries, together with Avicenna's Canon, and reached a cultured readership well beyond the medical realm.

Thus, the idea of intelligence as a measure for sizing up persons does not seem to belong to the classic culture, although the long pursuit of the old civilizations has included negative attitudes toward physical disability as part of mental illness. The seeds of intelligence as a measure for classifying humans were sown at the end of the twelfth century, with the expansion of towns and trade and the consequent onset of state and ecclesiastical administration. In the Middle Ages, intelligence was conceived as an entity hanging between heaven and earth, and divine and human. In the thirteenth century, Thomas Aquinas interprets human intelligence as a defective derivation of the divine intelligence and attributes the comprehension skill of less intelligent persons as a greater lack of completeness and universality than that of the general population. It is the medieval scholasticism that used the term intellect to translate the Greek word "nous" (understanding), as opposed to reason ("dianoia"). The concept of human *nous* somehow stems from its cosmic reference, which is not just a recipient of order, but a creator of it. Similarly, the brain is matter "configured" (shaped, structured) by the soul, which itself is a "configured" thing, but also a "configurer" for the brain.

According to Galen, the brain is instrumental in transforming the "vital spirit" (one of the three-part system of spirits derived from the Arabic philosophy) into the "animal spirit," which controls both peripheral and central neural activities, from sensation and movement, to imagination, cognition, and memory. In his work "De animalibus,"

Albertus Magnus, described by his contemporaries as one of the greatest philosophers ever and named by Pope Pius XII patron saint of scientists, described "idiotae" as people who "do not discern the universal from particulars" and who are not able to make abstractions. During the Middle Ages, the distinction between mind and body was more complex and more fluid than in post-Cartesian thought, complicated by different views on the soul, the relationship between thought and affect, and the position of mental functions in the body.

During the Renaissance, further development of urbanization and commerce was associated with increasing demands for quality and speed of performance, as traceable from standard medical textbooks, especially the many commentaries on The Art of Medicine. Most humanist writers extended Cicero's concept of "ingenium" in a normative way and used it as a reference to differentiate persons in terms of cleverness and speediness, while some other exponents of the humanistic medicine pioneered a new descriptive meaning and defined ingenium as an operation of the intellect. In his reinterpretation of the Articella, Niccolò da Lonigo (called Leoniceno), the leader of the humanistic movement which overcame the medieval setting, used ingenium for "quickness of apprehension," with a specific reference to the everyday cleverness, as opposite to the scholastics' meaning. Paracelsus and Felix Platter, and other eminent Renaissance physicians, introduced the concept of "feebleminded[ness]," as a condition to be differentiated from "foolish[ness]" and mental illness [10, 11]. Platter included "weakness" (imbecillitas), slowness of wit (ingenium) in his four headings of cognitive alterations, together with "consternation" (absence), "defatigation" (about sleep), and "alienation." He is also considered as a pioneer of psychiatry of ID/IDD as he was the first to offer a multi-level description of ID and to consider associated behavioral epiphenomena in terms of possible co-occurrent "syndromic" symptoms or sets of related symptoms. Other Galenist medical writers of the Renaissance described mental ill-health as the result of excesses or deficiencies of invisible material elements, such as "humors" and "animal spirits" (also called "soul spirits"). They associated the lack of one or more of these matters with different mental alterations having in common only some kind of slowness, such as lethargy, melancholy, and stupidity.

In the seventeenth century, speediness continued to have a central place in the evaluation of mental skills, and mental disability was increasingly considered as the opposite of mental geniality. The success of Descartes' theories strengthened this contraposition and attributed to slowness (and disability) an extremely negative meaning. In Descartes' thought, the mind, the source of mental functions, is no longer made of invisible matter (humors or soul spirits), but of a nonphysical and non-spatial substance. Cognitive speed and other intellectual functions are independent of the body because intellection is an operation of the mind alone. Ingenium takes on such importance to become the marker of the human species and lose any element of overlap with the medieval concept of "divine intelligences."

In 1664, Willis, an Oxford philosopher and pioneer of brain anatomy, described "stupiditas" and "idiocy" as disease-like states and proposed various etiological factors, including heredity, trauma, other diseases, and "animal spirits" [10, 11]. In theology, the term idiota continued to be used until the eighteenth century, with the meaning of someone lacking religious wisdom, or uneducated (indoctus). In 1690, the English philosopher John Locke [12], regarded as one of the founders of the Enlightenment in the eighteenth century, postulated that the mind at birth is a blank slate, with thoughts, beliefs, and personality being not innate, but learned through senses and experience. This revolutionary theory implied that person with learning disability could benefit from corrective experiences, and set the stage for the modern conception of rehabilitation. Locke also extended Albertus Magnus' point on "idiocy" as a lack of abstract skill and differentiates mental disabilities from physical ones, although his idea of mental disability overlapped with that of madness, especially in reference to intellectual and affective

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deficits [13]. Speed of cognitive performance and learning lost most of the previous significance in Locke's psychology and his eighteenth-century readers. In "The Improvement of the Mind," the theologian, logician, and educator, Isaac Watts states "presume not too much upon a bright genius, a ready wit, and good parts; for this, without labour and study, will never make a man of knowledge and wisdom … When they have lost their vivacity …, they become stupid and sottish" [14]. The British philosopher David Hartley proposed to classify individual intellectual performance by strength, vividness, intensity, and educability, but not speed. He interpreted ideas as the result of physical "vibrations," basing on principles of Newton's Optics [15]. In the mid-eighteenth century, the term "changeling" was used routinely for persons with ID. It was introduced by John Locke as an alternative for "idiot," to describe a creature which is human only in the body but without mind or soul.

Although speed of learning and other qualities of cognitive performance lost importance in the eighteenth-century theories of the mind, they gained a central position in the definition of the social status and social hierarchy. With the industrial revolution, more and more people fled to cities, working for slave wages and living in squalid conditions. Persons with ID were increasingly marginalized and left homeless and jobless. Parish authorities often bargained with factory owners to take one "imbecile" with every 20 workers. Together with the philosophers Rousseau and Hobbes, Locke argued against the idea that some individuals could prevail on or even subjugate other persons on the basis of their innate superior (sometimes divine) mental characteristics. However, it was the critical philosophy of Immanuel Kant [16–19] with its emphasis on autonomy, which laid the foundations for a truly persistent sociocultural revolution: "Every person is worthy of dignity not because of status or innate properties, but simply because of their human nature."

Kant argued that mental functions, including cognition, understanding, judgment, and reason, are immaterial active processes that convert raw sensory data into meaningful, ordered experiences through the instrumentality of innate abilities (*categories*). Things in themselves cannot be known; human beings perceive the world only the way their minds represent them. For the same reason and for their lack of substance, "reason," "cognition," and other mental operations are difficult to assess empirically as are deficiencies associated with these mental operations as additions to mental disorder [20].

The scientific attention given to ID/IDD began to increase at the end of the eighteenth century, after the French and American revolutions had restored value to human freedom and dignity and declared principles of equality, natural and imprescriptible rights. In 1793, Philippe Pinel, the leading French psychiatrist of his time, was the first to affirm that most persons labeled as "mentally deranged" were affected by a disease rather than being sinful or immoral by nature. Along with the English reformer William Turk, he created the method of "moral management," using gentle treatment and patience rather than physical abuse, chains, and other restrictions on the inmates of the Bicetre asylum, and later on those of the Salpetriere.

Jean Marc Gaspard Itard spent 5 years (1801–1806) on the education and rehabilitation of Victor, the wild boy from Aveyron [21]. Although the results of rehabilitation were only partial, his studies and methods were recognized by the French Academy of Sciences as notably worthy and opened the avenue for practice and research that spread widely to other countries in Europe and North America. On the thrust of Itard's work, Edouard Séguin, already sensitized to the ideas of Henri de Saint-Simon, deepened some techniques of qualification and started the study of the causes of ID, then called "idiocy." He founded the first private school in Paris dedicated to this area and in 1846 published the first systematic textbook. One year earlier, Jean-Étienne Dominique Esquirol, in his treatise on mental illness, had distinguished idiocy from imbecility, characterizing the former with a serious impairment of intellectual and moral abilities, the latter with a less marked limitation, in which knowledge and social relations did not reach an appropriate level for their age and education received [8, 22]. Seguin included this distinction in his textbook and

added two other categories which he termed mental weakness and superficial delay [8]. Further distinctions, both clinical and etiological, were made in the following years by Wilhelm Griesinger, John L.H. Down, and Desire-Magloire Bourneville, with references to cretinism, ethnic differences, and tuberous sclerosis [8, 23]. In 1891 at the Sorbonne University, Paris, Binet, and Simon developed the first intelligence test, specifically aimed at measuring children's abilities during different stages of development.

In the United States during the colonial and early republican periods, inhabitants of newly emerging small towns did not expel or denigrate persons who were slow to learn or to mature until 1840, when the concept of productive citizenship was claimed and idiots became a social and state problem. Before the civil war, care and control of persons with ID had already assumed a curious linkage [24]. It was Samuel Gridley Howe who challenged the widespread belief that "idiots" could not be taught and who established an experimental boarding school in South Boston for "Idiotic and Feeble-Minded Youth." Like Seguin, Howe firmly believed in the importance of community inclusion and wanted his schools to prepare children with ID/IDD to live within the wider society.

Intelligence and intellectual disability (ID) or intellectual developmental disorder (IDD), according to the most recent terminology, might not be natural kinds but the result of a historically contingent human attempt of self-definition and self-characterization. The seeds of intelligence as a measure for classifying humans were sown at the end of the twelfth century, with the expansion of towns and trades while a scientific attention toward ID/IDD started at the end of the eighteenth century, after the French and American revolutions had restored value to human freedom and dignity and declared principles of equality, natural, and imprescriptible rights. The first corrective interventions and the stage for the modern conception of rehabilitation were introduced during the Enlightenment period.

1.3 Definition and Terminology

The definition and the terms used for ID/IDD have always had relevant implications for research and clinical practice as well as for sociocultural attitudes and health policies. Definitions of ID/IDD have been provided across history in accordance with changing conceptualizations, but a scientific discussion on description and terminology started together with the advent of the international classification systems of mental disorders. The use of terms such as idiocy, imbecility, cretinism, moronity, feeblemindedness, mental subnormality, defectiveness, and mental deficiency has been maintained in these classification systems until the end of the mid-twentieth century [7, 8, 22, 23, 25, 26].

In 1904, Martin Barr published the first textbook on ID/IDD using the term "Mental Defectives" in the title. He differentiated between an idiot and imbecile by attributing to the former a considerably higher severity of impairment ("see nothing, feels nothing, hears nothing, and knows nothing") (p. 18) and summarizing different causes and prognoses. Idiots were considered as affected by an untreatable disease and sent to assistance only, while imbeciles were considered to be mentally defective and were trained to learn daily living skills through step-by-step, easy, and concrete instructions [25]. In 1908, Tredgold extended Barr's concepts and provided educational resources for professionals on appropriate diagnosis, treatment, eligibility for services, and long-term placement.

In 1952, the American Psychiatric Association [27] published the first Diagnostic and Statistical Manual (DSM-I) classifying this disorder as "chronic brain syndrome with mental deficiency" (p. 14) and "mental deficiency" (p. 23). In 1961, the American Association on Mental Retardation (AAMR), formerly American Association on Mental Deficiency (AAMD), introduced the term "mental retardation" to replace the previous terms, which became gravely stigmatized. This coincided with the introduction of the first definition and diagnostic criteria to be nearly universally adopted. The definition stated that the disorder "refers to subaverage general intellectual functioning

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which originates in the developmental period and is associated with impairment in adaptive behaviour" [28]. In 1973, the AAMR changed the term "subaverage" with "significantly subaverage" and the terms "impairment in adaptive functioning" with "concurrent with deficits in adaptive behavior." These changes were based on the need to increase the intelligence quotient (IQ) standard deviation from the population mean, which had been found to be overly inclusive and to determine excessive assignment of students to special education, and because the adaptive behavior criterion had been ignored in clinical practice. The 1983's definition included the IQ's standard measurement error and emphasized the importance of clinical judgment in the diagnostic process as well as the impact of social milieus in facilitating or impeding the development of intelligence.

A paradigm shift occurred in 1992, when the AAMR moved the emphasis of the definition from a disorder (neither mental nor medical) to a disability: "mental retardation refers to a particular state (not a trait, Ed.) of functioning that begins in childhood in which limitations in intelligence coexist with related limitations in adaptive skills" ([29], p. 9). The focus of the disability construct is defined by stating that the individual condition is expressed through an interaction between the affected person and the environment ([29], p. 9). The impairment of adaptive functioning was defined by the presence of significant disabilities in at least two out of ten specific adaptive skills, which replaced the previous deficits in adaptive behavior. The DSM maintained its definitions aligned with that of the AAMR in the third and fourth editions.

In the second half of the last decade of twentieth century, the terms "mental retardation" coexisted with "intellectual disability," which was considered to be more aligned with the new conceptualization of the developmental condition. ID/IDD was increasingly accepted and included in the name of some scientific journals and international scientific organizations, such as the International Association for the Scientific Study of Intellectual Disabilities, the Section of Psychiatry of Intellectual Disability of the World Psychiatric Association, and the European Association for Mental Health in Intellectual Disability.

In 2002, the AAMR updated its definition by specifically stating that mental retardation is a disability and aligning its position with that of the World Health Organization's (WHO) International Classification of Functioning (ICF). It emphasized that mental retardation implies significant limitations in the areas of intellectual functioning and adaptive behavior, with the two areas having equal footing and the latter being organized into the conceptual, social, and practical domains. In 2007, the AAMR changed its name to American Association of Intellectual and Developmental Disabilities (AAIDD) and in 2010 included the term ID in its updated manual, putting the construct of disability first and leaving the consideration of the developmental impairment as an invariant trait of a person. The primacy of the concept of disability implies the social–ecological context to have a main causative role for the difficulty of the person with developmental impairment to achieve a good adaptation to the environment as well as self-worth, well-being, and self-determination [6, 30]. In this model, the grading of ID severity is represented by "patterns and intensity of supports needed" (i.e., intermittent, limited, extensive, and pervasive), with a considerable difference from the DSM severity codes. In 2013, the AAIDD revised the fidelity of its approach to the ICF model by proposing a more complex functionality approach, which includes health issues and the need for disease classification codes [31].

In the fifth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-5) [1], the American Psychiatric Association (APA) replaced "mental retardation" with "intellectual disability" and the parenthetical name "intellectual developmental disorder," to harmonize with the first proposal of the WHO working group for ICD-11 and to underline that diagnosed deficits in cognitive and adaptive skills begin in the developmental period. The ICD-11 [2] has adopted the term "disorders of intellectual development (DID)."

Difficulties in defining and positioning the ID have been maintained up to the new sys-

tems of international classification of diseases and disorders [DSM-5 [1] and ICD-11 [2]] and in recent years have raised a more heated scientific debate than ever before.

Currently, there are two main approaches: the first, promoted by the AAIDD and aligned with the international classification of functioning (ICF; [32]), focusing on disability as characterized by significant impairment of intellectual functioning and adaptive behavior; the second, supported mainly by the Section of Psychiatry of Intellectual Disability of the World Psychiatric Association (WPA-SPID), is instead polynomial-polysemic, multidimensional approach which interprets ID as a condition to be defined in accordance with different contexts. With specific reference to the classificatory systems, it should be defined as a meta-syndromic group of "intellectual development disorders" in the ICD, and as a condition of complex "intellectual disability" in the ICF.

Extreme positions in this debate might have extremely negative implications. The conceptualization of ID uniquely as a "disability" would be followed by its exclusion from the ICD, and consequently by an enormous reduction of eligibility for specific healthcare, particularly mental healthcare (see ▶ Chap. 9 for the extent of mental health needs) as well as for educational and social services [33]. In fact, it is the ICD and not the ICF that is used by the 194 WHO member countries to define the responsibilities of governments to provide healthcare and other services to their citizens. Furthermore, the removal of ID from the ICD would have a major impact on both national and global health statistics as well as on health policy and service availability to this vulnerable population [34]. Interventions related to dietary intake (i.e., for disorders such as phenylketonuria), psychopharmacotherapy, or education are addressed for attenuation of overall deficits and related activity functioning and social participation. On the other hand, the consideration of ID solely as a health condition would have an extremely negative impact on the provision of social and educational services, as well as public attitude, policy formulation, and future legislation. Furthermore, it would be inconsistent with the current position of many governments, international organizations, and advocacy groups.

The choice of DSM-5 to adopt the term ID, but to accompany it (in parenthesis) by IDD also reflects the ongoing difficulties in definition and positioning. Moreover, the USA has not adopted ICF.

In 2013, the first WHO working group on mental retardation for the ICD-11 provided a valuable attempt to overcome these issues by proposing a polysemic-polynomial approach [6]. Specifically, it has been proposed that a distinction is needed between "intellectual developmental disorder" (a clinical meta-syndrome coded at ICD) and "ID" (its functioning/disability counterpart coded at ICF), since these terms have different scientific, social, and policy applications, although they describe two different but related aspects of the same construct (IDD/ID). Indeed, according to the WHO working group, these two terms should be regarded as "semantically similar" but not as "mirror codes" [35]. It has to be considered that in most countries, service eligibility and treatment selection for people with ID/IDD are heavily influenced by diagnostic classification, and a reliable, valid, and feasible diagnostic system seems needed to guarantee better health services for persons with ID. Conversely, in low- and middle-income (LAMI) countries, mental health professionals of any kind maybe lacking, and often not even physicians are available for people with ID/IDD. Therefore, the priorities of identifying and providing tools to enable a widespread, efficient, and accurate identification and prioritization of service needs for people with ID/IDD are among the aims of the ICD-11. Moreover, it is important that the ICD-11 provides the basis for documenting the prevalence of ID and its functional consequences, because many individuals remain undiagnosed for years especially in LAMI countries. For the development of healthcare programs, it is also important to consider the amount of resources available for diagnosis as opposed to treatment. However, it must be understood that any marked changes to the ICD may not be incorporated into clinical practice in many countries for some years;

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therefore, it is preferable to keep assessment as user-friendly as possible to encourage its use.

Different definitions of ID/IDD have been provided across history in accordance with several conceptualizations. Currently, there are two main approaches: the first prioritizes the aspects associated with the concept of "disability" such as intellectual functioning and adaptive behavior over those associated with the concept of "disorder"; the second advances a polynomial-polysemic, multidimensional approach that interprets ID/IDD as a condition to be defined according to the different classificatory contexts such as classification of diseases and classification of functioning.

1.4 Nosology

As mentioned earlier, in the DSM-I [27] ID/IDD was split into "chronic brain syndrome with mental deficiency" and "mental deficiency" (p. 23). The former disorder was defined by the presence of "a defect of intelligence existing since birth, without demonstrating an organic brain disease or known prenatal cause." This group included "only those cases formerly known as familial or 'idiopathic' mental deficiencies."

In 1961, with the introduction of the term mental retardation, the AAMR, formerly AAMD, proposed more objective diagnostic criteria, including tests and reference scores for the assessment of intelligence and adaptive skills. The main criterion was an IQ lower than 84 or one standard deviation below the average of the general population. The impairment of adaptive functioning was divided into four severity levels: mild, moderate, severe, and profound retardation [28].

The diagnostic criteria for mental retardation (MR) remained unchanged until 1965 when the WHO included it in the International Classification of Diseases, Eight Revision (ICD-8), with some considerable changes from the DSM [36].

The DSM Third Edition (DSM-III) in 1980 [37], and the DSM Third Edition-Revised (DSM-III-R) in 1987 [38], included MR under "developmental disorders," which were considered as personality issues and placed in Axis II.[1]

In 1992, the AAMR introduced a multidimensional classificatory approach aligned with the above-mentioned conceptual shift from a disorder-centered model to a disability-centered model, which included the four dimensions of (a) intellectual functioning and adaptive skills; (b) psychological/emotional considerations; (c) biomedical, social, behavioral, and education factors; and (d) environmental considerations. Furthermore, the 1992 AAMR diagnostic manual proposed to arise the IQ ceiling from 70 to 75 and to replace IQ with support need in the definition of diagnostic subcategories, which was not accepted by the wide majority of the American scientific community [39].

The DSM-IV [40] aligned itself with the AAMR in defining mental retardation, while it departed from the upper limits of the severity range, which were kept at lower levels, and in multiaxial rather than multidimensional classification. This Axis II positioning was maintained in DSM-IV and DSM-IV-TR [41], where MR was included under the Chapter "Disorders Usually First Diagnosed in Infancy, Childhood, or Adolescence."

The diagnostic criteria of the AAMR manual were revised in 1973 and 1983, the former comprised a reduction of the IQ cut-off to two standard deviations below the average of the general population and an extension of the age of onset to 18 years, and the latter consisted of an insertion of the IQ standard measurement error [42]. These criteria were basically embraced by the DSM-III (Diagnostic and Statistical Manual of mental disorders—III edition, [37]) and the ICD-10 (International Classification of

1 A complete DSM-III diagnosis enabled coding on five separate axes. All clinical psychiatric syndromes were coded on axis I, with the exception of specific developmental disorders and personality disorders, which were coded on axis II. Axis III was used for coding physical disorders or physical conditions, axis IV for coding severity level of psychosocial stressors, and axis V for the highest level of adaptive functioning in the past year.

Diseases—tenth edition, [43]), and used for the first time in almost the whole world.

In the ICD-10 ID/IDD appeared with the name "mental retardation" in the chapter V (Mental and Behavioural Disorders), with some disability aspects being regarded as psychiatric symptoms. Genetic syndromes including ID/IDD, such as Prader–Willi or Fragile X, were coded in the ICD independently from associated cognitive impairment.

In 2009, the WHO established a Working Group for the new classification of MR (WHO WG-MR) reporting to the WHO International Advisory Group for the Revision of ICD-10 Mental and Behavioral Disorders in the ICD-11. Strikingly, the WHO WG-MR was the first international and multidisciplinary expert group established by an international organization to debate the issue of the classification of ID/IDD and to produce an international consensus document incorporating views of representatives of the main professional, scientific, and self-advocacy organizations, including an APA delegate to harmonize with the ongoing DSM-5 Task Force. The WG-MR produced a position paper in 2011 [44] with an extensive consensus report, which represented the basis for the official recommendations sent to the WHO Advisory Group on ICD-11 in 2012.

The terminology, nosology, and the positioning of ID/IDD within the taxonomy of mental disorders have long been controversial; indeed, the debate has continued for more than two decades in parallel to the dispute on the concepts of intelligence, human functioning, and disability [45]. ID/IDD is regarded as a cluster of syndromes that typically emerge early in life and imply different patterns of severe and persistent cognitive impairments together with altered adaptive behavior and personal skills. Despite the high variability of clinical presentation, this cluster of syndromes has been associated with a complex array of neurodevelopmental underpinnings. Classification and diagnostic criteria are therefore crucial issues and have important implications for epidemiology and interventions as well as outcome measures, service provision, and planning. Moreover, the debate on ID/IDD has implications for the classification of other neurodevelopmental disorders such as autism spectrum disorder where a specifier "with or without accompanying intellectual impairment" is required.

As already explained for the naming, the question of whether ID/IDD should be considered a health condition, a disability, or even a life condition is still debated in many contexts. For example, different interpretations of the condition exist in different fields, such as psychology, sociology, anthropology, and education. These interpretations still influence the classification of ID/IDD and consequently the choice of diagnostic criteria.

The main issues which are still debated are the age-at-onset cut-off, the consideration of specific cognitive functions as alternative to overall IQ, and the relationship between cognitive impairments and adaptive skills.

In DSM-5, ID/IDD is included under the meta-category of neurodevelopmental disorders to make clear that the focus is on the disorder category and not the disability construct. There, the diagnostic criteria are (a) overall intellectual functioning significantly (two standard deviations) below the mean of the general population; (b) deficit in adaptive functioning (without continuous support), with particular reference to the sociocultural standards of personal independence and social responsibility; and (c) onset of intellectual and adaptive deficits during the development period, typically before school education. Deficits in adaptive functioning have to refer to at least one of the daily activities of life, such as communication, social participation, or autonomy, in different contexts, such as home, school, work, and community [1].

General intellectual functioning is defined by the IQ (IQ or IQ equivalents) obtained through evaluation with one or more standardized, individually administered intelligence tests (e.g., the Wechsler Intelligence Scale, the Stanford Binet, the Evaluation Battery of Kaufman or, in people with communication problems, the Leiter scale). An intellectual functioning significantly below the general population mean is defined by an IQ of about 70 or less.

Since before the publication of the DSM-5 [1], the severity of ID (then "mental retarda-

tion") was defined almost exclusively on the basis of IQ: mild for values between 50–55 and 70 (50–69 in the ICD-10), moderate between 35–40 and 50–55 ("medium," 35–49 in the ICD-10), severe between 20–25 and 35–40 (20–34 in the ICD-10), profound for values below 25–20 (<20 in the ICD-10). In the DSM-5, the severity levels are defined on the basis of adaptive functioning and not on the IQ, especially for the forms that fall in the lower part of the range of deficits, with specific reference to conceptualization, social and practical domains.

In the ICD-11, the primary parent category for ID/IDD is that of "Neurodevelopmental Disorders." In the last decade, the neurodevelopmental focus has represented one of the aspects upon which the whole international scientific community has agreed most. IDD encompasses a broad grouping of heterogeneous developmental conditions which result from significant interference with the growth and maturation of the brain, especially during the prenatal and perinatal periods. The disorders included in this meta-structure are not mainly organized by the criteria of onset in childhood and adolescence, as in ICD-10 or DSM-IV, but by etiopathogenetic factors, such as genetic alteration or abnormal neural circuit development, and clinical aspects, such as dysfunctions in cognition, learning, communication, behavior, early emergence, co-occurrence, and lifespan course.

The ICD-11 has renamed ID/IDD with "Disorders of intellectual Development" with reference to "a group of etiologically diverse conditions originating during the developmental period characterized by significantly below average intellectual functioning and adaptive behavior that are approximately two or more standard deviations below the general population mean (approximately less than 2.5 standard deviation), based on appropriately normed, individually administered standardized tests. Where appropriately normed and standardized tests are not available, diagnosis of disorders of intellectual development requires greater reliance on clinical judgment based on appropriate assessment of comparable behavioural indicators" [2, 33].

The nosographic approach to ID/IDD has considerably changed across time, passing from a unitary and scarcely defined diagnosis of "mental deficiency" to a group of lifespan conditions characterized by early onset significantly below average intellectual functioning and adaptive behavior.

1.4.1 Diagnostic Criteria

1.4.1.1 Early Cognitive Impairment

Intelligence, according to the ICD, is considered as an umbrella term that is age-appropriate and includes cognitive functioning, learning, adaptive behavior, and skills, meeting the standards of culture-appropriate demands of daily life.

ID/IDD can be envisioned as an early cognitive "meta-syndrome" analogous to the syndrome of dementia in later life [6]. The two meta-syndromes share similar characteristics: a variety of causes can lead to the two above-mentioned conditions, they can involve extensive impairment of core cognitive functions required for daily living, and the clinical evaluation process involves a broad consideration of biological, personal, and environmental factors.

Particularly, ID/IDD is a condition that involves the entire lifespan. ID/IDD is characterized by a marked impairment of core cognitive functions necessary for the development of knowledge, reasoning, and symbolic representation and that deserves the consideration of the developmental phases and life transitions of the person. Therefore, a developmental approach that accounts for the complex causal factors known to impact the acquisition of cognitive abilities and adaptive behaviors is necessary to understand ID/IDD and to provide the appropriate interventions and long-term care [46].

To note, there is still no conceptual map or hierarchy of the key cognitive functions and domains to be considered in ID/IDD [47], and different terms are often used for the same functions, and vice versa. Nevertheless, a series of cognitive domains are significantly impaired in persons with ID/IDD. These core cognitive domains mainly include percep-

tual reasoning, working memory, processing speed, and verbal comprehension [48, 49].

However, it has to be considered that different combinations of cognitive impairments and atypical features can manifest in ID/IDD [50]. For example, persons with Down syndrome usually manifest impairments in specific areas of language, long-term memory, and motor performance, while showing relative strengths in visuospatial construction [51]. In contrast, persons with Williams Syndrome show deficits in attention, visuospatial construction, short-term memory, and planning [52], while showing a distinctive pattern of auditory processing and relative strengths in auditory processing and concrete language [53].

The assessment of cognitive functions in ID/IDD is generally performed using an IQ test, a score derived from an intelligence test which should be used for diagnosis only with consideration of locally standardized norms. Usually, an IQ score of 70 or below is indicative but not sufficient to diagnose ID/IDD. Other alternative clinical and cognitive assessments could be used including locally standardized developmental tests. Indeed, cognitive assessment should not be limited to standardized IQ but also include neuropsychological testing, for example, assessment of executive functioning in order to determine an individual's profile. Moreover, retesting during critical life periods is recommended, as distinct cognitive developmental trajectories have been identified in different conditions of ID/IDD and because there may be cognitive losses related to aging. Besides the identification of cognitive impairments, the diagnosis of ID/IDD should also be associated with the assessment of difficulties in different domains of learning, including academic, social, and practical knowledge. For example, objectives of the elementary school curriculum are expected to be difficult to meet for persons with ID/IDD. Reading, writing, and calculating are hard tasks to achieve and will need considerable effort and repetition. These difficulties can also vary across levels of cognitive functioning and other aspects of the disability and of the environment, as well as across different etiological conditions. Indeed, persons with Down syndrome show more problems in motor learning compared with other IDD syndromes [49].

1.4.1.2 Limitations in Adaptive Behavior

The person diagnosed with ID/IDD also manifests limitations in the context of adaptive behavior that is, meeting the demands of daily life expected of same-age peers, as well as demands of cultural and community environment. These difficulties include limitations in relevant skills for daily life, previously categorized into conceptual, social, and practical skills [54]. According to ICD-11, "conceptual skills are those that involve the application of knowledge (e.g., reading, writing, calculating, solving problems, and making decisions) and communication; social skills include managing interpersonal interactions and relationships, social responsibility, following rules and obeying laws, as well as avoiding victimization; and practical skills are involved in areas such as self-care, health and safety, occupational skills, recreation, use of money, mobility and transportation, as well as use of home appliances and technological devices" [2].

Becoming independent and self-directed or having adequate self-care in the context of everyday life will require practice and long-term monitoring, coaching, and support. Practical examples can be represented by dressing skills and how to appropriately interact with others. Moreover, persons with ID/IDD often have difficulties in managing their behavior, emotions, and interpersonal relationships, and maintaining motivation in the learning process.

1.4.1.3 Severity/Extent (Subtypes)

Though the degree of cognitive deficits and adaptive skills vary from person to person, in most instances it is possible to categorize ID/IDD into one of the four following clinical severity levels: mild, moderate, severe, and profound. The prevalence is higher for mild, followed by moderate subtypes, while severe and profound subtypes show similar lower rates [55, 56].

In the ICD-11, the severity of an ID/IDD is assigned "on the basis of the level at which

the majority of the individual's intellectual ability and adaptive behaviour skills across all three domains (i.e., conceptual, social, and practical skills) fall. For example, if intellectual functioning and two of three adaptive behaviour domains are determined to be 3 to 4 standard deviations below the mean, Moderate Disorder would be the most appropriate diagnosis. However, this formulation may vary according to the nature and purpose of the assessment as well as the importance of the behaviour in question in relation to the individual's overall functioning" [2].

1.4.1.4 Age of Onset and Course

ID/IDD is an early-onset syndrome generally manifesting in infancy, childhood, and extending into adolescence. No specific temporal qualifier is necessary for the diagnosis [57]. The ICD-11 states that "among adults… who come to clinical attention without a previous diagnosis, it is possible to establish developmental onset through the person's history, i.e., retrospective diagnosis" [2].

Although ID/IDD is usually a stable diagnosis, it is considered a dynamic health condition, since there can be significant variability in cognition and functioning across different clinical severity levels throughout the lifecycle [58] and may show high individual and condition-related variation in developmental trajectories (e.g., the development of sensory and cognitive functioning in adults with Down syndrome). Therefore, it is highly recommended to perform reassessment of the individual at key developmental phases, life transitions (e.g., at school entrance, puberty, transition from educational to occupational setting, with changes in living arrangements, early and later adulthood), and after significant life events and/or traumatic events. The experience of life events and trauma can be particularly challenging for a person with ID/IDD who has limitations in adaptive functioning. While the timing and type of life transitions vary across societies, generally individuals with ID/IDD need additional support to adapt to changes in routine, structure, educational, or living arrangements. Traumatic life events (e.g., learning of the sudden death of a family member, being in a life-threatening accident, being physically or sexually assaulted) are important predictors of psychopathology in this population [57].

ID/IDD is influenced by many factors including the maturation of the brain, gene–environment interactions, environment, culture, education, availability of supports and training opportunities as well as by the physical and mental health of the person [59]. To note, most individuals with ID/IDD continue to acquire skills and competencies, especially with optimal care, training, supports, education, and opportunities for learning. A summary of the most important ID/IDD features and vulnerability factors across different age periods are provided in ◘ Table 1.1.

ID/IDD refers to a group of etiologically diverse conditions originating during the developmental period characterized by deficits in intellectual functions and adaptive behavior. These characteristics can vary significantly across individuals at different clinical severity levels throughout the lifecycle. Therefore, it is highly recommended to perform reassessments of individuals with this condition at key developmental phases, life transitions, and after significant life and/or traumatic events.

1.5 Clinical and Functional Properties

The alignment of ICD-11 with ICF enables professionals to explore and compare the conceptual, clinical, and functional properties of IDD and ID and to explore differences across age groups, life situations, and settings. As described earlier, "intellectual developmental disorder" (IDD, clinical meta-syndrome coded in ICD) and "intellectual disability" (ID, its functioning/disability counterpart coded in ICF) describe differently, but related aspects of the same construct. Therefore, it can be assumed that the disorder category (IDD) and the concept of intellectual disability (ID) may not necessarily always represent

Table 1.1 Main IDD features and vulnerability factors across the lifespan

Adolescence	Puberty and adolescence may cause increased social, emotional, and behavioral difficulties for individuals with ID/IDD. During this stage, major transitions take place in physical growth and sexual maturation. There may be also additional changes in educational and/or care settings
Young adulthood	During young adulthood, individuals with mild or moderate ID/IDD often have difficulties with personal relationships, managing finances, and planning. In most societies, there are also other significant life transitions for the person with ID/IDD (e.g., moving to adult services, moving from living at home to a group home, moving from an educational to an occupational setting, or getting married), and this can put an individual with ID/IDD at increased risk for problem behaviors and psychiatric illness. Indeed, psychiatric illnesses may have their onset in young adulthood. It is important to note that persons diagnosed with mild ID/IDD in childhood may no longer meet criteria for ID/IDD in later life, as their skills may be better suited to the new environmental demands or because of new brain maturation and learning. Even in this optimal group, individuals with a BIF diagnosis will require specific health attention and support in areas such as education, work, and the justice system. Unlike most people without ID, people with ID/IDD often do not move out of parental home at this age, which has implication for their family caregivers
Adulthood	Many older adults with mild ID/IDD may continue to work and live independently. There may be known condition-specific health risks that should be checked for in mid-adulthood [60]. For example, older adults with Down syndrome have a higher prevalence of Alzheimer's disease and epilepsy. Family life transitions include bereavement of older family members and parents
Old age	In addition to the onset of condition-specific health problems, age-related health conditions occur in old age [61]. Dementia occurs at a higher rate in adults with ID/IDD compared with the non-ID general population [62]. In general, aging is accelerated in many genetic conditions associated with ID/IDD, for example, in Down syndrome. Early retirement is available for persons with ID/IDD in countries with extensive social inclusion programs

the exact same functional characteristics. A minimal set of ICF codes to record manifestation and other functional properties of IDD is available within the ICD-11.

The ICD-11 Content Model [63] lists "functioning properties" as one parameter to capture knowledge that underpins the definition of an ICD entity. According to the ICD-11 reference guide, "signs and symptoms in the ICD are aligned with body functions in the ICF" (see 3.3 Functioning in ICD and joint use with ICF, [64]). For IDD, assumingly "signs and symptoms" (manifestation properties) would refer to the ICF Category "b117 Intellectual functions." Consequently, the "supplementary section for functioning assessment" of the ICD-11 does not include "intellectual functioning" in any of the three subsets: (1) ICF items represented in the WHODAS 2.0 36 Item version, (2) ICF items proposed for a "Brief Model Disability Survey (MDS)," and (3) a set of ICF items considered "Generic functioning domains" (Body functions, activities, and participation domains, excluding codes already included in (1) or (2)).

The "Clinical Descriptions and Diagnostic Guidelines (CDDG)" for ICD-11 (ICD-11 CDDG, developed in close coordination with DSM-5 diagnostic criteria; [65]) define essential features of IDD as problems with "intellectual functioning" and with "adaptive behavior" (DSM-5: adaptive functioning). The CDDG for ICD-11 go beyond the manifestation property (IQ as indicator of intellectual functioning) by considering how people function in daily life [66]. The "adaptive behavioral indicators" and "intellectual functioning behavioral indicators" are grouped into three domains ("Communication skills," "Social skills," and "Daily living skills") with separate

indicators for "early childhood," "childhood and adolescence," and "adulthood" [67]. These indicators can be used to group individuals on the basis of one of four severity levels (i.e., "mild," "moderate," "severe," and "profound") across all domains. The rationales for two separate sets of functional indicators (intellectual functioning and adaptive functioning) are low correlations between the two, especially in high functioning ID, and, therefore, the need to use both to identify ID [68, 69]. A more precise mapping of adaptive functioning is important to differentiate between a range of neuropsychiatric syndromes [45], but also to understand changes in functioning across the lifespan and as a result of intervention and service provision. Independent of manifestation and functional properties of IDD, ICF categories may be selected to describe additional functional problems of specific individuals or groups or to identify areas of strengths. The assignment of ICF categories largely depends on the setting and the assessment or documentation purposes. Environmental and personal factors might be considered as potential contributors to the overall life experience and to identify intervention strategies to improve service provision.

To document functional characteristics with ICF codes and explore the dynamics of functioning and disability requires compatible assessment instruments to collect valid and reliable data. The use of the ICF as an information system is guided by data generation (i.e., assessing disability) and knowledge creation (i.e., theorizing disability) processes. The ICF is able to represent assessment data from diverse sources (e.g., tests, interviews, observation) in a coherent information system, but also helps to clarify constructs such as "intelligence" or "disability" by mapping definitions or models into the ICF. In other words, assessment information is aggregated into the ICF, and functional information represented in the ICF is used to describe the problem of an individual or group (data-information-knowledge hierarchy) [70]. For example, the CDDG for ICD-11 include functioning properties based on the "Vineland Adaptive Behaviour Scale" [71] to define the concept of "adaptive behavior" [67]. The choice of assessment tools, therefore, does not only predetermine the information, but also implicitly guides knowledge creation processes (clinical sense-making, decision-making, intervention, or service planning). The definition of functional properties, therefore, depends on conceptual models, on the purpose of identifying functional properties and on the choice of assessment tools. "Intelligence" is assessed with standardized psychometric tests (e.g., WISC-IV, WAIS-IV, K-ABC, etc.), but "cognition" or "cognitive functions" may be more relevant constructs to understand specific functional problems associated with learning and problem-solving [45]. "Cognition" as assessed by the WHODAS 2.0 includes interview questions for "attention function," "memory functions," "solving problems," "basic learning," "communicating with—receiving—spoken messages," and "conversation." In conclusion, the functional properties of IDD or ID largely depend on the choice of underlying concepts, choice of assessment methods, and the purpose of assessment.

Lastly, it should not be forgotten that mapping functioning and disability is also about understanding people's life experiences, not only about defining IDD or exploring functional problems associated with ID. The subjective "lived experience" includes concepts such as well-being that are beyond the ICF, but conceptually compatible with it [32]. Psycho-educational, sociocultural, and justice perspectives complement the biomedical perspective to approach IDD [72]. For example, article 24 of the Convention on the Rights of Persons with Disabilities [73] demands that states create educational opportunities to support the development of personality, talents, and creativity of persons with disabilities. The human rights perspective emphasizes issues of personal freedom, availability of choices, and subjective well-being. What gives meaning to the lives of people? Which strengths, skills, aspirations, and talents lie dormant and are not yet mobilized? What is a person's potential to develop functioning not yet developed? Static assessment methods used to document current functioning are inadequate to generate information about a person's potential to develop functioning. Dynamic assessment

methods may be more appropriate to fully explore communication abilities [74] or search for potentials [75]. Dynamic assessment includes experimental approaches to expand abilities [76] and to reflect on the interactive nature of disability as conceptualized by the ICF. The "locus of disability" may be more adequately described as a gap or discrepancy between personal competence and environmental demands [72] than as "functional properties" of the individual. For example, problem behaviors such as aggression or self-injurious behavior associated with IDD and ID [33] may be indicators of such a gap or mismatch.

Defining functional properties of IDD is an ongoing process guided by further development of underlying concepts and the choice of assessment tools and strategies. Identification of individuals and groups also depends not only on the selection of adequate functioning components, but also on the choice of indicators and cut-off or threshold criteria. The perspective assumed by professionals will have an impact on disaggregating complex life situations into functioning and disability components, environmental, and personal factors. If an interactive approach to understanding disability is adopted, the functioning of social systems may be more relevant than the functioning of an individual.

Signs and symptoms of ID/IDD as described in ICD-11 are aligned with body functions in the ICF. Further definition of individual functioning can be obtained through evaluation of life experiences and well-being as well as other psycho-educational, sociocultural, and justice aspects.

1.6 Adaptive Behavior and Skill

Adaptive behaviors are the everyday skills we learn and perform to meet the expectations and societal demands [77]. These expectations and demands increase in complexity as the individual's chronological age increases. Hence, we expect different adaptive behaviors from a child, than from an adolescent or an adult. Adaptive behavior consists of a broad set of skills, encompassing conceptual skills (e.g., functional academics, money and time concepts, communication, self-direction), social skills (e.g., interpersonal skills, social problem-solving, following social rules and laws), and practical skills (e.g., personal hygiene, home living skills, work skills). Empirical support—using a sample composed of both people with and without intellectual disability—supported this three-factor structure of adaptive behavior [78]. Although related to some extent to intellectual functioning, adaptive behavior remains a separate and independent construct [79–82]. A person's inability to learn and/or be able to independently perform these skills may be an indicator of the presence of a neurodevelopmental disorder. The condition that is diagnosed when there is manifestation of significant deficits in adaptive behavior, along with significant deficits in intellectual functioning occurring during the developmental period, is intellectual disability [1, 77] or disorders of intellectual development. Overall, intellectual disability and disorders of intellectual development refer to the same condition or disorder and include the same individuals.

Deficits in adaptive behavior may be attributable to any number of factors, including (a) the person's incapacity to learn and/or perform some skills despite sufficient and appropriate opportunities; (b) a lack of exposure or opportunities to learn and perform some skills; (c) motivational deficits that results in the person choosing not to learn and/or perform some skills; and/or (d) the presence of a comorbid disorder that interferes with the person's learning and/or performing some skills.

1.6.1 Assessment

According to Coulter and Morrow [83], the assessment of adaptive behavior has historically been done for two primary purposes: establish a diagnosis/eligibility determination (i.e., does the person present with significant deficits in adaptive behavior) and identify intervention areas (i.e., individual education plan, individual support plan, identify strengths, and weaknesses). Doll [84, 85] is

seen as the author of the first standardized measure of adaptive behavior, called the Vineland Social Maturity Scale. Since the publication of the Vineland Social Maturity Scale, more than 200 measures of adaptive behavior have been inventoried at different points in time [86, 87]. A vast majority of these measures are used for the assessment of discrete adaptive skills for the purpose of measuring skills deficits with the goal of planning didactic training or implementing or evaluating skills acquisition program. These instruments might be a brief inventory, checklist, or questionnaire focused on a specific area or ability (e.g., social skills, communication skills, toilet training, work skills). These assessments typically involve obtaining information from a third-party respondent (e.g., parent, teacher, direct support professional), but a limited number of assessments involve a direct measure of the person's knowledge of adaptive skills (e.g., Independent Living Scales [88], Street Survival Skills Questionnaire [89]), while others may be administered as a semi-structured interview with the assessed person who provides a self-report of their skills (e.g., Adaptive Behavior Assessment System [90]).

Generally speaking, for the purpose of conducting a robust assessment of adaptive behavior aimed at making a diagnosis of intellectual disability, a number of elements are strongly recommended. The assessment should include, when possible, information obtained from a comprehensive and psychometrically robust standardized assessment tool for adaptive behavior, informed by direct observations from multiple respondents [77, 91]. There are less than a handful of standardized scales that are sufficiently robust for use when the purpose of the assessment is for making a diagnosis or determination of intellectual disability [67, 91]. These recommended instruments include Adaptive Behavior Assessment System—Third Edition (ABAS-3; [92]), Adaptive Behavior Diagnostic Scale (ABDS; [93]), Diagnostic Adaptive Behavior Scale (DABS; [94]), Scales of Independent Behavior, Revised (SIB-R; [95]), and Vineland Adaptive Behavior Scale—Third Edition (Vineland-3; [71]). All these standardized assessment tools are sufficiently psychometrically robust and comprehensive to provide a good standardized measure of a person's adaptive behavior for the purpose of determining ID/IDD.

Alternative sources of adaptive behavior information can be obtained from clinical interviews with people who have lived with or have recently observed, on a regular basis, the person function in their home, school, work, and/or community. Other records that might provide helpful corroborating adaptive behavior information include social and family history, medical records, school records, educational and psychological evaluations, work records, etc. Tassé and his colleagues [67] proposed a number of behavioral indicators of adaptive behavior to guide clinicians in assessing adaptive behavior when standardized adaptive scales are not available.

1.6.2 Intervention

Teaching and promoting the acquisition of adaptive behaviors should be an essential goal of any intervention. The outcome of increasing a person's repertoire of adaptive behavior will often lead to greater independence, personal autonomy, and self-direction as well as a reduced perception among laypeople that the person is disabled. The DSM-5 [1] moved away from using the level of intellectual functioning and proposed relying on the person's level of adaptive functioning as the determinant of the severity of intellectual disability, because adaptive behavior is a better estimate of the person's overall functioning and level of support needs. With the proper level of instruction and support, people with intellectual disability can learn new adaptive skills throughout their life and their overall functioning will generally improve [77].

Adaptive behavior is a complex separate and independent construct of intellectual functioning and equally essential in documentation of ID/IDD. It includes conceptual, social, and practical skills, which can be assessed through a number of robust and reliable instruments. Teaching and promoting the acquisition of adaptive behavior should be an essential goal of

any intervention. Adequate and personalized supports can improve one's adaptive skills as well as his/her overall functioning and quality of life.

1.7 IQ and Cognitive Functions

Measurement of intelligence on the basis of the IQ score plays a central role in the classification of ID/IDD and often represents the only measure undertaken to assess persons with early cognitive difficulties. As mentioned above, an IQ significantly below the general population mean of the population (usually >2 standard deviation units) is the common reference for ID/IDD, and this score is assumed to estimate a person's ability to manage environmental demands and produce adaptive behaviors. This process can result in the burden of diagnostic labels that may fail to capture performance on specific cognitive functions. In fact, evidence from neuropsychology, genetics, neuroimaging, and functional anatomy has demonstrated high variability across different cognitive abilities within individuals [96, 97], thus challenging the current concept of unitary intelligence and the use of IQ.

1.7.1 Uni-component Models of Intelligence

Historically, Spearman [98] was the first to propose a uni-component model of intelligence. He observed that test scores on different cognitive tasks correlated with each other and concluded that this was explained by one underlying common factor, that is, "g factor." In the last 30 years, several studies found statistically significant correlations between neural activation, particularly within the frontal lobes, and performance on a number of tests of reasoning and intelligence [99, 100]. Other researchers found more widespread gray and white matter regions, distributed across several lobes of the brain, to be associated with performance on intelligence tests, a theory which has prevailed to the current day [101, 102]. Research involving individuals with focal brain damage has found activations associated with both the g factor [103] and executive functions, such as working memory, verbal comprehension, and perceptual organization, in both frontal and parietal cortices [104], supporting a distributed intelligence network.

Studies have also reported that executive functions underlying the g factor could vary independently of each other, and that IQ score stability could also hide marked variations in verbal and performance abilities [102]. Furthermore, gray matter co-varying with the g factor does not always belong to those cortical regions suggested to be the seat of general intelligence [103].

In evaluating the utility of psychometric theories supporting a uni-component model, it is necessary to consider whether a deficit in a single cognitive function may have a neuropsychological overshadowing effect, resulting in an artificially lower IQ score, or whether a low IQ score is incorrectly assumed to be the explanation for any anomaly of neuropsychological functioning [105]. These potential pitfalls arise, given that the tools for assessing IQ were not originally developed to evaluate below average performance, but to measure a child's abilities that might be predictive of academic achievement that would indicate the need for additional support. For the same reasons, such tools cannot measure IQ below 40. Hence, this floor effect precludes investigation of how different severities of ID/IDD may be associated with distinct and heterogeneous forms of cognitive functioning, as well as the associations with other variables of interest, such as genetic factors or vulnerability to psychopathological features.

1.7.2 Multicomponent Models of Intelligence

Many scientists theorized that human cognition could not be explained by a unique factor. Luria [106] postulated the existence of three basic units, resulting from interactions between different brain structures: attentional–arousal, integration-sensory input, and executive planning organization. Gardner [107] questioned the validity of IQ and IQ tests as indicators of cognitive functioning,

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claiming that the representation of the intelligence requires a combination of multiple, specific abilities, which he called "talents." With the theory called "Planning, Attention, Simultaneous, and Successive—PASS," Das and Naglieri [108–110] proposed the existence of interdependent but separate functional systems and developed a measure of individual differences on the basis of cognitive processing, called Cognitive Assessment System (CAS; [111]). Finally, in Goleman's theory of emotional intelligence [112], there is a wide array of competencies and skills which are theorized to drive adjustment and satisfaction in life. According to Anderson's Minimal Cognitive Architecture model [113], cognitive functions represent hierarchically organized and interconnected schemas, according to their role in executing a task or behavior. Individuals who are faster in cognitive processing and in gathering information are more likely to solve problems and have higher IQ [114]. The most widely accepted model of multicomponent intelligence was prompted by Cattell and Horn's theory of fluid and crystallized intelligence and subsequently integrated it within Carroll's concept of triple stratification. This model, upon which the most recent versions of WISC and WAIS have been developed, postulates the existence of nine functions at a broad level, and over 70 more specific, narrow skills. Broad functions include quantitative reasoning, auditory and visual processing, processing speed, reading and writing skills, and long- and short-term memory [115]. As theories of intelligence have evolved over the years, from a single factor to more differentiated intelligence structures, new instruments for measuring IQ have increasingly included scores for subindices, but their utility to describe cognitive profiles of persons with ID across the range of severity has remained questionable.

1.7.3 Specific Cognitive Functions and Executive Functions

There is little consensus on what executive functions actually mean and how they are distinguished from specific cognitive functions. In the neuropsychological literature, they are described as higher order abilities involving (1) volition—determining what one needs or wants and conceptualizing its future realization; (2) planning and decision-making—organizing the steps needed to carry out intentions and/or achieve goals; (3) purposive action—maintaining, switching, and stopping activity in an orderly and integrated manner; and (4) self-regulation—reduction of erratic responding and flexibility of responding to shifting task demands [116]. According to the International Classification of Functioning, Disability and Health (ICF: [32]), a cognitive profile should include both specific functions, such as memory, attention, perception, thought, space–time orientation, and executive functions, such as planning, decision-making, inhibition, regulation/correction, and action.

There are many studies reporting correlations between neuroanatomical areas and specific cognitive functions, independent of general intelligence [97]. Pascual-Leone combined various neurophysiological and brain-imaging techniques in order to identify the correlates of functioning with respect to different areas and neural systems, demonstrating that the brain represents a modular structure, and that focal damage causes only a limited impairment of overall intellectual functioning [117]. The prefrontal cortex has repeatedly been found to be active during attention switching tasks, and prefrontal cortex injury, especially on the left side, causes impairment in attention switching [118, 119]. Poor performance in switching tasks was also found in patients with lesions in the language cortex [120] or in the inferior frontal gyrus of the right hemisphere [121, 122].

In evaluating executive functions in children with ID/IDD through the Behavioural Assessment of the Dysexecutive Syndrome (BADS-C, [123]) and the Cambridge Executive Functioning Assessment (CEFA, [124]), Willner et al. [125] found scores on both tests to be only weakly related to receptive language skill, and even more weakly to IQ. Interestingly, working memory—the ability to keep something in mind while manipulating it simultaneously (e.g., repeating a string of random numbers in reverse order)—seemed to play a key role in this floor effect.

Several other studies have found significant associations between executive functions, particularly working memory, and general intelligence [96, 126]. However, some more precise assessments indicate that only updating working memory correlates with intelligence, whereas inhibiting responses and shifting mental sets do not; these two functions seemed to be related with IQ only via their covariance with updating [96, 127, 128]. Gansler and collaborators [129] found that assessment of intelligence based on executive functions accounts for more variability in activities of daily living is better predicted by health status, and was predicted less well by educational status than traditional IQ measures.

In the field of ID/IDD, researchers have found that the investigation of relationships between brain structure and specific cognitive functions to be more informative than the IQ per se.

In a recent literature review [45], neuroimaging studies of persons with Down syndrome or Fragile X syndrome were shown to be based on specific cognitive or executive functions more than on overall intelligence. Specifically, none of the five studies of persons with Down syndrome and 12 of the 15 studies of persons with Fragile X syndrome identified in the review included any overall measure of intelligence [45].

To date, knowledge on alterations of very specific cognitive functions is limited, as well as their impact on "higher order" executive functioning abilities. The most frequently studied functions are working memory and executive functions and more specifically orientation response and attention switch [45].

The case for using specific cognitive functions in defining ID/IDD is also supported by findings of recent research on cognitive and behavioral phenotypes. Different genetic syndromes with comparable IQs were associated with very different cognitive phenotypes, with respect to both relatively intact and impaired functions [45]. Similarly, general IQ scores of children with Williams–Beuren syndrome, Prader–Willi syndrome, and Fragile X syndrome, with similar sociocultural and socioeconomic backgrounds, were found to be associated with significant differences in verbal IQ and verbal and performance subtests. Vocabulary and comprehension subtest scores were significantly higher in Williams–Beuren syndrome in comparison with Prader–Willi and Fragile X syndromes, and block design and object assembly scores were significantly higher in Prader–Willi syndrome than in Williams–Beuren and Fragile X syndromes [130].

Variability between and within phenotypes is also present in autism co-occurring with ID/IDD. Of particular interest is the finding that in people with autism, low IQ scores are not necessarily associated with impairment of overall cognitive functioning, but with abnormalities in information processing, which in turn have pervasive effects on the overall functioning of the individual [131, 132].

In summary, individual differences in specific cognitive functions are highly relevant in differentiating ID/IDD phenotypes, and in understanding their biological underpinnings, whereas IQ measures do not provide such differentiation.

Unfortunately, to date, knowledge of alterations of very specific cognitive functions in ID/IDD of different origins is limited, as well as the impact of such specific functions on "higher order" executive functioning abilities. In one of the few studies aimed at this, Scerif and collaborators [133] compared visual attention in Fragile X and Williams syndrome, showing that early manifestations of inhibitory deficits affect disengaging and set-shifting abilities in Fragile X syndrome and selective attention in Williams syndrome, with different implications for impulsivity and self-control, working memory, and organization of thoughts and behavior to reach a goal (planning, self-correcting, verifying, and adapting).

1.7.4 Integration of Cognitive and Emotional Processes

Assessment of IQ does not include measurement of emotional skills. In spite of this omission, the reciprocal influence between emotion and cognition has received considerable attention, with very interesting models being proposed, including those by Ciompi, Plutchik,

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or LeDoux [45, 134–137]. It has been suggested that an emotionally charged activating stimulus or a condition of emotional distress may affect the quality of an individual's cognitive performance [138], particularly memory recall [139].

Investigation of the interplay between emotional and cognitive processes has been fostered by a new conceptualization of mental disorder proposed in a project titled Research Domain Criteria (RDoC, [140]). The RDoC defines the relationship between behaviors and brain activities, and correlates clinical phenomena to the functioning state of neurobiological circuits [140]. The RDoC framework proposed the "construct" as the basic unit of analysis, which summarizes all data related to a specific domain of functioning, that is, genetic, molecular, anatomical, behavioral, and symptomatology. This model pays particular attention to the study of emotion, motivation, and social processes and their relationships with cognitive functioning. Five constructs were defined, such as negative affectivity, positive affectivity, cognition, social behavior, and arousal/regulator system. The "cognition" construct includes major cognitive functions, indicated by the prevailing literature, to comprise the neural basis of behavior, that is, attention, perception, working memory, declarative memory, language, and cognitive control.

In recent decades, the relationship between cognition, emotion, and social interaction has increasingly become the object of study in many areas of psychology and cognitive neuroscience with the name of "social cognition." Social cognitive processes have been clustered in three main domains: (a) perceptual processing of social information such as facial expression and emotional body language (social perception), (b) grasping others' cognitive or emotional states (social understanding), and (c) planning behaviors and acting taking into consideration one's own and others goals (social decision-making) [141]. Impairments in one or more of these domains represent a prominent concern in the several cases in which ID/IDD co-occur with autism spectrum disorder.

Another relationship between cognition and affect, not measured by IQ tests, has been theorized by Salovey and Mayer and further developed by Goleman through the concept of emotional Intelligence [142, 143]. According to Mayer and Salovey, emotional intelligence is "the ability to perceive emotions, to access and generate emotions so as to assist thought, to understand emotions and emotional knowledge, and to reflectively regulate emotions so as to promote emotional and intellectual growth [143]." Goleman focused on the more functional aspects of interpersonal and personal strengths and agreed with Salovey and Mayer in dividing emotional intelligence into the following five components: self-awareness, self-regulation, internal motivation, empathy, and social skills [112]. Self-awareness is the ability to recognize and understand personal moods, emotions, and drives, as well as their effect on others. Hallmarks of self-awareness include self-confidence, realistic self-assessment, and a self-deprecating sense of humor. Self-regulation consists of the ability to control or redirect disruptive impulses and moods, and the propensity to suspend judgment and to think before acting. Hallmarks include trustworthiness and integrity, comfort with ambiguity, and openness to change. Internal motivation is a drive to action linked to an inner vision of what is important in life, a joy in doing something, curiosity in learning, and a disposition to pursue goals with energy and persistence. Hallmarks include a strong determination in achieving aims and optimism even in the face of challenges. Empathy consists of the ability to understand others' emotional makeup and treating people according to their emotional reactions. Social skills are expressed by aptitude in managing relationships and building social networks, mainly through the identification of a common ground with others. A common hallmark of social skills is represented by effectiveness in building and maintaining between-individuals and group relationships. According to Goleman, emotional competencies within each component of emotional intelligence are not innate, but rather learned capabilities that must be worked on and can be developed to achieve outstanding performance [142]. The concept of emotional intelligence and its measures have been criticized,

for (a) not being a construct of intelligence or cognitive ability; (b) lacking correlations between its components; (c) being a merger or conflate of previous psychological constructs, such as relational skills and emotional states; (d) having little predictive value for success in higher education, work, long-term romantic relationships, etc.; and (e) not having validated measures [144–146].

There is a dearth of research examining emotional intelligence in persons with ID/IDD. A few studies focusing on younger individuals found difficulties in domains like social cognition and emotional knowledge, although explicit reference to the term *emotional intelligence* is limited to two studies [147–152]. On the other hand, a recent literature review concludes that persons with ID possess emotional capacities similar to persons with typical development, with some exceptions depending on specific cause and severity level of ID/IDD [153].

A construct in the social cognitive sciences to explain the association of cognitive and emotional processes in respect to social understanding is represented by the "Theory of Mind" (ToM) mechanism, which is defined as the ability to understand and take into account another individual's mental state [154–156]. Although there is still disagreement about the precise nature of the ToM mechanisms, there is a widely accepted consensus that what needs to be explained about social understanding is the capacity for the so-called mentalizing or mindreading, which has been considered to include at least three relatively distinct cognitive abilities: to (a) attribute mental states to the hidden minds of others, (b) explain the behavior of others in terms of these hidden mental states, and (c) predict their future behavior accordingly [157]. A large number of studies show that ToM develops over the lifespan [158–160], but most of the research has been conducted with children of preschool and elementary school age, displaying low levels of ToM development [161–163]. ToM has also been criticized and other processes, for example, embodied interaction, have been proposed to play an essential role in social understanding [157].

Further descriptions of ToM and characterization in persons with autism spectrum disorder (ASD) are provided in ▶ Chap. 16.

1.7.5 Concluding Considerations on IQ and Cognitive Functions

IQ scores alone can be limited when used as an indicator of the complexity and dynamic nature of human cognitive functioning [164]. A common and comprehensive model of intelligence, which fully reconciles both general cognitive capacity and specific cognitive abilities, is lacking. Even though there is evidence to support both the uni-component and multicomponent models of intelligence, the latter appear to be more appropriate for explaining the high variability of cognitive functioning in ID/IDD. Indeed, experimental data indicate that the same IQ score can correspond to very different cognitive profiles as well as functional capacities.

Neuropsychological studies indicate that traditional intelligence tests have notable limitations, most notably capturing those cognitive functions and sub-functions in which the literature has supported as being more sensitive to individual abilities, brain injury, and disabilities [116]. The most frequently used assessment tools may provide incomplete and non-personalized data, lacking real-world validity in clinical practice with persons with ID/IDD. Cumulative scores, referring to macro-areas of cognitive functioning (e.g., verbal abilities, perceptual abilities), often fail to capture the effectiveness or importance of single skills and cognitive domains (e.g., self-control, learning ability). The DSM-5 indicates that "IQ test scores are approximations of conceptual functioning but may be insufficient to assess reasoning in real-life situations and mastery of practical tasks" and that consequently "a person with an IQ score above 70 may have such severe adaptive behaviour problems" . .. that their "actual functioning is comparable to that of individuals with a lower IQ score."

Full-scale IQ is a metric that many researchers and clinicians consider to be

outmoded and ready to be augmented by more meaningful indicators [165–167]. The WPA-SPID and the first working group for ICD-11 proposed a diagnostic approach complementing measurement of IQ with assessment of specific cognitive functions, and a contextualized description of consequent adaptive and learning difficulties [33, 44, 168]. This approach was adopted also by the DSM-5; in the chapter on diagnostic features of ID (Intellectual Developmental Disorder), it states that "Individual cognitive profiles based on neuropsychological testing are more useful for understanding intellectual abilities than a single IQ score. Such testing may identify areas of relative strengths and weaknesses, an assessment important for academic and vocational planning." Within this approach, cognitive skills should be assessed in the most comprehensive way possible, through direct clinical examination, semi-structured observations, and tests, referring to complex executive functioning, including perceptual reasoning, processing speed, verbal comprehension, as well as to very specific cognitive functions, such as attention orientation, attention switch, visual–spatial perception, or working memory.

The evaluation should aim to identify the neuropsychological characteristics that have the greatest impact on the person's quality of life, not only via cognitive skills but also associated behaviors, personal skills, adjustment, and autonomy. This would require professionals to be familiar with several instruments, in order to quickly select the most appropriate ones on the basis of the person's characteristics and the evaluation context, and cultural background.

This approach may favor the understanding of the link between cognitive alterations and psychopathological vulnerability across the lifespan, as well as bring enormous advantages to a more inclusive cultural attitude toward ID/IDD and other neurodevelopmental disabilities, providing a paradigm shift from "intellectually below average IQ" to "neuropsychological characterization of relative strengths and weaknesses." According to this paradigm, every person would have a neuropsychological profile specific to themselves. However, some people with and without ID/IDD could even share one or more specific cognitive dysfunctions and be distinguished only by their severity and their impact on individual functioning.

IQ scores alone can be limiting when used as an indicator of the complexity and dynamic nature of human cognitive functioning. Even though there is evidence to support both the uni-component and multicomponent models of intelligence, the latter appears to be more appropriate to explain the high variability of cognitive functioning in persons with ID/IDD. Indeed, experimental data indicate that the same IQ score can correspond to very different cognitive profiles. A comprehensive consideration of cognitive functioning may favor the understanding of the link between cognitive alterations and psychopathological vulnerability across the lifespan, as well as bring enormous advantages to a more inclusive cultural attitude toward ID/IDD and other neurodevelopmental disabilities, providing a paradigm shift from "intellectually below average IQ" to "neuropsychological characterization of relative strengths and weaknesses."

1.8 Development

A multitude of meanings is associated with the term "development," which can be regarded as the process of change in the person's acquisition of skills and abilities finalized to ultimately reach their human potential and to understand their role in the society. Although there can be a great variability, in its typical course the developmental process of children brings them progressively from the condition of complete dependency on others to achieving independence of their needs and well-being. Starting from these premises, by definition children with ID/IDD are those who develop at a rate significantly below average and make progresses at a rate that is

significantly slower than the one expected for children of the same age. Nevertheless, also the development of children with ID/IDD varies from person to person, since many conditions underlying ID/IDD have unique developmental trajectories [169]. Moreover, dynamic interactions between biological and environmental factors can influence the course of the development over time [170]. Indeed cognitive, language, emotional and social abilities, and competences are specifically influenced by experience and environmental factors, generally following a hierarchical sequence in their evolution [171]. Consequently, the timetable and rates of achievement of developmental milestones for children with ID/IDD can be difficult to predict as well as eventual resulting outcomes in the adult age and the acquisition or the disruption of the skills specifically associated with each developmental period can present unique challenges for a child or adolescent with ID/IDD and his/her family or caregivers (Table 1.2).

Divergence from typical developmental sequences may be particularly evident when secondary to the presence of specific genetic syndromes. For example, severe to profound ID/IDD and an early period of developmental regression preceded by a phase of normal development are typically present in Rett syndrome [172]. Therefore, an early diagnosis of ID/IDD is critical, also because children with delayed or no identified diagnosis usually progress poorer than those with a clear diagnosis [173]. Finally, clinical attention should not only be paid to childhood but also to all developmental critical periods [174, 175], since understanding the dynamics of development in different sensitive periods can guide the timing of interventions in order to improve the trajectory and global outcome of development [176].

1.8.1 Infancy

The presence of ID/IDD can modify or disrupt the typical development of attachment [177–180], self-regulation [181], and environmental awareness and exploration during infancy. For example, some functions linked to the strengthening of attachment, such as eye contact or social smile, can be absent in children with ID, and the presence of an autism spectrum disorder often linked to ID/IDD can further present obstacles to the attachment process [182–184]. Moreover, postnatal hospitalizations and prolonged medical interventions in cases of severe neurological or physical conditions can secondarily interfere with normal attachment. Conversely, delays in motor coordination and/or comorbid medical disorders, such as seizures, often create a status of dependence on family members or caregivers that can result in excessive parental vigilance. In this context, families that receive a diagnosis of ID/IDD for their child often experience anger, denial and grief, and frustration and can question their parental skills, with feelings that can interfere with the attachment processes [177, 178, 185, 186].

1.8.2 Early Childhood

When not identified during infancy, ID/IDD can be usually diagnosed during early childhood.

Language development is frequently delayed or altered in children with ID/IDD, and deficits in communication development have been proven to be early predictors of behavioral difficulties in persons with ID/IDD. Indeed, frustration at not being able to fully or entirely communicate needs or desires may lead to disruptive or self-injurious behavior [183, 187, 188]. Moreover, poor social cues can also be secondary to an inability to successfully drive the communication process apart from the difficulties in following the flow of the communication. As a consequence, social withdrawal, isolation, or increased reliance on selected caregivers may be inadvertently reinforced. Therefore, great clinical attention has to be paid to language difficulties, since early interventions must not be delayed. Moreover, children with specific language disorders may develop effective alternative communication systems to express their needs [189].

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Table 1.2 Main developmental stages and issues associated with ID across the lifespan

Infancy	Attachment Self-regulation Environmental awareness Exploration Overdependence on caregivers Social withdrawal
Early childhood	Self-care skills delay Personal mastery and self-esteem Frustration and tension with parents Overprotection and inhibition of individual initiative Communication needs if not met properly can lead to challenging behaviors Isolation or increased reliance on selected caregivers Delay or lack of spontaneous play, substituted by undirected or self-stimulatory behavior
Childhood	Taunting and rejection by peers Increasing awareness of one's limitations Social withdrawal and isolation Depression Externalizing or acting-out behavior Difficulties in meeting language and abstract concepts demands Learning skills
Adolescence	Academic failure Education goals change from life skills to vocational activities Difficulty in adapting to life changes Dependence on parents, effect on self-esteem, and learning of social skills Increased relationship and emotional difficulties Issues related to delay of puberty and sexual development Risk of abuse and bullying
Adulthood	Independent living Self-reliance in fulfilling roles Employment and other vocational activities Intimacy and reciprocity in relationships Psychopathological vulnerability Parental aging and ill-health issues
Aging	Emotions are more predictable and less labile Investments toward meaningful others increase Negative emotions become more infrequent Emotions associated with negative life experiences may be more intense and affect physiological functioning Aim of control processes shifts from changing the world to changing the self Vulnerability to age-related physical and mental health problems Increase in support needs

Considering the type of play typical for the age, the beginning of spontaneous and meaningful play may be delayed or completely missing in children with ID/IDD [190]. Indeed, in severe forms of ID/IDD, undirected or self-stimulatory behavior can be present instead of appropriate play. In the case of children with mild/moderate ID, some forms of symbolic play may develop when they are about to enter school, and also isolated or parallel play may predominate especially when communication skills are significantly impaired.

The presence of fine and gross motor delays can interfere with the acquisition of self-care skills. For those with more severe forms of ID/IDD, there is the possibility of a lifelong inability to perform activities of daily

living [191]. In the case of less severe forms of ID/IDD, self-care skills acquisition can only be delayed or is partially incomplete compared with children of the same age without ID/IDD [192].

Finally, many factors during early childhood can influence the development of the child's personality [193]. The acquisition of adaptive, communication, emotional and self-regulation skills, and how caregivers contribute to the entire process can have significant implications. Indeed, parental response, both emotionally and in terms of expectations, can greatly impact children in the development of personal mastery. The parent–child relationship may suffer from unrealistic expectations by the family and/or caregivers, while minimizing expectations may inhibit the acquisition of skills and generate frustration and lack of initiative in the child with ID [194]. For example, children with less severe ID/IDD may express the desire to perform different tasks without the appropriate skills, and this can lead to increased conflict with family members and/or caregivers. For children with milder severity of ID/IDD, self-esteem and perceived trust can form the basis of interpersonal relationships and can help to build a sense of self in the world. For children with more severe delays, the caregiver's ability to assist the child effectively in helping to regulate the different responses to internal and environmental stimuli can contribute to create a better lifelong style of behavior.

1.8.3 Childhood

Entry into school may coincide with the first exposure for children with ID/IDD to peers without disabilities. Therefore, it can be regarded as a challenging time, particularly for children with mild ID/IDD. This can be the first time for a child with ID/IDD to encounter terms such as "slow learner," or to feel different, or to experience peer taunting or rejection, although increased academic mainstreaming has elevated the awareness of many typical children regarding disabilities [195, 196]. As a consequence, isolation and social withdrawal can be expected, as well as depression or anger with externalizing and/or acting-out behaviors [197]. Most children with ID/IDD need special resource support in the classroom, since they may particularly struggle with progressively more demanding language concepts and abstract tasks. In this regard, teachers caring for many children with different special needs are challenged to work on specific communication and behavioral skills of the individual child [198, 199].

1.8.4 Adolescence

For youth with ID/IDD and their families, adolescence represents the most challenging developmental phase within the whole lifespan. Issues related to the transition from childhood to adulthood may be delayed or exaggerated, especially in becoming independent from parents, self-esteem, and social skills [200, 201]. Relationships inside and outside the family become more complex, and environmental factors have a deeper impact on the person's development than in previous ages, with consequent relational and emotional difficulties much greater than in adolescents of the general population. Problems in adapting to life changes are often expressed behaviorally, since the adolescent may be unable to communicate their ensuing frustration and confusion. These difficulties are further worsened by the delay of puberty, sexual development, and related body changes [202, 203], and the acquisition of social skills in comparison to peers. This gap increases the risk of abuse and bullying [204]. Also, families' experiences become even more challenging than before, since the person with ID may exacerbate their problem behaviors and increase the family burden and stigma [205].

During adolescence, academic failures can be common. In the case of the presence of more severe delays, the education program of the adolescent will shift from preparation to higher education to the acquisition and consolidation of life skills [206, 207] and vocational activities. Moreover, this life period is also typically characterized by the participation in extracurricular and community activities. In this context, the preparation of the

adolescent to the achievement of developmental tasks related to the social role and expectations of the society in general are crucial to ease the transition to adult life.

1.8.5 Adulthood

Key developmental milestones of adult life include emotional autonomy, independent living, self-reliance in fulfilling roles, employment, intimacy, and reciprocity in relationships. Delays and difficulties in reaching these stages, which depend on interaction of a number of personal and environmental factors, can determine or aggravate problems of self-esteem, environmental mastery, emotional control, adaptation, and stigma [208–210]. A common causal factor is represented by an overprotective or custodial attitude, which can contribute to delay the acquisition of autonomy and favor the development of a sense of insecurity and inadequacy, which can in turn reinforce submissive, renouncing, or obstinate and challenging behaviors [211].

In many cases, the continuation of these psychological adversities can contribute to the onset of psychopathological co-occurrences, especially in the context of anxiety and mood disorders [212–214].

1.8.6 Aging

During aging, experienced emotions are more predictable and less labile. Social networks narrow, but investments toward meaningful others increase. Negative emotions become more infrequent, but those associated with negative life experiences may be more intense and affect physiological functioning and ultimately physical health.

In general, primary control processes directed at modifying the environment to meet one's wishes leave the scene to secondary control processes during late adulthood, with a shift to change the self in order to be in line with the environment [215]. In the case of persons with ID/IDD, this observation is not frequently true, since they often lack resort to secondary control strategies of adjusting expectations and therefore have a high probability to fail to pursue more attainable goals when certain primary control goals become unattainable during aging.

Furthermore, psychophysical decline associated with aging occurs earlier and more intensely in people with ID/IDD than in the general population [216]. Sensory declines are common and can negatively impact communication and social interaction. Physiological functioning is regulated less well and physical reserves decrease, although occasionally some protection can be given by improvement of self-regulation. Age-related psychophysical problems and consequent contextual changes require more support, especially in case of parental death, and many people with ID/IDD have to end their lives in community residential settings [217]. In the last decades, life expectancy of people with ID/IDD and low-functioning ASD has progressively increased, reflecting a proportionate rise of issues related to moving from the family home after the death of parents and the mourning process [218].

It has been acknowledged that the experience of bereavement in people with ID/IDD is globally similar to that in the general population [218, 219], although there are some studies indicating relevant differences, especially in terms of prolonged and atypical grief which is often unrecognized [220–222], and a definitive position is yet to be reached.

Particular conditions related to ID/IDD are associated with life-shortening and degenerative illnesses, such as Down syndrome, which is linked with the early onset of Alzheimer's disease [223].

1.8.7 Emotional Development

The importance of emotional aspects in the definition of the developmental level for every person with ID/IDD has been underlined by many authors [224–226], with some of them going so far as to say that persons with ID are emotionally impaired or follow a different emotional development in respect to typically developing persons [227–229]. Generally, individuals with ID/IDD pass through the same

periods of emotional development as typically developing persons do, although with an increased risk of delay and incompleteness [227, 230–234]. The level of emotional development seems to correlate with the severity of ID/IDD and the co-occurrence of ASD [234]. Nevertheless, quality of evidence on emotional development in persons with ID/IDD is very limited, and no consensus has been reached on definitions and assessment procedures or on the relationship with adaptive skills, cognitive level, behavioral issues, and co-occurrence of psychopathology.

Some authors suggest that at each level of emotional development, different emotional needs and motivations, coping abilities, adaptive skills, and consequently different behavioral patterns can be found [227, 235–237], in combination and interaction with the well-known social, sensorimotor, and cognitive functions [238–241].

Newborns perceive and respond to a wide range of stimuli by expressing simple emotions; later, the emotional response begins to be modulated by the effect of the caregiver or interacting partner [242, 243]. The subsequent development of "joint attention," which denotes the emergence of affective sharing, appears around the second year of life [244, 245]. Around the third year of life, emotion regulation achieves more complex stages, and children become able to manipulate the emotional states of others [246]. Preschool children begin to have a basic insight into the causes and consequences of emotions and are able to differentiate and regulate their affective states, while further development of empathy, pro-social and moral thinking, and behaviors are observed in school-aged children [247].

The most famous developmental model specific for people with ID/IDD is the one developed by Anton Došen and collaborators. It includes five levels of socio-emotional development, which are aligned to the above-mentioned milestones of the developmental trajectories and the associated maturation of the respective brain circuits [227, 233, 248].

1.8.8 Conclusive Considerations on Development

The longitudinal trajectory of ID/IDD can be variable and distinct from that of typical development, especially when specific etiological conditions are taken into account [249, 250]. The different patterns of the longitudinal course can include a progressive trajectory often associated with metabolic disorders (e.g., the manifestations of the underlying process occur after birth and evolve over time into a progressively deteriorating course); an acquired trajectory with normal course of development up to the point of an insult with subsequent modification of the course of development (e.g., traumatic brain injury or severe psychosocial deprivation); and a non-progressive linear trajectory especially in environmental-related disorders with fluctuating developmental course, with periods of stress, and periods of nurturance, or positive intervention, or both [33]. The consideration of these different developmental patterns has relevant consequences for the diagnostic process. Indeed, "at any given cross-sectional encounter point with convergence of the various possible trajectories it is difficult for a clinician to make an informed decision about the developmental course of ID/IDD." Since many genetic, metabolic, and environmental factors can lead to the development of ID/IDD across the lifespan, it is, therefore, especially vital to undertake full medical history, physical examination, and requisite workup, to determine the principal cause of ID/IDD [251]. In this context, the introduction of a longitudinal developmental perspective is likely to improve the understanding of how different presentations of ID/IDD, with or without a known etiology, may change and evolve over time. In addition, this approach could lead to a better definition of how different genotypes of known conditions associated with IDD may lead to specific behaviors and cognitive profiles during specific periods of the lifespan [33]. For example, young children with Down syndrome show increasingly better receptive language skills in advance of

expressive abilities [252, 253], while boys with Fragile X syndrome show more pronounced patterns of higher level abilities in simultaneous processing than in sequential processing over the course of their development [254].

- Persons with ID/IDD develop at a significantly below average rate and slower than the general population, depending on a dynamic interaction between several biological and environmental factors. Nevertheless, the timetable and rates of achievement of developmental milestones can vary considerably from one person to another. An early diagnosis and monitoring of ID/IDD and associated developmental and psychopathological issues is crucial, since the understanding of the dynamics of development in different sensitive periods can guide the timing of interventions and improve the global outcome.

- In recent years, increasing attention is paid to emotional aspects in the definition of the developmental level.

1.9 Integrated Assessment

1.9.1 Integrated (Multidimensional) Assessment

As described above, persons with ID/IDD present a complex multidimensional and multi-systemic vulnerability, which expresses itself through a high rate of physical and mental ill-health, as well as problem behaviors. Thus, a well-timed, sensitive, and comprehensive assessment represents a fundamental process to define their overall health condition appropriately. This assessment should include as many aspects as possible within all health-threatening areas, such as biological, psychological, and socio-environmental.

To date, scientific evidence confirms that integration of different aspects of care has many advantages, including improvements in personal outcomes and diagnostic and prognostic abilities of health professionals ([255, 256]; see also ▶ Chap. 13).

With the term "integrated assessment," we refer to a comprehensive process aimed to collect quantitative and qualitative data through the use of multiple standardized and informal instruments and procedures. Comprehensive assessment data obtained should be integrated, interpreted, and summarized, including indirect and preexisting sources. The primary aim of this kind of assessment is to enable multidisciplinary team to make accurate diagnoses and to define a profile of the person's strengths, weaknesses, and needs, in order to plan the most effective interventions.

To conduct an integrated and multidisciplinary assessment, different professionals must work together to build up a shared project with the person and his/her family; these professionals must also be able to create connections of meaning and value concerning information collected, personal history, and his/her experiences. Bio-psycho-social approaches encourage the use of multidisciplinary case formulation to integrate different information, to explain the development and maintenance of physical and mental health problems, and to plan adequate interventions to address those problems [257].

1.9.2 Comprehensive Assessment and Evaluation

Professionals conducting a comprehensive and integrated assessment should have a good knowledge of the condition of ID/IDD in order to identify individual peculiarities and the need for specific adjustments.

No single data source is sufficient: the use of different informants, records, measures, procedures, and practices is necessary to enable multidisciplinary teams to have a comprehensive and reliable assessment.

The first step to carry out the assessment process is to collect a detailed history of both personal and medical aspects. Then, the condition of ID/IDD itself has to be well defined, including level of IQ, specific cogni-

tive impairments (i.e., attention orientation, attention switch, working memory, etc.), language, functioning, adaptive skills, and physical issues, before moving to the assessment of current physical and mental status, including behavioral profile, and concluding with the evaluation of person-centered general features, especially quality of life.

1.9.2.1 Medical and Personal History

Information on an individual's medical history should consider the following aspects: maternal pregnancy, delivery progress, abnormalities at birth, specific investigations, diagnosis of ID/IDD, presence of organic/cognitive/behavioral disorders, hospitalizations, and pharmacological and non-pharmacological treatments. Primary care and community services should aim to ensure that older people with ID/IDD and or ASD can see the same healthcare practitioners, wherever possible, to help practitioners to become familiar with the person's medical history, to build a good relationship with the person, and to understand his/her usual behavior and communication needs.

Personal history should be targeted at least at the following:

- Family: original and current composition of the family, cultural and economic status, quality of relationships between members, most significant relationships, and average state of health, and psychiatric familiarity
- Social belonging outside the family: relationships outside the family with individuals and groups, quality of these relationships, level, and quality of participation in community life
- Life conditions: place of birth, quality of the housing and environment (including the level of external stimulation, the involvement in various activities, etc.) in which the person lived, home/house moving, level of education, supports for learning, training conducted, the current employment status, and preferred places
- Personal interests
- Strengths and weaknesses

1.9.2.2 Definition of the Condition of ID/IDD

Assessment of cognitive functioning provides information about a person's intellectual strengths and difficulties. Intelligence testing is usually undertaken using the Wechsler Adult Intelligence Scale (WAIS-IV, [258]). This tool is designed to measure intelligence in adolescents and adults aged between 16 and 90 years. The WAIS-IV determines four individual scores, which are combined to derive the IQ score. The four individual scores are related to verbal comprehension, working memory, perceptual reasoning, and processing speed. The Leiter International Performance Scale-Revised (Leiter-R; [259]) is another standardized nonverbal measure of intelligence, often used to estimate the nonverbal problem-solving potential of individuals who do not use speech. The test is particularly suitable for children and adolescents, from 2 to 20 years, with cognitive impairment and or verbal disorders. The Leiter-R is composed of standardized batteries: Visualization and Reasoning (VR), which consists of 10 subtests for the measurement of nonverbal cognitive skills related to visualization, spatial abilities and reasoning, and Attention and Memory (AM), also consisting of 10 subtests.

Assessing and empowering personal functioning of people with ID/IDD is crucial in planning rehabilitative interventions. In the International Classification of Functioning (ICF; [32]), disability and functioning are viewed as outcomes of interactions between health conditions and contextual factors. The latter includes both external environmental factors and internal personal factors, which refers to personal characteristics such as gender, age, coping styles, social background, education, profession, past and current experience, overall behavior pattern, and other factors that influence how the person experiences disability. ICF classifies three levels of human functioning: functioning at the level of body or body part, the whole person, and the whole person in a social context. According to this classification, disability involves dysfunctioning at one or more levels: impairments,

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activity limitations, and participation restrictions. The definitions of these components of ICF can be summarized as follows: body functions (including psychological functions), body structures, impairments, activity, participation, activity limitations, participation restriction, and environmental factors.

Language

Communication Assessment Profile (CASP) [260] assesses the communicative abilities of adults with severe to mild learning disabilities. It examines eight aspects of language and communication, including the person's understanding and use of everyday words and sentences. Sound discrimination and conversational skills are also assessed.

Another tool that can also be used with people with ID/IDD is the Peabody Picture Vocabulary Test, Fourth Edition (PPVT-4) [261]. It is one of the most commonly used assessment tests that measure verbal ability in standard American English vocabulary. It measures the receptive processing of examinees from 2 to over 90 years old.

Adaptive Skills

Adaptive behavior refers to the set of conceptual, social, and practical skills that have been learned by the persons to function in their daily life [262].

The Vineland Adaptive Behavior Scales (Vineland-3; [71, 263]) and the Adaptive Behavior Assessment System (ABAS II; [264]) are the best known and most widely used tools for the assessment of adaptive skills. They detail conceptual, social, and practical domains into a variety of areas, including conceptual skills (i.e., arithmetic skills, knowing numbers and shapes), communication (i.e., comprehension and production of written and oral language), daily living skills (i.e., self-care, home living, professional activities), socialization (i.e., interpersonal relationships, social problem-solving, recreational activities, respect of social norms, community use), health and safety (i.e., following safety rules, showing caution when needed, staying out of danger, and knowing when to get help), leisure (i.e., playing, hobbies, following rules in games, planning fun activities), self-direction (i.e., self -control, making choices, starting and completing tasks, following a routine, and following directions), and motor skills (both gross and fine).

ID/IDD is a condition which is maintained throughout the lifespan, and it complicates individual transitions between ages, which are often difficult in itself (e.g., from adolescence to adulthood, from adulthood to seniority). Different contexts that are commonly associated with the different life phases may represent significant obstacles for people with ID/IDD. Nevertheless, consideration of these transition phases and, in general, the adoption of a lifespan perspective by healthcare services is often limited. All the professionals involved should be aware of the critical points of transition and their responsibilities.

Emotional Development

Emotional aspects deserve a place in the assessment of every person with ID/IDD. In fact, emotional impairment or peculiarities in emotional development are common in many people with ID/IDD. Furthermore, emotional development is reported to influence and be influenced by changes occurring in other areas, like the motor or linguistic, cognitive, and social [33]. The most common assessment tool of emotional development in people with ID/IDD of any age is the "Scheme of Appraisal of Emotional Development" (SAED) [227], a semi-structured interview that evaluates the achieved developmental level in 10 basic aspects of emotional development, with the following domains: Dealing with his/her own body, interaction with a caregiver, experience of self, object permanency, anxiety, interaction with peers, handling of material objects, verbal communication, affect differentiation, and aggression regulation. According to the five possible developmental levels (adaptation, socialization, individuation, identification, reality awareness), values from 1 to 5 can be assigned in this ordinal scaled measure corresponding to certain developmental achievements and age equivalents in typically developing children. Two modified versions of the SAED have been produced during the first half of the last decade Scale for Emotional Development—Revised (SED-R) [265] and

Scale for Emotional Development—Second Revision (SED-R 2) [266], keeping the same five-stage model of emotional development, but adding three domains. More recently, a short, psychometrically sound and adult-oriented version has also been developed, with the name of SED-S (Scale of Emotional Development—Short) [267].

1.9.2.3 Physical Assessment

In persons with ID/IDD, the identification of physical health problems is usually more complex than in the general population. This occurrence is due to several peculiarities in the perception, presentation, and communication of symptoms. In particular, the discrepancy in primary healthcare received by people with ID is due to the following barriers: mobility and sensory impairments, behavior problems, language difficulties, and inadequate knowledge and attitudes of staff. First of all, in order to conduct an appropriate and comprehensive assessment, healthcare professionals have to make some adjustments so that people with ID/IDD may have equal access to medical care. Practitioners should explain clearly to a person with ID/IDD what will happen during any medical appointments or examinations. If the person agrees, they should also share the information with his/her family member, carer, or advocate. This evaluation should be carried out in a place that is familiar, welcoming, and appropriate to personal needs. Managers in healthcare settings should provide adequate information to family members and carers so that they can help people with ID/IDD to find an appropriate service. Alternatively, they should identify a practitioner with experience in the field to contact people with ID and their family members, carers, and advocates [268]. In the context of assessment, professionals should discuss changes that may occur with age with persons with ID/IDD and their family members, asking them about symptoms of common age-related conditions or changes in any existing conditions, including blood pressure and serum cholesterol level, diabetes, hearing loss, and sight problems, osteoporosis, malnutrition, dementia, or mental health problems. They should give people accessible and clear advice about keeping well as they grow, for example, telling them about, and helping them access, services such as breast screening, smear tests, testicular and prostate checks, dental checks, hearing and sight tests, and podiatry [268] (see resource section in ▶ https://spectrom.wixsite.com/project and also ▶ https://www.nds.org.au/images/resources/Supporting-Practice-Leadership%2D%2D-A-collation-of-resources.pdf).

1.9.2.4 Mental State Assessment

In persons with ID/IDD, the assessment of mental functioning and evaluation of psychopathology is even more challenging than physical assessment. There are further difficulties associated with the lack of laboratory and instrumental markers for the co-occurrence of psychiatric syndromes. Besides the identification or exclusion of psychopathology, a comprehensive assessment should also focus on social conditions and psychosocial stressors.

Family relationships may significantly affect social and psychological well-being even after the person with ID/IDD has left their family home. The family environment can be seen as the primary agent of socialization. The needs, resources, priorities, and concerns of the family should also be identified. The family assessment is usually conducted through an interview with the parents.

Another factor to take into account is the exposure to life events, traumatic experiences, and environmental stressors, which seems to be particularly high in people with ID/IDD [269] (see ▶ Chap. 3). Several studies have shown that multiple life events can have an impact on behavior and mental health, determining changes in a person's emotional well-being, and quality of life [57, 270–273]. Therefore, professionals who conduct the integrated assessment should pay particular attention to these aspects, recording all the events considered relevant for the person, both positive and negative.

A detailed description of the mental state examination and the way it has to be carried out in persons with ID/IDD and low-functioning autism spectrum disorder is provided in ▶ Chap. 16.

1.9.2.5 Assessment of Problem Behaviors (ABA and Others)

As previously discussed, problem behaviors (PBs) are common in individuals with ID/IDD and can upset an individual's social, occupational, or academic daily functioning [274]. Mental health professionals who evaluate PBs should collect information about their topography, frequency, duration, intensity, and potential functions. There are three main types of behavioral assessment: standardized assessment, descriptive assessment, and functional analysis. Standardized assessment involves the administration of measures in which parents or other caregivers provide information about the person's behavior; in descriptive assessment, professionals evaluate situations in which the behavior occurs by observing the person in his/her natural setting and recording events that precede and follow behavior. Functional analysis methodology focuses on the identification of variables that influence the occurrence of problem behavior [275]. It is a process that identifies specific targeted behavior, the purpose of the behavior, and what factors maintain it. For assessment of internal and external predisposing, precipitating, and perpetuating factors for problem behavior, the Comprehensive Assessment of Triggers for behavior Scale (CATS) could be used (see ► https://spectrom.wixsite.com/project) (see ► Chap. 7).

1.9.2.6 Support Needs (Support Intensity Scale and Similar Measures)

Supports are resources and strategies that promote the development, education, interests, and personal well-being of a person and enhance individual functioning [262]. The term "support needs" refers to the pattern and intensity of supports necessary for a person to participate in activities linked with normative human functioning.

The Support Intensity Scale (SIS) [276] measures the individual's support needs in personal, work-related, and social activities to identify and describe the types and intensity of the support a person requires. The SIS was designed to be part of person-centered care processes that help all individuals identify their unique preferences, skills, and life goals (► https://aaidd.org/publications/supports-intensity-scale#.Ww6sLTihcdU).

The SIS is composed of three sections that investigate different aspects as follows: the first section asks about the support people may need with various activities (e.g., activities at home, in the community, and in the workplace), health and safety, social activities, and learning during the lifespan; the second section asks about a person's protection and advocacy-related support needs; and the third section investigates the person's unique medical and behavioral support needs.

Individual needs change over time; therefore, support must change as well. They must be developed and provided in age-appropriate settings, always considering that the person with ID/IDD should have the opportunity to engage in daily activities just like any other person, regardless of their intellectual abilities or impairments. Examples of other needs assessment instruments for people with ID/IDD are (a) Learning Disability version of the Cardinal Needs Schedule (LDCNS) [277] and (b) Camberwell Assessment of Need for Adults with Developmental and Intellectual Disabilities (CANDID) [278].

1.9.2.7 Quality of Life

An integrated assessment, as described above, promotes a holistic vision in which health and social problems are interconnected, allowing the identification of links between the different systems. Integrated approaches assume fundamental importance in evaluating personal well-being, experiences, satisfaction, and aspirations of people with ID/IDD and must be carefully considered for the implementation of interventions that translate into improvements of individual Quality of Life (QoL). The most recent literature suggests that the evaluation of QoL as the outcome measure of therapeutic interventions has discriminative capacity and sensitivity; moreover, its ability to indicate the real well-being of the person has resulted in being superior to all the efficacy parameters previously used [279, 280].

Thus, all the assessors should share results and feedback, including with the person with ID and their family members (see ► Chap. 15).

General References for an Integrated (Multidisciplinary) Assessment

Instruments and Procedures for Comprehensive Assessment and Evaluation

Accurate information about a person's status and needs must be derived from a variety of assessment tools and procedures to obtain a comprehensive set of quantitative and qualitative data. A comprehensive, integrated assessment should

1. Use a valid and the most current version of standardized assessment instruments.
2. Use multiple measures and data sources, including case history and interviews with parents, carers, relevant professionals, and the person with ID (if appropriate), the information provided by parents; direct observations or data-based information in multiple settings and on more than one situation; and standardized tests that are reliable and valid, as well as culturally, linguistically, developmentally, and age-appropriate.
3. Examine functioning and or ability levels across domains of motor, sensory, cognitive, communication, and behavior, including specific areas of cognitive and integrative difficulties in perception; memory; attention; sequencing; motor planning and coordination; and thinking, reasoning, and organization.
4. Integrate standardized and non-standardized data collected.

Professionals should evaluate personal functioning and skills across domains of motor, sensory, cognitive, communication, and behavior, including impairments in perception, memory, attention, planning and coordination, and reasoning. Finally, interdisciplinary team members should integrate and discuss the information gathered from using both standardized and non-standardized instruments.

Box 1.1 Advice for a multidisciplinary approach and inter-agency collaboration

- Sharing information on a need to know basis with everyone involved in the care of the person with ID/IDD including their family caregivers and where appropriate with the person with ID/IDD themselves
- Interdisciplinary and inter-agency case management and formulation of intervention
- Sharing assessment paths with all relevant services and stakeholders, including educational and disability services
- Sharing inter-agency and interdisciplinary resources for an effective outcome of assessment and intervention
- Promoting educational and training activities involving components of the mental health service network and subsequently of all the other sectors involved in the support of the person with ID/IDD (services for disability, health, education, and inclusion in the community)

Families and Advocates

Staff working with adults with ID/IDD should work in collaboration with their families and caregivers, providing the necessary support and trying to establish a good relationship with them, characterized by trust and empathy.

It is extremely important to involve parents in the assessment to obtain detailed information regarding the person's past. Important background information includes past psychiatric or medical information, past drug and other treatments, family history of psychiatric and medical problems, and details of the person's skills and abilities. It may also be appropriate to involve additional informants, such as other relatives and caregivers.

It is also useful to involve various professionals in the role of evaluators and to search for different informants. Health services

should increase information for families, carers, and other advocates on key components of the comprehensive assessment and evaluation process, expanding meaningful ways in which they can be involved in the process itself.

Consent and Participation in Assessment

Professionals who conduct the assessment should consider several factors which can influence a person with ID's ability to understand and to express his/her opinions (► www.intellectualdisability.info/mental-health/articles/classification-and-assessment-of-psychiatric-disorders-in-adults-with-intellectual-disabilities).

The seriousness and the type of disability influence individual's communication skills, together with a series of factors, such as neurological factors, psychiatric disorders, drug side effects, the setting, and the communication style of the others. Also, the tendency to acquiescence may represent a limitation to the collection of information. Therefore, all these factors should be taken into account to improve communication (see ► Box 1.1 and ◘ Table 1.3). Service providers should ensure that care and support for people with ID are tailored to their needs, strengths, and preferences.

In order to provide an effective and complete assessment, professionals should find out how the person prefers to communicate and receive information; give people written information (such as appointment letters and reminders) in an accessible format (see examples of accessible psychotropic medication leaflets; ► https://spectrom.wixsite.com/project); use visual aids and short, clear sentences during consultations and conversations; and talk to the person's family members and carers if appropriate, and with the person's consent. If necessary, services should provide an independent interpreter so that people can communicate in their first language.

Healthcare and social care practitioners should encourage people to express their views and make their own decisions, assess their capacity to make decisions, and involve family members, carers, and advocates, when appropriate. With the person's consent, they should actively involve members of the person's support network in the planning and delivery of their care and share information with their family members, carers, or advocate, for example, about any changes that might be needed to their support. It is crucial to determine the needs and preferences of the person, ensuring these are not overshadowed by the preferences of others, including when the person lacks capacity. Consent and participation in assessment have to be pursued also taking into account local legal rules, such as the mental health act and mental capacity act in the UK or the Law 180 in Italy.

Concluding Considerations on Assessment

Resources for effective assessment (e.g., appropriate and current assessment tools, continuing professional development), evidence-based and high-quality instructions, time for data collection and analysis, and collaboration should be provided. Services providers should take responsibility for making available to people with ID and their families professionals with experiences and support that promote the implementation of positive practices based on research. Policy-makers, administrators, and educators should examine the findings of the assessment to ensure that people with ID/IDD are provided with opportunities to perform at achievement levels beyond their chronological age or grade. Each member of the team must consider the person's cultural and linguistic background when choosing and interpreting assessments. Behaviors that may be viewed as part of autism spectrum disorders or symptomatology in one culture may have a different interpretation in another culture (e.g., not establishing eye contact is often considered a characteristic of autism, while in some cultures direct eye contact can be viewed as disrespectful or rude) [281, 282]. The assessment team will need to become familiar with the pattern of behaviors demonstrated by the person. Working as a team, the professionals involved in the assessment should also observe the person's behavior and performance across different environments.

Table 1.3 Advice for an optimal setting to assess persons with ID/IDD

Context	Offer a time frame which is appropriate to the characteristics of the person
Environment	Provide a relaxing, familiar/friendly environment (for example, the person's home) Minimize physical barriers Use sittings suitable for the disability of the person Position yourself in a way which is adequate to communicate with the person and his/her carers (e.g., continually looking at them or do not stay too far). Minimize environmental noise (close the window, switch off the TV, etc.)
Considering the presence of sensory disabilities	
In the case of vision problems: Attract/focus the person's attention, for example, by starting the sentence with his or her name or touching his/her hand or arm gently before speaking In case of hearing problems: Position yourself so that the person can see your mouth and face; in case of unilateral impairment, position yourself on the side where the hearing is better Check that the person wearing his hearing aid is turned on, and the battery is not discharged Articulate clearly and slowly Use all the communication devices that the person has at his/her disposal Ensure that you have caught the person's attention before starting to speak, for example, touching his/her hand or arm	
People with ID/IDD with verbal communication skills	
Be an active listener Ask for clarifications to verify that you understand correctly Use clear language and short sentences Avoid technical terms Avoid intellectually complex concepts (to be concrete) Avoid sentences with complex constructions, such as those with conditional verbs Repeat the questions and, if necessary, reformulate them in a simplified Verify that the person has understood the question Allow the person with ID/IDD time to answer before moving to the next question Prefer open questions Articulate clearly	
People with ID without verbal communication skills	
Look at the person you are communicating with Use intonation, body language, and posture for encouraging the person to express himself/herself Support interaction with drawings and symbols (having available materials like illustrated volumes can be useful) Some people can communicate through sign language, both standardized and personalized Some people need tools and technological supports Some people may need the help of a speech therapist, a communication facilitator/partner, or another professional Ask the person's communication partner what is the person's prefer the way of communication	
Interview techniques	
A. Making some simple questions at the beginning of the interview will help the person with ID/IDD feel comfortable B. Evaluate any discrepancy between expressive and receptive language C. In some cases, the use of images, drawings, or other communication methods maybe necessary D. When possible, avoid using direct questions (e.g., avoiding questions like "you do not like living here, do you?" In favor of "how are you at home?") E. Use simple and appropriate language F. Avoid double senses, metaphors, idiomatic expressions, medical jargon, etc. G. Make sure that the person understands H. If necessary, repeat the question several times	

1

- Well-timed, sensitive, and comprehensive assessment represents a fundamental process to appropriately define overall health condition of persons with ID/IDD and a profile of strengths, weaknesses, and needs, in order to plan the most effective interventions. Most relevant aspects to be assessed include personal and medical history, the condition of ID/IDD itself (specific cognitive impairments, language, adaptive skills, functioning), current physical and mental status, behavioral profile, personal experiences, and quality of life.
- To conduct an integrated and multidisciplinary assessment, different professionals must work together to build up a shared project with the person and his/her family.

Tip

It is hoped that future research and practice will give increasing importance to the condition of ID/IDD, as it currently represents a disregarded topic in psychiatry and, to a lesser extent, in overall medicine.

ID/IDD represents a complex syndrome grouping (meta-syndrome), which needs a polynomious-polysemic approach to be named and defined for different audiences and purposes. The evolution of its diagnostic criteria attempted to maximize clinical consensus but masked a wide degree of heterogeneity between and within individuals for many aspects. To understand this multi-level heterogeneity, future research and practice should follow a transdiagnostic approach utilizing organizing concepts that move beyond unitary intelligence and developmental comorbidity. This is particularly relevant in the current neuroscientific context, which is progressing toward precision medicine, person-centered, lifespan-informed, shared decision-making, and collaborative planning of care to provide holistic support for each unique individual with ID/IDD.

Key Points

- The nosographic approach to ID/IDD has considerably changed across time. The present chapter reports the evolution of definitions and diagnostic criteria.
- ID/IDD refers to a group of etiologically diverse conditions originating during the developmental period characterized by significantly below average intellectual functioning and adaptive behavior.
- Models of specific cognitive functions reflect more faithfully the characterization and the efficiency of individual intellectual profile than IQ measure only.
- Dynamic interactions between biological and environmental factors can influence the course of the individual development over time, generally following a hierarchical sequence in their evolution.
- A multidimensional and interdisciplinary assessment is fundamental to appropriately define overall health condition of persons with ID/IDD and a profile of strengths, weaknesses, and needs as well as to plan the most effective interventions.

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Prevalence and Aetiopathogenesis of Intellectual Developmental Disorders

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Contents

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Learning Objectives

The primary objective of this chapter is to provide an overview of the epidemiology of intellectual disability by providing data from more recent research. The aim was not to be comprehensive but to provide the reader an understanding of the prevalence, incidence, mortality rates, risk factors, and mental health services–related epidemiology and identify the major gaps in knowledge that require further research. Aetiology and treatment modalities are dealt with in more detail in subsequent chapters, but this chapter provides a broader perspective on some aetiological causes and availability and quality of public mental health services related to intellectual disability across different countries.

2.1 Introduction

Communities understanding of intellectual disability (ID) and its definition have varied over the centuries [1–4]. The way the community has treated people with intellectual disabilities (PWIDs) has also been chequered with numerous instances of abuse and neglect [5]. Even now, PWIDs continue to be ostracized and abused in many communities, and even their families suffer stigma and negative perceptions. However, given these negative perceptions, there are also positive developments that have happened over the decades to improve the lives of PWIDs. First, new screening methods have not only led to early detection of genetic abnormalities but also led to taking corrective steps where possible. Second, numerous evidence-based mental health services and supportive care facilities, including academic and vocational skills training, have been developed to cater to the needs of PWIDs. Third, both national and international policies have been framed to safeguard the interests of PWIDs globally.

The definition of ID has also evolved over the decades as more was understood about aetiology of ID, and ability of PWIDs to adapt to their disability and function within the society. Currently, ID is defined as "a disability characterized by significant limitations both in intellectual functioning and in adaptive behaviour, which covers many everyday social and practical skills" [6]. The onset of ID is determined by a mix of genetic and environmental factors and often manifests in early childhood. With age, PWIDs adapt their behaviours to their needs. This depends on the severity of their disorder and support systems available to them. They are able to participate at different function levels in the community, participate in routine activities and professional or academic activities and contribute effectively to the society. Current norm for assessing ID takes this into account and includes a detailed evaluation of adaptive behaviours, besides an intellectual quotient (IQ). Adaptive behaviour includes "conceptual skills [such as] language and literacy; money, time, and number concepts, and self-direction; social skills [like] interpersonal skills, social responsibility, self-esteem, gullibility, naïveté (i.e., wariness), social problem solving, and the ability to follow rules/obey laws and to avoid being victimized; and practical skills [which include] activities of daily living (personal care), occupational skills, healthcare, travel/transportation, schedules/routines, safety, use of money, use of the telephone" [6].

Given the current global population of 7.3 billion and estimated prevalence of 1% [7], there are about 73 million PWIDs, worldwide. It is critical to not only understand the causes for ID but also understand the types of health services available for PWIDs. Better knowledge about the epidemiology of IDs and available services will help improve the services, conduct research to plug the gaps in knowledge and inform policy-makers to develop strategies to improve the lives of PWIDs. This chapter summarizes the epidemiology of ID and related health services for PWIDs and provides a broad understanding of ID.

2.2 Prevalence of Intellectual Disabilities

2

At the outset, it is important to recognize that it is difficult to compare prevalence studies as they vastly differ in terms of conceptualization of ID, diagnostic criteria followed and the methods adopted to identify ID. King and colleagues [3] while discussing the various factors that tend to influence the prevalence of ID globally observed that definitions used for ID by different surveys determine how ID is identified in the community; age of ascertainment also plays a crucial role as with age adaptive behaviours modify the functional ability of PWIDs; identification of ID also varies according to age based on the specific health condition causing it (with 10 years being a modal age for identification globally); different mortality rates for PWIDs that vary by age. Identification commonly occurs when one starts schooling, so often that is the age when point prevalence shows a peak. Additionally, the prevalence is also determined by the survey instruments used and populations surveyed, such as community/clinical/special institution-based studies.

The prevalence of ID is estimated between 1% and 3%, and the incidence is estimated to be around 1.8% [4, 8]. In developed countries like Finland or the Netherlands, the prevalence of ID is currently less than 1%, while it may rise to 4–5% in more deprived regions of the world and can reach up to 6% in some countries in Eastern Europe [9]. Among the four different severity degrees, 85% have mild ID, 10% moderate, 4% severe and 2% profound [3]. Mild ID being present in 80 to 85% of the world's population [10] could explain why many PWIDs are not identified until they reach school age and start having difficulties acquiring and learning new skills. The prevalence of Borderline Intellectual Functioning (BIF) is estimated to be 12.3%, and Hassiotis and collaborators [11] found that in a sample of 8.450 adults living as part of a family in the United Kingdom, approximately one-eighth had BIF. The prevalence of ID is higher in males than females in both adults and children/adolescents. Among adults, the male-to-female ratio varied between 0.7 and 0.9 [7].

The National Health Interview Survey provides trends in the prevalence of ID among 3–17-year-olds from 1996 to 2008 in the United States. Boyle and colleagues [12] while reporting on the trends found it to be relatively stable around 7/1000 children with the latest round of data from 2006 to 2008 providing an estimate of 6.7/1000 children. However, over the same period the prevalence of learning disorders increased by 5.5% while that of ID reduced marginally, which might be a reflection of administrative data classifying more cases as learning disorders compared to ID, in order to reduce stigma associated with ID. A meta-analysis of 52 community-based studies has observed an overall prevalence of 10.37 ID cases per 1000 population in the world. The prevalence was highest in the low-income countries at 16.41 cases per 1000 population, followed by middle-income countries (15.94 cases per 1000 population) and lowest in high-income countries at 9.21 cases per 1000 population [7]. Highest rates were seen among children/adolescents, those from rural or urban slum populations, and among females. McKenzie and colleagues [13] conducted a systematic review of the prevalence and incidence of intellectual disabilities between 2010 and 2015 and reported a range of prevalence between 0.05% and 1.55%. They offered several reasons for this variation. First, the IQ levels of general population which increases at regular intervals due to environmental factors, a phenomenon known as Flynn effect, are not captured reliably by IQ tests. To overcome the Flynn effect, IQ tests are re-standardized at regular intervals to increase the difficulty level of test items. But a disadvantage is that more people fall below the IQ of 70 [14, 15]. Therefore, IQ scores are not sufficient to diagnose ID. Second, definition of ID has been modified over the numerous revisions of the

standard diagnostic systems which resulted in modifications of the diagnostic criteria of ID. There is still a debate among researchers about whether to include adaptive behaviour capabilities on top of IQ score, or use each of them separately. The American Association of Intellectual and Developmental Disabilities outlines 10 domains of adaptive behaviour and uses both IQ and adaptive behaviours to identify ID and its severity [6]. ID is closely associated with autism spectrum disorder, and trends show that the prevalence of autism spectrum disorder is increasing but prevalence of ID is decreasing because of a diagnostic preference for autism in response to administrative policies of providing services [16–18]. Third, male-to-female ratio is decreasing. The prevalence of autism is higher among males, and it is also believed that between 40% and 60% of individuals with autism spectrum disorder have ID [19, 20]. Fourth, prenatal diagnosis for certain categories of ID, namely, Down's syndrome, Edwards syndrome and Patau syndrome resulting in medical terminations of pregnancies, is also contributing to the decrease in ID prevalence. Additionally, improved public health and risk prevention strategies such as educating about alcohol consumption during pregnancies and reduction in lead-contaminated drinking water may have lowered the risk of ID in population [21]. Fifth, differences in population characteristics, average parental age, increasing maternal age and sex ratio have also been identified as the factors for variations in the prevalence and incidence of ID [13, 22]. King and collaborators [3] found that mild, moderate, severe and profound ID is reported in 85%, 10%, 4% and 2% of the population affected with ID, respectively. A brief summary of more recent studies on prevalence of ID is given in Table 2.1.

The prevalence of ID is estimated between 1% and 3%, and the incidence is estimated to be around 1.8%. Prevalence seems to be higher in children and adolescents, those in low- and middle-income countries, and among females.

2.3 Incidence of Intellectual Disabilities

Compared to prevalence, incidence is a more effective measure of magnitude of ID. However, very few incidence studies have been conducted. A longitudinal study on children with median age of 14 years in Sweden reported cumulative incidence of 0.62% [23], while another study on adults with 50 years of age reported cumulative incidence of 1.58% among males and 0.96% among females [24]. Based upon two cohort studies conducted in Finland in 1985–1986 (n = 9432) and in 1966 (n = 11,965), the cumulative incidence of ID was found 12.6/1000 for ID [8]. However, Heikura and colleagues [8] reported no change in incidence of profound category but a mild shift rate from severe and moderate towards mild ID between 1966 and 1986 cohorts. The cumulative incidence for mild ID was observed to be 7.5/1000 compared to 5/1000 in younger cohort. Katusic and collaborators [25] have reported differences in incidence of ID with reference to gender and degree of ID in the United States. In a cohort of 5919 children born between 1976 and 1980, the cumulative incidence for severe ID was observed more than twice among females than males, and for mild ID it was twice in males than females. For males, cumulative incidence of ID was reported to be 8.3/1000 population and for females 10/1000 population. However, cumulative incidence was found to be 1.7 times greater in males than females [25]. In an epidemiological study of 5070 babies born between 1978 and 1987 in Japan, the incidence of ID was 0.87%. The incidence of ID was 0.51% for mild and 0.36% for severe category [26].

2.4 Mortality in Intellectual Disabilities

Lifespan of people with ID has improved significantly due to advancement in health care, rehabilitation services and personal care. However, worldwide, the mortality is signifi-

Table 2.1 Trends in prevalence of ID

Country	Authors and year	Sample features	Prevalence
Australia	Haider et al. 2013 [160]	Adult population survey	0.1%
	Leonard et al. 2011 [161]	Children/adolescents and adult's population survey	1.3%
Canada	Lin et al. 2013 [162]	Administrative health data used to derive estimates of the prevalence of adult ID	0.1%
China	Zheng et al. 2011 [163]	Nationally representative surveys conducted in 1987 and 2006; age-adjusted prevalence of ID estimated	1.3%
Denmark	Pedersen et al. 2014 [24]	Data of residents who received health services between 2000 to 2012	0.1%
India	Girimaji and Srinath 2010 [146]	2064 children aged 0–16 years were screened for ID on ICD 10 criteria	3%
	Lakhan and Mawson 2016 [22]	Community survey; a total of 8797 tribal population were screened for ID; prevalence of ID among children up to age 18 years was reported	0.6%
Italy	ISTAT 2018 [164]; Salvini F., 2018 [165]	Students with ID	1.9%
USA	Boyle et al. 2011 [12]	Data analysis of National Health Interview Survey (NHIS) datasets 1997–2008; data collected every 3 years from children aged 3–17 years	Rates at each time point 1997, 2000, 2003, and 2006 were 6.8/1000, 7.3/1000, 7.5/1000, and 6.7/1000, respectively
	Braun et al. 2015 [19]	Population-based developmental disabilities surveillance programme data from 1991 to 2010 for 8-year-olds in metropolitan Atlanta	1.6%

cantly higher in ID population compared to their age-matched non-ID peers [27–29]. It appears that the reduction of life expectancy is positively correlated with the severity of ID [30]. In Finland, Patja and colleagues [31] followed a cohort of more than 2400 PWIDs for 35 years and estimated the mortality rate to be 18/1000 person-years, with mortality increasing with severity. Data from Scotland showed that survival rates at 76 years were significantly lower among children with ID [32]. The relative risk of survival reduced to 0.79 and 0.63 for an IQ level that was one and two standard deviations below normal, respectively. Thorpe and colleagues [33] have found a positive relationship between factors such as mood disorder, poor baseline functioning, poor cognitive abilities and psychotropic medication and mortality in PWIDs [33]. Some specific health conditions such as increased prevalence of comorbid Alzheimer's disease and cardiovascular disorders have been associated with ID and also lead to increased mortality [34]. Some of the early work and recent studies on mortality, survival rate and life expectancy are presented in Table 2.2.

Community-based data show that the cumulative incidence of ID was found to be 12.6/1000 for ID. Mortality rate is about 18/1000 person-years, increasing with ID severity. Some syndrome-specific

Table 2.2 Recent estimations of mortality, survival and life expectancy

Country	Authors and year	Sample features	Mortality	Remarks
Australia	Bittles et al. 2002 [166]	Analysed data of 8724 people with ID for their survival rate	The life expectancy was found to be 74 years for mild ID, 67.6 years for moderate ID and 58.6 years for people with severe ID	The strong positive association was found between severity of ID and lower life expectancy rate
England	Tyrer and McGrother 2009 [167]	A cohort of 503 people with ID was followed up	17% of the cohort with moderate and profound ID died within 14 years	The standardized mortality rate was very high for congenital abnormalities (28%), nervous system (5.4%), mental disorders (3.7%) and bronchopneumonia (2.1%)
Finland	Patja et al. 2001 [31]	To investigate cause-specific mortality in ID, a nationally representative sample of 2369 ID cohort was followed up for 35 years	18 per 1000 person-years	The mortality was positively associated with severity of ID. Compared to general population, specific conditions such as cardiovascular and respiratory diseases were found highly associated with mortality in people with ID. But there were no differences in mortality rate with reference to gender
Germany	Dieckmann et al. 2015 [27]	Two sample data of ID population from two different geographical locations in Germany for 2007 to 2009 were analysed and compared with the general population	The life expectancy for males (70.9 years) was higher than females (72.8 years) in Westphalia–Lippe; but lower for males (65.3 years) than the females (69.9 years) in Baden–Wuerttemberg	The life expectancy for male and female ID was found improved, but it was found lower than the general population. The life expectancy of ID population was also found varied in terms of geographical locations
India	Lakhan and Kishore 2016b [29]	Analysed National Sample Survey Organisation data for 2002	The mortality rate in people with ID increases with age in India, particularly after the age of 19 years	This is secondary analysis of the data so it did not provide any information if any specific variable of ID were associated with ID

(continued)

Table 2.2 (continued)

Country	Authors and year	Sample features	Mortality	Remarks
Israel	Merrick 2002 [168]	The data of division for mental retardation of Israel were analysed for 53 residential centres, 1280 persons in community-related residential facilities and about 13,000 persons who participate in day care facilities	A study reported a mortality rate of 10.25 deaths per 1000 population for people with ID living in residential care facilities between 1991 and 1997	It is much higher than the rates reported for their counterparts availing the community day care facilities
Netherland	Maaskant et al. 2002 [28]	Data of 29,290 children with ID aged 5 years, who were registered in the National Case Register of Netherland between 1991 and 1995 were analysed for mortality	The survival age was found to be 41 years for ID and 46 years for ID with Down's syndrome	The mortality rate did not differ with gender
UK	Tyrer et al. 2007 [169]	Adults with ID of age 20 years or over ($N = 2436$), with moderate to profound ID living in Leicestershire and Rutland, UK, between 1993 and 2005 were analysed with the general population of 700,000 people	The cause-specific standardized mortality rate (SMR) was found nine times higher in men (SMR = 883; 95% CI 560–1325) and 17 times higher in women (SMR = 1722; 95% CI = 964–2840) in their 20s compared to the general population of this age	Study found three times higher all-cause and disease-specific mortality rate among ID people compared to the general population, with particular disadvantage for Down's syndrome
USA	Shavelle et al. 2014 [170]	California Department of Developmental Services database of 64,207 people age 5 and older was analysed from 2000 to 2010	The excess death rate increased with age from 0.1 to 6.8/1000 in moderate to mild and 3.4 to 6.7 in profound to severe ID	This study indicates that the mortality rates increase with severity of ID

morbidities such as Alzheimer's disease and cardiovascular disorders also lead to increased mortality.

2.5 Prevalence of Co-Occurring Autism Spectrum Disorder

About 34% of PWIDs have pervasive autistic traits [35–37], while up to 60% of patients with autism have ID [38–44]. In terms of a dual diagnosis (i.e. a developmental disorder such as autistic spectrum disorder (ASD) or ID and psychiatric disorder), there is increased risk of underestimating ASD in PWIDs when schizophrenia is diagnosed [45–47]. About 40% of the items included in diagnostic tools for PWIDs and used to screen for psychosis commonly receive a high score when autism is present [48]. A further consideration giving rise to increased prevalence of ASD may be linked to inaccurate diagnosis and a growing recognition that autistic disorders can be associated with other conditions [49, 50]. In the United States, increase in prevalence was associated with declines in other diagnostic categories, indicating that diagnostic substitution had occurred [51]. A background but key issue in these considerations is to delineate robustly the boundary between ASD and ID. Both ID and ASD are metasyndromic groups to which many different clinical conditions belong [52]. However, many PWIDs have some autistic aspects, as well as many persons with ASD have lower intellectual functioning compared to the general population [20, 53].

About 34% of PWIDs have pervasive autistic traits, while up to 60% of patients with autism have ID. Many PWIDs have some autistic aspects, as well as many persons with ASD have lower intellectual functioning compared to the general population. Both ID and ASD are metasyndromic groups to which many different clinical conditions belong.

2.6 Aetiological Factors and Physical and Psychological Conditions Associated with Intellectual Disabilities

Considering that ID is a disability with implications for life-long consequences for the individual and the family, and necessitates high service provision and social costs, it is necessary to identify the causes in order to take appropriate preventive steps. ID is associated with several biological and psychosocial factors. The biological factors are further divided into genetic and non-genetic factors. The risk factors may act individually or through an interactive effect [54].

In most cases, the aetiology for ID is unknown. Genetic causes are the leading causes among the identifiable causes for ID. The etiological factors could also be antenatal, perinatal, postnatal, brain trauma during delivery, birth asphyxia, intrauterine growth retardation, infections affecting the nervous system, hypothyroidism and iodine deficiency, and lead poisoning [7, 55]. Karam and collaborators [56] have found that the aetiology of ID is 44.4% environmental, 20.5% genetics, 12.6% idiopathic, 13.2% neonatal sequelae and 9.3% other diseases. A study from India found 64.4% genetic, 20.4% perinatal, 12% central nervous system malformation, 3.6% prenatal and 2.4% postnatal causes for ID in India [57]. Vissers and collaborators [58] had reviewed genetic studies in ID and found that with new techniques in genetics research such as genomic microarrays and next-generation sequencing-based technologies, there has been a spurt in genetics-based research in ID though saturation in that area is still far away. There have been attempts to study aetiology from a cultural perspective. One such variable is consanguinity but it did not show strong association with ID [59, 60]. One consistent finding, at least in low- and middle-income countries (LMICs), is that the excess rate of ID is related to preventable aetiological factors such as teratogens,

diet deficiencies, pregnancy and birth-related conditions [61, 62]. However, the cause is unidentified in 60% of PWIDs in LMIC [60]. In general, the causes of ID are unknown for up to 60% of cases [63]; for the rest ID represents a heterogeneous group caused by a different combination of specific causative factors, both genetic and environmental [61]. Known causes are classified by the time they occur in respect to birth, as prenatal, perinatal and postnatal.

2.6.1 Prenatal Causes

2.6.1.1 Genetic Factors

A proportion of cases ranging from 17% to 50% are caused by a genetic disorder [63–66], while non-syndromic ID accounts for 30–50% of cases [67, 68]. Conventionally, genetic forms of ID are divided into two major categories: syndromic ID characterized by associated clinical, radiological, metabolic, or biological features, and non-syndromic forms in which cognitive impairment represents the only manifestation of the disease. Although this distinction remains useful for clinical approach, recent studies and detailed clinical follow-up indicated that distinctions between syndromic and non-syndromic ID are disappearing, and some of the latter could be recognized as syndromic forms [69–71]. Detailed analyses of database and literature searches reveal that more than a thousand of genes can cause ID. Furthermore, more than 290 genes are involved in clinical phenotypes, metabolic and neurological disorders associated with ID [72–74].

Genetic causes are divided into chromosomal (or genomic) mutations and gene (or point) mutations. Trisomy 21 is the chromosome abnormality responsible for over 95% of Down's syndrome, which is the best-known chromosomal cause of ID [63, 75]. Other well-known genomic syndromes are Turner syndrome [76, 77], Klinefelter syndrome [78], and Cri du Chat syndrome [79]. The most prevalent conditions due to gene mutation include Fragile X syndrome [80, 81], galactosemia [82, 83], lipidosis [84], phenylketonuria [85, 86], neurofibromatosis (or von Recklinghausen's disease) [87], Williams's syndrome [88], and Prader–Willi syndrome [89]. ID occurs in other genetic syndromes like tuberous sclerosis [90], Huntington's disease [91, 92], mucopolysaccharidosis [93], alkaptonuria [94], and porphyria [74, 95, 96]. The risk of transmission of recessive genes is greatly increased by consanguineous marriage. For a couple of first cousins, the chance of passing on chromosomal alterations is about five times higher than for couples who are not blood relatives [96]. Idiopathic ID supports the hypothesis that rare de novo point mutations can be significant causal factors of ID [97].

2.6.1.2 Epigenetic Mechanisms

Recent research has shown interest in epigenetic mechanisms, which refer to changes that affect gene activity and expression without changing the DNA sequence or, in a narrower sense, to heritable phenotype changes that do not involve DNA alterations. These two different acceptations imply a different reference to the individual's lifespan, from conception to senescence, or to trans-generational timeframe. Evidence indicates that the fidelity of the transmission of DNA methylation patterns, through which epigenetic modifications are done, is much lower than that of DNA nucleotide sequences and that therefore de novo epimutations can occur more frequently than de novo mutations.

Negative prenatal environmental factors, such as pregnancy distress, exposure to toxic substances (e.g. cannabis, cocaine, ethanol) and viral infections, seem to be able to alter the normal brain development and to contribute to the onset of a neurodevelopmental disorder with mediation of epigenetic mechanisms [98–101]. In animals, the genes most frequently involved in these mechanisms are those that encode the glucocorticoid receptor (Nr3c1) and the brain-derived neurotrophic factor [102]. In mammals, including humans, the most recent studies have focused

on two main mechanisms: epimutations during embryogenesis and epigenetic memory induced by environmental factors, with reference to the effect of pharmacological treatments. It seems to be possible that environment-induced epigenetic modifications can occur throughout an individual's lifespan, even if prenatal development is identified as the period of maximum responsiveness [103]. In this phase, an extensive "epigenetic reprogramming" can take place, that is a pluripotent redefinition of the epigenome. In cell replications that occur from the zygote formation to the blastocyst nesting, even the entire architecture of DNA methylations can undergo a wide cancellation or remodeling [102, 104]. The epigenetic modifications that occur in the last part of the gestation and after birth appear to be much lower, not only in number but also for the type of control exercised on gene expression. These modifications are defined "epigenetic programming", to distinguish it from the aforementioned "reprogramming" and to refer to the changes that guide cell differentiation, from stem cells to various types of neurons and glial cells [105, 106]. In summary, recent insights on epigenetics indicate that the DNA methylation variations determined by environmental factors that can contribute most to the pathogenesis of neurodevelopment disorders or adult psychiatric disorders (or both) are those that occur during the foetal period.

Reprogramming, which allows the zygote to erase the epigenetic markings inherited from the parental gametes, seems to find an exception in parental imprinting. Parents' genome undergoes the effect of specific epigenetic modifications during the meiosis, which is the parents' chromosomes separation to give rise to oocytes or spermatozoa. This process gives a subclass of homologous loci, called "imprinted genes", an exclusive expression of the genetic information inherited from one of the parents. Some of these genes are expressed if inherited from the father, others if inherited from the mother. The other parental allele is, in fact, maintained in a state of repression by DNA methylation. These imprinted genes play a significant role in the brain development, and incorrect imprinting or inappropriate expression is the basis of some syndromes including ID, such as Angelman, Prader–Willi, Beckwith–Wiedemann or Silver–Russell.

Mutations in Methyl CpG-binding protein 2 (MeCP2) and ATP-dependent helicase X-linked protein gene are already well established in Rett syndrome, Down's syndrome, and some X-linked cases [107–110]. However, in most cases such modifications are known to be affected by environmental factors before and after birth [111]. In recent years, a new protein able to recognize epigenetic changes has been identified. This protein, called ZFP57, was also found to contribute to the conservation of these epigenetic changes, from the embryo to the adult, and to enable their transmission to the offspring. Even more recently, the genetic structure of this protein has been defined, which allowed to confirm the hypothesis that also the mechanisms underlying epigenetic modifications have a genetic basis [112, 113].

Among the several relevant implications, epigenetics, as an expression of the complex relationship between genes and environment, may represent a possible explanation of the frequent association between negative life events and psychopathological vulnerability in persons with ID and ASD, an area of research and clinical practice that had always received limited consideration, despite the fact that its importance had been widely documented in the general population.

2.6.1.3 Biological and Environmental Factors

Prenatal causes of ID include congenital infections such as cytomegalovirus, toxoplasmosis, herpes, rubella, influenza and human immunodeficiency virus [114]. The human foetus seems unable to produce an effective immunological response in early pregnancy; the ability of children to produce antibodies increases significantly only between the 6 and

12 months of age. Exposure to radiations; pollutants; heavy metals; harmful medications, such as thalidomide, phenytoin, valproate and warfarin in early pregnancy; and additive substances, such as alcohol, nicotine and cocaine, can also cause ID [67, 115–117]. Other prenatal risk factors are maternal hypoglycaemia, diabetes, hypoxemia and malnutrition [118, 119]. Evidence from experimental studies shows that malnutrition in utero can impact on brain development leading to nervous cells depletion, protein synthesis defect and abnormal electrical activity [120].

2.6.2 Perinatal Causes

Perinatal causes involve complications of labour and delivery, severe prematurity, very low birth weight, birth asphyxia and birth trauma. Furthermore, neonatal complications in the first 4 weeks of life included septicaemia, severe jaundice and hypoglycaemia [121, 122]. Low birth weight at term delivery can be due to genetic causes. In other cases, poor nutritional contribution in uterus associated with placental insufficiency or other damaging agents may occur. These children, born small for gestational age, show retardation in foetal growth and subsequent neurological complications that are different to those related to shorter gestation period. True pre-term births before 26 weeks of gestation and pre-term births after less than 36 weeks of gestation are those at greatest risk of developing neurological damage, which increases in inverse proportion to their degree of maturity and birth weight (<1500 grams) [123, 124]. Other perinatal causes include congenital infections like those listed among prenatal causes, although most infections in newborns are caused by bacteria of the birth canal, especially if it has an active infection.

2.6.3 Postnatal Causes

During the postnatal period, which includes infancy and childhood, brain infections such as encephalitis and bacterial meningitis may damage the brain and cause ID [125]. Furthermore, encephalic traumatism, chronic lead or other toxic exposure, severe and prolonged malnutrition can also lead to ID [81, 126]. Studies report a significant association between ID and exposure to inadequate caregivers, low level of stimulation and social deprivation [127, 128]. Malnutrition and negative socio-environmental conditions are often co-present in the postnatal history of some persons with mild ID.

In most cases, aetiology of ID is unknown. Genetic causes are the most common among identifiable causes. The etiological factors could also be antenatal, perinatal and postnatal. Some of the most common are brain trauma during delivery, birth asphyxia, intrauterine growth retardation, infections affecting the nervous system, hypothyroidism and iodine deficiency, and lead poisoning.

2.7 Psychological and Physical Comorbid Conditions

PWIDs suffer from higher rates of physical and other mental disorders [129, 130] compared to general population. Cooper and colleagues [129] have found that specific physical and mental health problems were significantly higher in adults with ID, with epilepsy being the commonest. Harris [4] has also reported that hearing impairment is present in 10% of cases and epilepsy in 5–30%. Mental health problems are four to five times more common in people with ID [131, 132]. Lundqvist [133] found that about 62% of PWIDs have at least one self-injurious, stereotyped or aggressive/destructive behaviour problem and 18.6% challenging behaviours. Lakhan and Kishore [134] found a higher prevalence of violent and destructive behaviours and misbehaviour with others among moderate and mild ID, and self-injurious, temper-tantrum and stereotype among profound and severe ID. Often physical health problems lead to some form of behaviour problems in PWIDs

[135]. However, it could be understood that the aetiology and comorbid conditions may vary depending on the age targeted and the setting in which the study is carried out.

PWIDs suffer from higher rates of physical and mental disorders compared to the general population, with epilepsy being the commonest (5–30%). It has also reported that hearing impairment is present in 10% of cases. Mental health problems are four to five times more common.

2.8 Mental Health Services for People with Intellectual Disabilities

Despite the existence of national and international policies and legislation, only 39% of countries in the world have policies or programmes related to ID [136] with mental health services being limited by poor availability, accessibility and adequacy [137], apart from few exceptions [130, 138–142]. This lack of services has a relevant impact on epidemiological knowledge, both for ID itself and the co-occurrence of mental disorders. Reversely, the lack of knowledge regarding the substantial epidemiological data on the prevalence of psychopathology in persons with ID represents in many countries a potential barrier to accessing mental health services [143]. In Southeast Asian countries, mental health services show a huge variation of type and range. In Hong Kong, Taiwan and South Korea, there is provision of inpatient, outpatient, day hospital and outreach services, for both children and adults with ID, whereas Vietnam provides a narrower range and greater orientation towards children. In Latin America, South Africa, China, Greece, Asia, India, Taiwan, Serbia and Bulgaria [139, 142–149], services are largely concentrated in secondary and tertiary centres of towns and cities, have a poor connection with other care centres, and are provided by inpatient psychiatric units at district general hospitals and specialized psychiatric hospitals, which are mainly located in big university centres [144]. Even in high-income countries the implementation of mental health policy does not uniformly address the needs of persons with ID. Furthermore, mental health services specialized for ID are uncommon, models of care are generally unclear, and data on service use or mental health outcomes are not systematically collected.

Although persons with ID tend not to refer themselves to mental health services, but rather be referred by family members or other caregivers, a number of barriers relating to the personal (individual) experience of access to services have been investigated and identified, including awareness of their need for care, fear of medical personnel, as well as low expectations of services and a reduced tendency to complain [150]. Some studies identified ethnic variation in the utilization of mental health service [151–155], often related to culturally shaped ideas about ID [156].

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2.9 Gaps in Knowledge and Future Research Needs

Major gaps in the extant literature on ID are in the areas of epidemiology from LMICs. While some studies are available from LMICs the quality of such are often limited by being small clinic-based studies. There are no large cohorts from LMICs which provide rich enough data as other cohort studies such as the Olmstead County study or other cohort studies available in Europe [157]. The absence of such studies hampers the ability to iden-

tify incidence rates, causal factors, trends in prevalence, risk factors amenable to embedded interventions and outcomes, and mortality. There is also a lack of clarity about the effect of mental disorders on behavioural manifestations in PWIDs [158], and it is still to be determined how mental disorders affect behavioural manifestations and to what degree in PWIDs.

Another area of research that is improving but still has some way to go is research related to genetics of ID [58]. While new techniques of exome sequencing and genome sequencing have improved understanding of the genetic sequence for many cases, others are still unknown. Added to this is the fact that few treatments based on genetic research are available, and research needs to be undertaken to understand how treatment based on modifying genetics can be implemented in a cost-effective manner at population level.

Estimates from the United States indicate that the lifetime cost of ID was USD 1.01 million per person in 2003 (CDC), and data from Australia suggest that almost AUD 14.7 billion/year is incurred as cost for managing care for PWIDs and 85% is opportunity cost due to lost time [159]. However, similar research from LMICs is limited. Other consequences are the inadequate consideration of ID within global health issues and the increasing gap between health services for ID and unmet needs. [136].

Tip

A major gap in the extant literature on ID is in the area of epidemiology from LMICs.

While new techniques of exome sequencing and genome sequencing have definitely improved the understanding of the genetic sequence for many cases, others are still unknown.

There is a lack of clarity about the effect of mental disorders on behavioural manifestations in PWIDs.

2.10 Conclusion

The epidemiology of ID has evolved over the decades, and prevalence estimates have tended to be influenced by the changing definitions and diagnostic criteria of ID. Along with it there have been many studies to identify causal factors, and genetic factors are a major identifiable risk factor which has been the focus of research over decades. With recent advances in technology, genetics research has started to evolve too, and new areas of focus are being seen especially in the non-X-linked genetic causes. However, a lot remains to be clarified, and studies from LMICs are particularly few when it comes to understanding longitudinal trends in different parameters, including use of mental health services. Future research needs to plug those gaps in knowledge and identify newer areas of focus.

Key Points

- The prevalence of ID cases is reported as 10.37/1000 population, with higher rates in low- and middle-income countries compared to high-income countries.
- The cumulative incidence is about 12.6 cases/1000 population.
- The lifespan is directly correlated to the severity of ID.
- Though in most cases the aetiology is unknown, genetic causes are the main among identified causes.
- PWIDs suffer from higher rates of physical and mental disorders compared to the general population.
- Knowledge about appropriate mental health services for PWIDs is limited.
- Some major gaps in knowledge are lack of high-quality epidemiological studies from low- and middle-income countries; and few studies on incidence, quality of mental health services and cost-effectiveness.

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Psychological Distress and Physical Vulnerability

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Contents

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3

Learning Objectives

- Psychological distress.
- Causes of psychological distress.
- Manifestation of psychological distress.
- Common physical disorders in people with intellectual disabilities (ID).
- Reasons for increasing prevalence of physical disorders.
- Different stages of life for people with ID.

Tip

People with intellectual disabilities are at a higher risk of developing psychological distress as well as physical disorders.

3.1 Psychological Distress

3.1.1 Introduction

Psychological distress describes the experience of an individual discomforting emotional state in response to a specific stressor, threat or harmful situation [1]. A broad range of events may elicit psychological distress. These events include but are not limited to illness, loss of physical function, victimisation, social conflict and economic insecurity. Irrespective of the severity, psychological distress may contribute to the development of a range of physical and mental health disorders in those who are vulnerable, leading to significant public health concerns [2, 3].

3.1.2 High Risk Among Individuals with Intellectual Disabilities

People with intellectual disabilities (ID) experience psychological distress more often and to a higher extent compared to people without ID. Epidemiological evidence is scant because large-scale assessment of subjective distress states is difficult, especially for people with moderate to severe ID. Results for emotional distress based on proxy reports, clinical interviews and self-reports provide divergent answers [4]. However, epidemiological evidence on the rate of stressors points indirectly to high psychological distress. The rate of traumatic experiences and stressful social situations, for example, is heightened among people with ID [5–8].

3.1.3 Causes of Psychological Distress

Abuse and Violence

Individuals with ID experience high rates of violence and all forms of abuse [9]. Intentional harm experienced by individuals with ID is almost always perpetrated by someone they know, such as a family member, and is commonly exhibited as sexual, physical and verbal/emotional abuse [9]. Individuals with ID are vulnerable to even paid caregivers who may be neglectful or abusive. Compared to the general population, people with ID often experience interpersonal violence. A meta-analysis revealed prevalence figures of 21.2% for violence experiences in children and 6.1% in adults with an ID [10, 11]. An earlier study reported abuse and mistreatment of up to 68% for girls and 30% for boys with an ID [12].

Maladaptive Coping Strategies

People with ID may also experience psychological distress to a higher intensity and chronicity due to maladaptive coping with stressors as these occur to people with and without ID alike. According to Lovallo's [13] model, primary appraisal of stressors as threats or non-threats occurs in cognitive subsystems shaped by experience. If a stressor is labelled on the basis of similar experiences as a threat, it is attended to more closely and appraised in relation to one's self-perceived skills and resources. Outcome of this second-

ary appraisal spurs behavioural, cognitive and physiological responses accompanied by emotions. If people experience their skills and resources as deficient in the face of perceived threat, social engagement with others gives way to noxious cognitive and physiological reactivity as a self-preserving fall-back option [14].

Attachment Styles

People learn to appraise stressors and engage other people in addressing threats in the context of attachment relationships [15]. The low rate of secure attachment relationships among children with ID and the high rate of growing up in residential care with less opportunities for the formation of attachment relationships [16], therefore, increase the vulnerability of people with ID for psychological distress. Psychological interventions addressing attachment needs as well as facilitating the communication of distress have been found to improve stress regulation [17], facilitate the training of adaptive behavioural responses to challenging situations [18] and reduce anxiety [19].

Cognitive Biases

Young people with ID show biases in processing social information. Even innocuous interpersonal events are interpreted more often as hostile [20]. These biases increase the likelihood of escalating aggressive conflict, further exacerbating psychological distress. Attributional style and stressors independently predict depressed mood [21].

Stigma

Stigma increases psychological distress in people with ID. People with ID can be more vulnerable to the psychological distress due to stigma [22]. Some researchers have hypothesised that self-reported stigma is positively associated with psychological distress and negatively associated with quality of life, contact with services, and adherence to treatment in people with ID [23]. There is evidence to suggest that stigma and even the perception of stigma may contribute to poor psychological health in people with ID by increasing psychological distress and reducing quality of life [23].

Communication Impairment

Communication impairment and the challenges to express pain [24] and emotions in a clear and direct way are factors that make recognising psychological distress in individuals with ID difficult, making it necessary to then look at the individual's behaviour. Challenging behaviour may also be a person's attempt to get needs met when they experience lower levels of well-being [25]. The vulnerability of individuals with ID to experience traumatic physical experiences and stressful social situations combined with their inability to engage others in dealing with their emotions creates challenges for caregivers and professionals as well as medical staff.

- Individuals with ID are more likely to experience psychological distress compared to their peers without ID.
- Multiple stressful and traumatic life events, maladaptive coping strategies, attachment styles, socio-environmental factors and communication difficulties have been postulated as some of the reasons for the increased psychological distress.

3.1.4 Manifestation of Psychological Distress

Behavioural Manifestations

Manifestation of symptoms with regard to psychological distress might differ between the general and the ID population. In people with ID, symptoms are more prominently expressed on a behavioural and non-verbal level [26]. People with ID might react in states of strong psychological distress, for example, after sexual abuse, with regression, like bedwetting, or complain of diffuse pain and present symptoms of self-injurious behaviour. Mental health symptoms following trauma exposure might include dissociative behaviour, motor agitation, flight reactions and aggression. This often leads to misidentifying psychological distress or posttraumatic stress

disorder (PTSD) symptoms as symptoms of other mental health or behavioural disorders [27]. In this context, the relevance of an elaborate diagnostic protocol appears evident.

3

- **Mental Disorders**

According to Reeve [28], symptoms of psychological distress fall short of meeting the DSM-5 [29] criteria for a clinical diagnosis of anxiety. However, when left unrecognised and untreated, normal anxiety that can contribute to positive effects such as improved attention and performance can become over-learned, habitual or otherwise dysfunctional. This phenomenon can lead to disruptive behaviour, inefficient brain patterns and avoidance of pleasurable experiences, thereby contributing to psychological distress [28]. For instance, Hartley et al. [30] found that adults with ID who sought reassurance for their distress from caregiving staff to an excessive extent, more often experienced rejection and other negative responses from staff, which in turn was associated with elevated rate of depression. Psychological distress should, therefore, have an important place in diagnostic formulation, caregiver education and care planning.

Besides the higher rate to experience a traumatic event or after being exposed to repetitive traumatising incidents, people with ID show a higher vulnerability to develop PTSD. PTSD is a disorder which a person may develop after being involved in or witnessing traumatic events. PTSD may be described differently in some situations: if a person's symptoms emerge more than 6 months after experiencing trauma, this might be called "delayed-onset PTSD". If a person experienced trauma in an early age or it lasted for a long time, you might describe it as "complex PTSD". Not everyone experiencing trauma will develop PTSD. Best known protective factors are a high intelligence level, as well as reliable memory functions. Thus, a lower developmental level in intelligence, typically going along with lower cognitive competences, will raise the incidence of PTSD in people with ID. In addition, the lower levels in verbal expression often limit people with ID to report such complex experiences [31]. People with ID also suffer from painful conditions more often [32]. Improving and preventing psychologically or physically harmful conditions is therefore bound to reduce psychological distress as well [7].

- Psychological distress increases the risk of mental illness such as depressive and anxiety disorders in people with ID.
- Psychological distress can also manifest as behavioural challenges mainly as aggression.

3.1.5 Assessing Psychological Distress

The Bangor Life Events Schedule for Intellectual Disabilities (BLESID) [33] or the Lifestress Inventory (LI) [34] [5] can be used to assess psychological distress in a systematic way. The BLESID, covering a broad range of 24 life events, can be applied both, as a self- and a third-party report instrument. The Lifestress Inventory, a self-report measure, assesses current stressors with 30 items addressing daily situations or life events, with 13 items on negative interpersonal relations. Traumatic events can be assessed via the Trauma Information Form [35], and PTSD can be assessed by the Lancaster and Northgate Trauma Scale (LANTS) [36] or the Impact of Event Scale–Intellectual Disabilities (IES-IDs) [35].

3.1.6 Life Events and Traumatic Experiences

Regarding the contribution of life events (LE) to this vulnerability, evidence is sparse and methodologically limited. Many authors report that people with ID are more frequently exposed to stressful and traumatic environmental conditions than the general population [5, 31, 37–39], and this may impair their resilience to stressors [40, 41]. In particular, children with ID are resulted to be exposed three/four times more than their counterparts with normal intellectual functioning [42].

It has also been suggested that people with ID generally have more complex living

circumstances and less control over their lives [43, 44]. These conditions include the institutionalised living and dependency on caregivers [45–49], isolation, neglect and/or marginalisation [50–52], reduced and impoverished social networks [53, 54], poor housing and lower income [55], reduced access to adequate forms of affective or practical support [56].

Furthermore, the susceptibility to further adverse LE after trauma [57] may be related to dysfunctional coping strategies [58]. Some authors have identified an association between the impairment of cognitive and emotional skills [59], the inadequate levels of social support [54], and all positive and negative LE. People with ID have specific cognitive profiles [60, 61] and less strategies to cope with environmental stressors. Actually, they encounter a number of difficulties in understanding changes and relationships between events [62] as well as in processing and expressing their distress related to the past experiences [47]. Thus, they might act this out behaviourally, displaying functional problems like challenging behaviours [63, 64].

Starting from the childhood, many families and socioeconomic risk factors are associated with emotional/behavioural problems in people with ID, such as poor physical and mental health, challenging behaviour, and hyperactivity. Family and socioeconomic risk factors which have been more frequently reported are bullying, problems of justice, violence and abuse, cohabitation with a single parent, precarious or inadequate housing, parental unemployment, parents with drug addiction, living in a lower socioeconomic community [65].

As suggested by Herman and Evenhuis [66], the risk of psychiatric problems in older people with ID may be increased due to the exposure to more age-related LE than younger adults [66]. In a sample of 988 adults aged over 50, 97% had been subjected to multiple LE during the past year. The most frequently reported LE were the change of staff in setting/day care (85.05%), holiday (65.3%), new residents (55.1%), mild physical illness (47.2%) and change of main professional caregiver (39.5%). Depressive and anxiety symptoms were significantly heightened in individuals who reported more total and negative LE during the preceding year. This association remained significant also after controlling for a depressive or an anxiety disorder.

Surveys conducted in the last two decades of the twentieth century have reported positive correlations between lifetime exposure to LE and the development of physical, somatoform and psychiatric disorders in adulthood [67–79].

Martorell and colleagues [79] surveyed 177 individuals with mild to moderate ID about the impact of LE and traumatic experiences on their mental health. The retrospective survey was performed by a checklist created ad hoc and the Trauma History Screen (THS) [80]. The psychiatric evaluation was carried out through the administration of the PAS-ADD Checklist (Psychiatric Assessment Schedule for Adults with Developmental Disability) [81]. Results showed the presence of at least one traumatic event in the lifetime of 75% of the participants and at least one traumatic event within the 12 months preceding the study in 50% of cases. The statistical analysis also revealed a significant correlation between exposure to traumatic events and – to a lesser extent – LE, and the likelihood of developing a mental disorder, classified according to the ICD-10 criteria [82].

There is evidence that LE during the previous 6 months are significantly predictive of levels of psychological trauma, behavioural changes (both in frequency and severity) [31, 83] and functioning problems in general [84]. Some studies have found that the high prevalence of challenging behaviour in persons with ID is associated with LE and trauma [85, 86], although it has been confirmed for aggressive behaviour, but not for self-injurious or stereotyped behaviours [87].

Owen and colleagues [87] examined the correlations between LE, problems behaviours and mental health in a residential population of adults with ID. On average, each resident had been subjected to 3.5 negative LE in the past 12 months. Changes of staff and residence, conflicts with care staff and other housemates, family relationships and bereavements, and injuries or illnesses were the events most frequently reported by the participants.

Destructive behaviour and heightened risk for depressive/anxiety disorders were more frequently found in residents who had experienced a higher number of LE.

In 2004, Hastings [73] and collaborators evaluated a large sample of more than 1000 individuals living in various arrangements in Northeast England. The authors found a relationship between the exposure to at least one or two LE in the previous year and the pervasiveness of psychiatric symptoms. The disorders, mainly belonging to the affective spectrum, were detected with the PAS-ADD Checklist [81]. The five LE most frequently reported were changes of residence (15.5%), serious illness of relatives or close friends (9%), serious problems with close relatives or friends (8.8%), serious illness or self-injury (8.5%), and death of family members or close relatives (8.3%) [73].

The LE most frequently associated with mental health problems in people with ID is relocation [73, 77, 87], while among traumatising events abuses, especially sexual abuse, and bereavements are more likely [88–90].

Among traumas, abuse has been the most studied. Reiter and colleagues [90] found that students with ID were more frequently abused – physically, sexually and emotionally – than their peers. In addition to the higher incidence of victimisation in this population, it has also been shown that the abuse often goes unreported or, when reported, it tends to be disregarded. Analysing various life histories, some authors have revealed the presence of sexual abuse as a predictor of self-injury, misuse of alcohol and drugs, PTSD, low self-esteem, anger, depression, guilty relationship problems and behavioural problems [91–93].

Other LE and traumatic experiences observed are unemployed/seeking job, problems with police or authority, alcohol problems and major financial crisis, which have also been frequently associated with personality disorders. On the other hand, serious illnesses or injuries, laid off/sacked from work, and retirement from work have been associated with depression [89].

Different studies have highlighted a positive correlation between the exposure to LE/traumas and the presence of PTSD [83, 91, 94, 95], schizophrenia, personality disorders, depression and adjustment reactions [31, 76, 77]. Findings of a recent longitudinal study [49] have shown that LE were related to depressive, anxiety and psychotic symptoms, as well anger and aggression.

Also, dementia appears to be related to exposure to LE [75, 96]. In particular, the speed of cognitive decline has been found to correlate with the type and the number of LE. Relocations, experiences of loss/separation from significant persons and health problems seem to be the more frequent [75]. Recently, the research has focused on the role of trauma also in the aetiology of obesity [31]. Regarding the role of gender, studies have found no statistically significant differences in the type of LE between men and women [89].

In conclusion, current data confirm that the LE and/or traumas often precede psychological problems, and so they could be considered as risk factors. Evidence is still needed for a better understanding of the possible causal role of LE in the aetiology of mental disorders. The effects of negative events seen in the literature on the general population are not necessarily transferable to people with ID [36].

It is difficult to understand what the threshold is to consider identifying an experience as negative or traumatic for persons with ID. However, it is possible to conclude that an event may have a major impact on the person's ability to adapt and to manage life stressors, especially in case of more severe cognitive impairment [58, 97, 98].

Although the understanding of the role of positive and negative LE in the development of psychopathological vulnerability is becoming more and more important, the literature on traumatic experiences across the lifespan and mental health problems in people with ID is limited and not always consistent [49, 62, 73, 74, 76–78, 84, 87, 99], except for abuse [90, 100, 101], personal victimisation, bereavement [102] and PTSD [74, 75].

These difficulties in the clarification of the relationship between LE, ID and mental disorders are due to different aspects: inaccurate research methodologies, non-representative

samples and LE detection tools, which have not adequately adapted from the ones created for the general population. The instruments used often consist of self-report scales whose administration requires good cognitive, emotional and linguistic skills. Furthermore, the content of their items does not cover a wide timeframe but refers only to recent experiences [4, 5, 54, 80, 103–106].

Once these mechanisms are understood, treatment directions could be identified [49], even if the cognitive behaviour therapy and the eye movement desensitisation and reprocessing are already available and effective [31]. Future studies are needed to contribute to the understanding of the impact of LE and trauma by indicating the specific cognitive profiles of participants and by using prospective methodology [107].

3.2 Physical Vulnerability

Vulnerability is a term that is often used with regard to people with ID and other disabilities and has been defined as "…susceptibility to any kind of harm, whether physical, moral or spiritual, at the hands of an agent or agency" [108]. It is related to dynamics of disempowerment and lack of autonomy. There is, therefore, an important relationship in clinical practice between factors associated with vulnerability, and delivery and accessibility of health care through systems and clinicians. A scoping review of the concept of vulnerability and disparities in health care found evidence to support the authors' hypothesis of a direct correlation between co-existing factors of vulnerability and healthcare disparities [109].

People with ID are prone to experience physical disorders more than in the general population for a variety of reasons associated directly with ID and indirectly because of social challenges in maintaining good health [110]. Maintaining good physical health is particularly important for people with ID because of their vulnerability to developing physical health problems that can have an adverse impact on their quality of life and can be associated with significant inequalities in mortality and morbidity. This paragraph is confined to exploring the prevalence of physical disorders in people with ID and describe specific health problems.

3.2.1 Life Expectancy

Over recent decades, there has been a gradual increase in the life expectancy and a sustained reduction in age-standardised mortality rates in people with ID [111]. Life expectancy has increased from 12 years in 1949 to 60 years in 2004 for people with Down syndrome [112]. Studies have reported that people with mild ID may have similar life expectancy to the non-ID population but for people with more severe ID it is lower [113]. The life expectancy of people without a known organic cause for their ID has also gradually increased [114], but people with ID overall still continue to live shorter lives than people without ID in the United Kingdom [115].

Various studies over the last 20 years have reported on premature death in people with ID. A study in South London showed that people with ID are 58 times more likely to die before the age of 50 than the general population [116]. The MENCAP report in the United Kingdom, "Death by Indifference", on six deaths of people with ID described the role of institutional discrimination against people with ID in not providing adequate intervention and support in hospital and primary care [117]. The subsequent Independent Inquiry by Sir Jonathan Michael recommended the establishment of the Learning Disabilities Public Health Observatory in the United Kingdom [118]. A further confidential inquiry into the deaths of people with learning disabilities [119] was undertaken which investigated the known causes of death of people with ID in South West England and reported that men with ID live 13 years less and women live 20 years less than their counterparts without ID. Some of the reasons for premature deaths included significant difficulties or delays in diagnosis, or delays in further investigation, or specialist referral, and problems with their treatment [120].

3.2.2 Causes of Death

The main causes of death in people with ID are respiratory disorders and cardiovascular conditions secondary to congenital heart disease. In contrast, the leading cause of death in the general population is cancer, followed by ischaemic heart disease and cerebrovascular accidents. Cancer as a cause of death was less common compared to people in the general population [121–123]. Tyrer and McGrother [124] found a relatively high cause-specific mortality for deaths caused by congenital abnormalities and diseases of the nervous system in their study of 503 adults with ID who died during a 14-year follow-up period.

3.2.3 Aetiological Factors for Physical Illnesses and Vulnerability

The aetiological factors for physical illness in people with ID are varied and due to not only the presence of physical disorders but also inherited disorders, communication difficulties, behavioural problems, deficits in support and other social determinants of health. People with ID are at a higher risk of specific diseases compared with the non-ID general population [125]. The presence of certain genetic syndromes further increases the risk of physical illness, for example, the prevalence of hypothyroidism is significantly high in people with Down syndrome. Certain neurological conditions such as epilepsy occur more frequently in people with severe ID with underlying genetic syndromes or organic brain damage [126].

The ability to ask for help and access health care is important for physical health conditions to be diagnosed, treated and prevented. An inability to process information, make informed choices and plan to accept help may reduce a person's access to health care. Communication difficulties in describing symptoms may predispose to a lack of awareness by carers of symptoms or their misattribution to behavioural difficulties instead of identifying accurately a physical health cause. Deficits in adaptive behaviour further reduce the ability to seek assessment and treatment for physical conditions. Inability or lack of motivation to engage in activities increases risk factors such as obesity which in turn may lead to the development of physical illnesses. Lack of appropriately adjusted information and processes of identification and access, as well as lack of awareness in individuals, family and carers, also has an impact on engagement with health screening programmes, for example, breast, bowel and cervical cancers [127].

The occurrence of behavioural difficulties in people with ID can further reduce their access to health care leading to reduced opportunities for assessment, accurate diagnosis and effective treatment. People with ID are prescribed medications more than the general population, particularly in attempts to manage behavioural difficulties [128]. Such medications are associated with metabolic effects that can have significant impacts on a person's physical health, causing weight gain and predisposing them to developing diabetes mellitus.

- Individuals with ID are also more likely to experience physical disorders compared to their peers without ID.
- Studies have highlighted reduced life expectancy of people with ID compared to their peers without ID.
- Reasons for increased physical disorders and premature deaths can be associated with the aetiology of ID, communication difficulties, behavioural problems and lack of social support among many other reasons.
- People with ID can struggle to access health care for various reasons.
- Among physical disorders, epilepsy is one of the commonest conditions seen in people with ID.
- Certain physical health conditions are commonly seen among certain groups of people with ID.
- For example, people with Down syndrome and ID are at a higher risk of hypothyroidism.

3.2.4 Common Physical Health Problems

Morbidity

Physical health morbidities are high in people with ID. A large population-based study in Scotland showed that only 32% of people with ID had no other health condition compared with 51.6% without ID and that adults with ID are more likely to have one to four physical health conditions [125]. Epilepsy (OR-31), constipation (OR-11) and visual impairments (OR-7.8) were significantly more prevalent in the ID population. Interestingly, cardiovascular-related conditions such as coronary heart disease, peripheral vascular disease, hypertension and atrial fibrillation had lower prevalence in this study. Authors also reported hearing loss, eczema, dyspepsia, thyroid disorders and Parkinson's disease to be twice as common in people with ID than non-ID population.

Central Nervous System

The commonest neurological condition in people with ID is epilepsy with a prevalence of 1 in 4 that compares with a prevalence rate of 4 to 10 per 1000 in the general population. Prevalence increases with the severity of ID [129, 130]. Epilepsy is a complex disorder that often has familial and genetic influences, as supported by twin studies [131]. This may further explain the higher prevalence of epilepsy in people with more severe ID and complexities associated with the diagnosis and treatment.

People with genetic disorders are more vulnerable to develop seizure disorders where the prevalence can vary according to the specific condition. Landau–Kleffner syndrome, Dravet syndrome, Doose syndrome and Rett syndrome are some of the genetic conditions associated with both epilepsy and ID. Certain genetic syndromes are associated with different types of seizures. Approximately 60% of people with Angelman syndrome experience multiple seizures with atonic, generalised tonic clonic, absence and complex partial seizures [132]. According to Robertson et al. [130], the pooled estimate of the prevalence of epilepsy in people with Down syndrome was lower than in the total ID population at 10–13%. This contrasts with Uppal and collaborators [133] who found a prevalence rate of epilepsy of 20%.

Epilepsy in people with ID has a poor outcome with about 66% of people with ID experiencing seizures despite using antiepileptic medications [129]. A person with epilepsy can die during or following a seizure for no obvious reason known as Sudden Unexpected Death in Epilepsy (SUDEP). Kiani and colleagues [134] report that SUDEP was the second commonest cause of death with a standardised mortality ratio of 37.6 for men and 52 for women in people with ID. Poorly controlled epilepsy is a risk factor for SUDEP, and people with ID who have epilepsy have a high mortality rate [135].

Metabolic Disorders

Inborn errors of metabolisms are a group of rare genetic conditions that can be associated with and/ or lead to ID. Van Karnebeek and Stockler [136], in a systematic literature review, identified a total of 81 treatable forms of inborn errors of metabolisms in which ID was a major feature. Early recognition, identification and treatment can allow initiation of treatments to prevent or minimise brain damage and improve health outcomes, for example, phenylketonuria. More common are the acquired disorders of metabolism. The functional systems of the body or metabolism include eating, sleeping, hunger and temperature control. Disorders of metabolism can affect many systems in the body and over time result in significant and severe effects on multiple organs.

Diabetes is a common metabolic disorder in which there is under-secretion of insulin that regulates the utilisation of sugar and fats by the body. Complications of diabetes include renal damage, cardiovascular events, cerebrovascular events and blindness. The global prevalence of diabetes has increased from 4.7% in 1980 to 8.5% in 2014 [137]. Similar increases can be expected in people with ID, but the prevalence of diabetes in this

population remains unknown [138]. There is evidence to support the assertion that people with ID are at high risk of diabetes, and the risk of diabetes in people with certain genetic syndromes such as Prader–Willi is approximately 25% for non-insulin-dependent diabetes mellitus with the mean age at onset as early as 20 years [139]. The risk of diabetes in people with Down syndrome has been debated on account of the autoimmune nature of diabetes, but the risk is considered high as evidenced in a Dutch study in children where the risk is threefold higher in children with Down syndrome compared to those without the syndrome [140].

Thyroid diseases, especially hypothyroidism, are over-represented in people with ID [141] especially in people with Down syndrome who have a high rate of thyroid disorders primarily of autoimmune origin. The lifetime prevalence of thyroid disease in people with Down syndrome is estimated at 63% with the incidence of primary congenital hypothyroidism in infants with Down syndrome approximately 28 times more than in the general population [142]. Cheung and colleagues [143] have reported a lifetime prevalence of hypocalcaemia of 80% in people with 22q11.2DS which is one of the commonest microdeletions and is typically attributable to hypoparathyroidism in this group.

Respiratory Disorders

Respiratory conditions are higher in people with ID but studies on the prevalence of asthma in people with ID have shown mixed findings. Gale and colleagues [144] reported the prevalence of asthma in people with ID is double that of the general population in the United Kingdom. Similar findings were reported from studies in Scotland [125], while others report no difference in prevalence between ID and the general population [141]. Irrespective of that, a case–control study has shown that people who died as a result of severe asthma following hospital admission were more likely to have ID [145] emphasising the importance of effective treatment of asthma in people with ID.

Even though Chronic Obstructive Pulmonary Disease (COPD) is a common respiratory condition secondary to smoking in the general population, there are no studies on the prevalence of COPD in people with ID. Recurrent chest infections, on the other hand, are commonly seen among children with severe ID [146] that increase morbidity and mortality in people with ID. There are multiple reasons for recurrent chest infections that include aspiration secondary to swallowing difficulties and gastro-oesophageal reflux, decreased cough efficacy due to expiratory muscle dysfunction and/or kyphoscoliosis, and malnutrition leading to respiratory muscle weakness. The presence of cerebral palsy further increases the risk. Almost 50% of people with Prader–Willi syndrome across different age groups reported a history of recurrent respiratory infections [139].

Obstructive Sleep Apnoea (OSA) is another common condition especially affecting certain groups of people with ID. People with Down syndrome are at a higher risk of OSA due to hypotonic upper airway muscles. Obesity itself is a risk factor that further increases the risk of OSA in people with Down syndrome and Prader–Willi syndrome.

Cardiovascular Disorders

Congenital cardiac abnormalities are more prevalent in certain genetic syndromes, especially in Down syndrome. A Swedish cohort study examining cardiovascular diseases showed that 54% of infants with Down syndrome had congenital cardiac defects compared with the risk of congenital heart disease of 1% in the general population. The most common congenital heart conditions in people with Down syndrome are atrioventricular septal defects (42%), ventriculo-septal defects (22%) and atrial septal defects (16%) [147].

Velocardiofacial syndrome or 22q11 Deletion syndrome (22q11DS) is a relatively common microdeletion syndrome, with an incidence estimated to be between 1 in 4000 and 1 in 6000 [148, 149], causing ID with various physical and mental health conditions. Cardiac abnormalities are commonly found

in this syndrome that includes interrupted aortic arch type B, persistent truncus arteriosus, tetralogy of Fallot and isolated ventricular septal defect or transposition [150].

Gastrointestinal Disorders

Dysphagia, or difficulty in swallowing, is highly prevalent in people with ID. Hypertonia or hypotonia of the swallowing muscles along with poor coordination during swallowing are considered to be among potential aetiologies. Dyspepsia and gastro-oesophageal reflux are also common [151]. Constipation is a frequent health condition in people with ID where nearly 70% of institutionalised adults with ID had constipation [152] and in children with severe ID, the prevalence rate is as high as 50%. People with ID are at a higher risk of constipation as a side effect of the various medications they are prescribed (e.g. antipsychotics), reduced mobility and poor fluid intake. Cerebral palsy and Down syndrome are strongly associated with constipation.

Cancers

It was previously thought that people with ID had a lower prevalence of cancers, but more contemporary analysis of data appears to show rates comparable to the general population [153]. There are some differences in the relative frequencies of types of cancer, for example, people with ID are at a much higher risk of gastrointestinal cancer. Rates and patterns of cancers may be changing with increased longevity. A significant challenge in the diagnosis and treatment of cancer is the variability in access to and uptake of screening programmes. Women with ID have a much lower participation rate in cervical and breast screening programmes than women without ID.

In the United Kingdom, figures from the Joint Health and Social Care Self-Assessment Framework demonstrated variability in uptake across three national screening programmes for cervical, breast and bowel cancer. For the bowel screening programme, 41.6% of people with ID were screened compared with 50.4% for the general population. Breast cancer screening was carried out on 39% of women with ID (general population 55.9%) and cervical cancer screening was the lowest with 29% compared with 69.1% [127].

Barriers to uptake of screening include the lack of routine use of accessible materials including invitations to screening, lack of reasonable adjustments to appointment systems, availability of adequate time and adaptations for restrictions in mobility. Professionals cite communication difficulties as a significant barrier and the awareness and attitudes of the professionals themselves have a significant impact. People with ID themselves may have limited knowledge and understanding of cancers and the need and benefits of screening [154].

Sensory Impairment

In a Dutch study of people with ID under the age of 50 years in an institutional setting, nearly 21% had hearing problems and 4% had visual problems. Such prevalence rates are significantly higher compared to the general population. The prevalence rate is higher with increasing severity of ID where 50% of people with severe and profound ID have visual impairments. The prevalence of hearing impairments was significantly high in people with Down syndrome and people with ID over the age of 50 [155]. Some of these conditions are treatable or steps can be taken to reduce the impairment due to sensory deficits, but they are often difficult to detect in people with severe ID.

Nutrition and Weight

Overweight is defined as a Body Mass Index (BMI) ratio of 25–29.9 and obesity BMI > 30 and are currently major health concerns for the general population. Obesity is one of the biggest risk factors for cardiovascular and cerebrovascular events. There has been a marked increase in the number of people with ID with obesity with a higher prevalence than the general population [156–158].

Multiple factors are thought to contribute to overweight and obesity in people with ID, and there is an association with certain genetic syndromes, for example, Down syndrome [158] and Prader–Willi syndrome [139].

Genetic studies have shown that genetic deletions and certain copy number variants are linked to obesity supporting the hypothesis that it is a heritable and highly heterogenous set of conditions [159]. Interestingly, the rate of obesity is lower with increasing severity of ID [158].

Many external factors lead to obesity that include a lack of healthy food choices, the side effects of medications such as antipsychotic drugs, a lack of exercise [156, 160], physical limitations and pain, little motivation and few resources and support to engage in activities. The environment in which a person lives has an impact on maintaining physical activity where people with ID living in less supervised settings are at higher risk of obesity compared to people who live in closely supervised ones [161]. Gender disparity in obesity is an interesting factor in its development. Kanter and Caballero [162] identified socio-cultural factors leading to gender disparity where in developing countries there are more obese women than men and that women with ID are at greater risk of obesity [156, 163, 164]. Such factors are larger body sizes considered as signs of healthiness and happiness alongside sex differences in metabolism and adipose tissue distribution.

- **Musculoskeletal Disorders**

People with ID experience musculoskeletal problems that cause significant functional impairment by reducing mobility and impairing ability to engage in educational and other activities which require fine and gross motor skills. Hypotonia is a common finding in Down syndrome and Prader–Willi syndrome and in people with cerebral palsy. People with ID are at a higher risk of low bone mineral density, osteoporosis and osteopenia often secondary to the use of antiepileptic medication to control seizures that reduces Vitamin D absorption. These factors along with reduced mobility predispose people to bone fractures that may also go unnoticed if they are immobile. Almost 50% of people with ID meet the criteria for screening of osteoporosis and osteopenia [165].

3.2.4.1 Prevention

Primary, secondary and tertiary prevention strategies are important in managing physical health conditions in people with ID. Examples of primary prevention interventions in childhood are screening for visual defects, hearing disorders and musculoskeletal disorders to prevent morbidity and/or to reduce the risk of impairment. Screening every person with ID for common health conditions and specific screening tests in people with congenital disorders is important, for example, testing for hypothyroidism in people with Down syndrome annually as per the guidelines from Down Syndrome Medical Interest Group. Secondary prevention is the detection and treatment of a condition prior to developing complications that involve recognising people at high risk and taking the necessary steps to test and treat. Tertiary prevention is reducing harm from a condition, for example, interventions to prevent a person with cerebral palsy from developing contractures. The biggest challenge with preventative strategies in people with ID is the paucity of research and policies and guidelines on screening and managing health conditions.

- Conditions related to central nervous system, metabolic system, respiratory system, cardiovascular system, gastrointestinal system and musculoskeletal system are commonly seen in people with ID.
- Improving physical health care of people with ID includes early identification and support to access health care.

3.2.5 Improving Health Care and Reducing Physical Vulnerabilities

To reduce health inequalities in people with ID, it is essential to recognise the obstacles in the person and in the system to them receiv-

ing appropriate care. Providing health services for people with ID can be challenging for many reasons not least the multi-morbidity affecting people with ID. In England, General Practitioners have responsibility to offer Annual Health Checks to their patients with ID. Despite various measures to increase uptake of Annual Health Checks only 50% had one [166].

One of the main challenges with improving health care for people with ID is the lack of research often due to multiple barriers such as funding, ethical issues and little interest by researchers in this area. As a result, issues relevant to diagnosis and treatment of physical health issues in people with ID are often not specifically addressed in guidelines with the potential of not identifying health problems as their presentations may be different to people in the general population. In addition, people with ID at high risk may not be recognised as "high risk" or clinicians may not be trained to examine and suspect health issues.

People with ID often express pain or distress through behavioural changes on account of poor communication skills. The behaviour can be perceived as "challenging" to carers and clinicians who may fail to understand the underlying physical cause of the behaviour. This may lead to inadequate assessment and inappropriate treatment. To overcome such difficulties, joint working between mental health clinicians and generalists is recommended. A lack of mental capacity to make decisions about physical health care can be an issue for some people with ID. Legal frameworks to support assessing mental capacity and making best interest decisions are important to support people with ID to access health care [167].

- **Summary**

People with ID are prone to physical health difficulties either directly related to the level of ID or as a consequence of treatments they receive. It is essential that clinicians working with people with ID are alert to unusual presentations, the impact of treatments, and are familiar with the health needs of this population in order to avoid greater morbidity. People with ID are a heterogenous group who experience a range of physical disorders as they grow older including long-term conditions. Skilled interventions by carers and clinicians can help to prevent the onset of physical health problems or to ameliorate their impact through education and support.

3.3 Lifespan and Transitions

3.3.1 Introduction

In the last decade, lifespan issues appear to have received increasing attention. There has been a growth in research and policies. Transition planning and supports for community inclusion, independent living and employment have become key areas of interest. Among many factors that have contributed to this development, a major role has been played by deinstitutionalisation, which created greater access to community-based services and living arrangements. Furthermore, educational advocacy and legislations have facilitated the access to educational opportunities. Provision of behavioural intervention programmes has produced dramatic increases in language, social and practical skills, with many individuals achieving skills necessary to adapt in community living. As of 2012, approximately 50,000 individuals with ID/autism spectrum disorder (ASD) per year turn 18 years old in the United States [168, 169]. Therefore, the challenges of transition to adulthood in ASD are now a significant public health issue [170, 171].

3.3.2 Adolescence

Deterioration in behavioural is seen in approximately one-third to one-fourth of adolescents with ID and ASD [172]. A major causal factor for such deterioration is postulated to be due to hormonal changes and associated sexual desires, especially in the context of limited knowledge and skills around safety and

relationship issues. In females with ID/ASD, this seems to be linked to menarche. The age of onset for menarche is often significantly earlier compared to women in the general population. Adolescence is also marked by various other changes in psychosocial development, educational and life activities, participation to community living and the related safety issues.

3.3.3 Education

Literature shows that adolescents with ID and ASD have lower levels of academic achievement in high school, and the majority of them have post-school services as part of their transition plan [173]. The data in the scientific literature indicate that 40% of individuals with ID/ASD attend college, and a smaller percentage graduate from College, with lower attendance rates among those with more severe symptoms or lack of access to services [168, 174–178]. Those young adults who are attending College or University or trying to pursue professional education may encounter various problems. Living away from home, managing one's daily life and finances, managing the unpredictable stresses of more independent living and navigating the relatively complex social environment of higher education are some of the challenges. However, there are several organisations that provide professional support staff and peer mentoring during College education. Furthermore, there are an increasing number of programmes designed to prepare and support young adults with ID/ASD prior to and during the University studies.

3.3.4 Sexuality

Sexuality encompasses more than just sexual behaviour. It includes self-image, emotions, values, attitudes, beliefs, behaviours and relationships. There are many wrongly held beliefs about ID/ASD individuals, referring to them as sexually immature and sparsely interested in romantic relationships or as asexual [179]. In contrast to these stereotypes, several studies have shown that sexuality is an important part of life for individuals with ASD and that they have general interest in solitary and dyadic sexual behaviours [180]. In fact, people with ID/ASD typically mature physically and sexually according to normal developmental stages. However, a young person with ASD can develop normally in some areas of social and emotional understanding and have difficulties in others. Difficulties in initiating and maintaining relationships appear to be the main difficulty that most people with ID/ASD experience than the lack of interest. The success or failure encountered by young people during their sexual development impacts their ability to effectively transition into adulthood.

Moreover, lack of understanding of rules of dating and romantic relationships, of social skills and also poor social decision-making in adults with ID/ASD may lead these individuals to engage in behaviours that are misinterpreted or get them into trouble, such as "stalking" behaviours [180]. This can have severe consequences, including loss of opportunities for employment, social isolation or even criminal prosecution. Moreover, individuals with ID/ASD report problematic sexual behaviours, including hypersexuality, paraphilic disorders, asexuality and gender-nonconforming feelings [181].

There has been a growing interest in this area recently. Some authors have suggested that specialised sexual education combined with social and communication skills training for adolescents may be beneficial. Such education would focus on improving knowledge so that inappropriate activity can be avoided [182, 183]. However, relatively few interventions and services have been developed and rigorously tested to improve social functioning for late adolescents and adults with ASD.

3.3.5 Adulthood

The transition to adulthood is one of the periods characterised by considerable stress and adjustment for people with ID/ASD and

their families [184]. One of the major difficulties in addressing transition to adulthood adequately is the tremendous heterogeneity of ID/ASD, including heterogeneity of underlying aetiology, clinical manifestations, levels of cognitive and adaptive functioning [185]. This heterogeneity makes essential thorough clinical evaluation and tailored transition plans, services, interventions to the goals and needs of the particular individuals. One of the major challenges is to help individuals, families and schools to plan the transition to adulthood in advance. Transitional issues are numerous and complex and often take time and planning to address these complexities [173].

Transition work should include several issues: helping to find appropriate jobs, providing support to progress in their work placement, considering options for housing, helping to gain more independent living skills and finding appropriate legal and financial counselling for longer term planning regarding financial and healthcare decision-making. Ideally, the individual, parents, teachers and other professionals should all participate in formulating the transition plan. There should be coordination between efforts of school and community agencies [173, 186].

One of the most complex transitions, which generally occur in adulthood, is represented by moving from the parental home to another setting such as independent, shared, supported or residential livings. Many skills are involved in independent living, including managing safety, cooking, cleaning, laundry, dressing and personal finances. In general, teaching and practice of these skills should be part of a transition plan to gain such skills relatively early, so that there is sufficient time for these skills to be acquired. Housing and residential issues are treated in ▶ Chap. 37.

Finding and maintaining a job is a priority during the transition to adulthood as it plays an important role in developing a sense of purpose and well-being, self-esteem, social and mental engagement, and financial independence. The several issues related to vocation and employment are treated in ▶ Chap. 38. Furthermore, young adults have substantial difficulties in meeting the increasing social subtlety of adolescent interactions relative to childhood social interactions.

Most adults with ASD tend to remain socially isolated [187, 188]. Several studies have found that a high percentage of people with ASD have no friendships or have only one friend [189–191]. Another critical transitional issue is the increased risk for periods of aggravation of behavioural symptoms (e.g. aggression, hyperactivity and insistence on sameness), seizures [192, 193] and mental health problems. The psychotropic medication usage tends to increase in individuals with ASD as they enter adolescence and adulthood [194], despite the fact that the evidence base for use of medications in adults with ASD is very limited [26, 195, 196].

The relatively poor outcomes for adults with ASD may be attributable, at least in part, to the relative lack of services for adults. This is further increased by limited research on adults with ASD. In many countries, mandated services and supports for individuals with ASD either disappear or are dramatically reduced after the age of 18–21 years [178, 191, 197]. The transition to adulthood is often a period of considerable stress and adjustment for close family members of persons with ASD\ID [184].

Main difficulties encountered by family members concern the coordination of many types of health, educational and other services. Parents often have longer term concerns about the capacity to move out of the home and to function in post-secondary education, employment and activities of daily living, what will happen to their child when the parents become older, sick or pass away. They have considerable worries about how to address legal issues about guardianship and financial and health decision-making [198]. In addition, these parents tend to experience higher levels of stress, more mental health symptoms, higher divorce rates than parents of typically developing children and higher demands for assistance [199, 200].

When necessary, the transition to adult services must be adequately prepared, not

only through the collaboration of services professionals for the two ages but also by providing young people with the necessary indications about the interventions they will need, where and from whom receive them. It is necessary for the young person and, when appropriate, the parents or caregivers to be involved in the treatment planning. Similar methods should be applied – mutatis mutandis – also to the transition, more gradual but also more subtle, from adulthood to senescence.

3.3.6 Ageing

Life expectancy of people with ID has increased over the last few decades although it is still lower than that of the general population by about 10–15 years. Increase in life expectancy has been linked with an increase in the prevalence of mental and physical ill-health disorders associated with ageing.

- There have been significant changes over the last few decades in the ways people with ID live following deinstitutionalisation and community inclusion.
- As a result, there have been changing expectations from people with ID in different aspects of life.
- Transitioning from one stage of life to another can lead to different challenges for people with ID.

Key Points

- Psychological distress is commonly seen among people with intellectual disabilities (ID)/autism spectrum disorder (ASD).
- Similarly, high rate of physical disorders are seen among people with ID/ASD associated with high mortality.
- People with ID/ASD are further vulnerable due to different stages of transition that they have to go through in their lives.

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Borderline Intellectual Functioning

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Contents

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Learning Objectives

- To understand prevalence of borderline intellectual functioning (BIF) and associated factors.
- To learn about the ascertainment and diagnosis of BIF.
- To become aware of mental health comorbidities and BIF across the lifespan.
- To understand the issues relating to service delivery and treatment provision to this group.

4.1 Introduction

Intelligence encompasses many different cognitive components that contribute to the ability for abstract thinking, self-realisation, consciousness, moral judgement and a range of adaptive behaviours such as education, employment, forming of relationships and so on. Intelligence is measured by tests that derive IQ scores which are standardised for various population groups. All testing is imperfect and subject to standard error which may be due to cultural or other norms and the construct that each test strives to measure.

In 1961, the American Association on Intellectual and Developmental Disabilities (AAIDD) (formerly American Association on Mental Retardation) suggested a categorisation of mental retardation, as intellectual developmental disorders were then known, which included borderline intellectual functioning (BIF) as the first of five levels of severity. The IQ range was given as 70–85 including people who were on the second to 16th percentile. Subsequently, this categorisation was adopted by the classification manuals of the American Psychiatric Association in DSM-II [1], DSM-III [2] and DSM-IV [3]. This classification tended to overdiagnose people from minority ethnic groups or those in lower social strata as it omitted the additional condition needed for a diagnosis, that of impaired adaptive functioning.

It was then recognised that in order to avoid the likely increase in false positives, that is, people identified as having BIF, the latter became a descriptive V-code rather than a disorder incorrectly, and the IQ upper ceiling for mild intellectual disabilities (ID) was set to 70. An unintended consequence of this change was the de-classifying of people who had reduced cognitive ability but were not deemed to have ID and therefore, were unable to receive services. As this was clearly causing difficulties, clinicians, researchers and policymakers tried to rectify the situation by (1) interpreting needs based on IQ scores flexibly, which is establishing service exemptions so individuals with certain diagnoses may receive services; and (2) raising the ceiling for ID from 70 to 75 given that this is the accepted standard error of IQ measurement. For example, an IQ score of 70 can be as high as 75 or as low as 65. For a full discussion of that debate, see Greenspan [4].

Interest in BIF is international. Wieland [5] argues that BIF is an important element in the onset, treatment and prognosis of mental disorders and the relegation of the concept to a V code "for Conditions Not Attributable to a Mental Disorder that are the Focus of Attention or Treatment" is detrimental to the quality of life of those with BIF and mental or other health comorbidities. In the International Classification of Diseases tenth edition-Clinical Modification (ICD-10-CM) [6], BIF was assigned a "residual code R41.8, other and unspecified symptoms and signs involving cognitive functions and awareness".

Salvador-Carrulla et al. [7] presents the dilemmas that dominate the current understanding of BIF and its classification as veering from the World Psychiatric Association view of BIF being a disorder (or metal condition) that should be included in the international classification of diseases (ICD) to the American Association on Intellectual and Developmental Disorders (AAIDD) view that BIF is a disability and therefore should be included in the International Classification of Functioning (ICF), Health and Disability [8, 9].

Further, Bertelli, Cooper and Salvador-Carulla [10] argue that defining conditions by IQ level is problematical because intellectual developmental disorders are multifactorial and individual abilities and skills may differ between persons at the same IQ level. They propose that assessing specific cognitive

functions may be more fruitful for ascertaining ID but also for combating the associated stigma for the sufferers.

These debates about diagnosis of BIF clearly underscore its high heterogeneity as a condition which is underpinned by both biological and psychosocial factors that increase individual risk for developing it. Consequently, the absence of consensus on what the term BIF should encompass and how it should be classified raises concern that such uncertainties will affect the information on prevalence of the condition and the provision of services to those in need.

4.2 Prevalence

Information about the prevalence of BIF is garnered from a number of sources including clinic populations, country-wide epidemiological surveys or simply estimations based on the population intelligence distributions. Therefore, published literature suggests that prevalence ranges from 13.5% to 18% [7, 11, 12]. These proportions, which all are based on IQ rather than IQ and adaptive behaviour qualification, are close to 14% which is derived by subtracting those with IQ less than 70 (second percentile) from those with an IQ around 85 (16th percentile). Prevalence seems to be relatively higher in males, persons with individual and family problems, negative parent behaviours, low socioeconomic position, and prisoners [13–19]. In the latter, the rate ranges from almost 6–32% [19].

Recent exploratory studies have shown the prevalence of BIF to likely be higher also in mental healthcare populations [12, 20, 21]. Discrepancies in the prevalence rates reported in each study are due to methods of ascertainment, sampling frame and definition/criteria for BIF including whether the additional criterion of adaptive functioning was used.

It is possible that BIF overlaps with the upper end of mild ID but on occasion also with specific learning difficulties, that is, impairment of scholastic ability in one area in the context of normal development and adaptive functioning. Further, given the known impact of severe mental illness, for example, psychosis, bipolar affective disorder, on IQ, it is not surprising that ascertainment of BIF status in adulthood in people with severe mental illness may be contentious in the absence of collateral developmental history.

> The prevalence of BIF ranges from 13.5% to 18% on the basis of the different sources. It seems to be relatively higher in males, persons with individual and family problems, negative parent behaviours, low socioeconomic position, and prisoners.

4.3 BIF and Cognition

Impairment in educational and scholastic achievement has been linked with BIF in school-age children. Underlying processes that have been implicated include information processing [22] measured with event-related potential in the absence of sensory deficits. The findings suggested that the BIF group (mean IQ 81) had deficits in attentional and information processing pathways compared with children of average intelligence mean (IQ 99). Grey matter and brain volume changes may underlie executive or motor function difficulties; they appear to be increased in certain parts of the brain (e.g. right temporal cortex) but decreased in others (e.g. right parahippocampal gyrus) in BIF young persons without other pathology (IQ 80) compared to children of average intelligence (mean IQ 118) [23]. Other studies have replicated such deficits in motor function in up to about 60% of samples drawn from schools for children with special needs (mean IQ 77) [24]. Impulse control may also be affected by BIF as shown by van der Meere et al. [25] in young boys with conduct disorder. They showed that children with BIF and conduct disorder respond faster than the control group, and that their best performances are related to the presence of valid-cue information. This propensity may be perceived as an action-oriented response style that is consistent with the sensation or thrill-seeking characteristics.

There are several genetic conditions with variable effects on IQ which may lack distinguishable characteristics and thus, it may not

be obvious that the sufferers have a number of neurodevelopmental problems that may hinder their academic progress.

The most common conditions associated with variable IQ are autism spectrum disorder [26], attention-deficit/hyperactivity disorder (ADHD) [27], foetal alcohol spectrum disorder [28], Fragile X syndrome premutation carriers [29], velocardiofacial syndrome [30], Prader–Willi syndrome [31] and Williams syndrome [32].

Children with those conditions share specific cognitive profiles but those may not be recognised in classrooms especially where the original genetic abnormality is not known or distinctive facial and other bodily markers may not be prominent and therefore missed. Common problems in the classroom may comprise disturbances in executive function, working memory, remote recall of information, declarative learning and memory, speed of information processing, and temporal sequencing, as well as visuospatial functioning.

Many children and young people with BIF with or without known genetic conditions are also called "slow learners" and may come to the attention of services at the start of primary education. Karande et al. [33] raised the issue of lack of recognition or awareness of BIF in parents of children with BIF who had been referred for assessment due to poor educational attainment.

These children often receive a statement of educational needs which entitles them to support if their difficulties are deemed serious and impede their progress. However, they may leave education without qualifications and therefore, represent a hidden problem. More often than not, those early difficulties have not been subject to remediation or early support and may continue to impact adult life outcomes in a considerable minority. ◘ Table 4.1 presents the main cognitive deficits in comorbid conditions associated with BIF.

BIF has been associated with deficits in attentional and information processing, motor function and impulse control.

4.4 BIF and Health

Analysis of data from the Longitudinal Study of Australian Children [37] showed that 23% of children with BIF were obese by age 7 and were more likely to have been exposed to socioeconomic disadvantage. These children were also rated higher by their parents for total difficulties on the Strengths and Difficulties Questionnaire which implies possible psychiatric morbidity.

Common comorbidities in children include other neurodevelopmental conditions such as ADHD, autism spectrum disorder, conduct disorder and symptoms such as inattention and impulsivity. Other potential presentations of psychiatric disorders may be problem behaviours or somatic complaints.

Van der Meere and colleagues [38] investigated the characteristics of poor impulse control in children with conduct disorder and BIF, comparing them with a control group using the alertness test [39]. They suggested that these children lack an inhibitory response. In their study of disruptive behaviour disorders, Villalobos et al. [40] analysed data from more than 1000 children and found that the presence of BIF considerably increased the amount of disruptive behaviours. Further, the presence of BIF is a poor prognostic factor for the management of children with ADHD.

A narrative review of the literature examining reported psychopathology in people with BIF suggests that the commonest mental conditions found in this population group are personality disorders, posttraumatic stress disorder (PTSD), psychosis, ADHD, bipolar disorder and sleep disorders [41].

Findings from an Israeli study of 173,542 adolescents with BIF screened for military service [42] showed that as a group they were twice as likely to be diagnosed with a psychiatric disorder using the ICD-9 [43], specifically antisocial personality disorder and non-affective psychosis and were more likely to use drugs. The study by Wieland [11] which compared a clinical adult population with and without BIF but with mental disorders with a group of adults with mild ID and

Table 4.1 Cognitive profiles of comorbid conditions associated with BIF

Generalised developmental disorders	*Neurocognitive features*
Autism spectrum disorder	Relative strengths in tasks requiring non-timed abstract reasoning and relative weaknesses in processing speed and comprehension [26]
Attention-deficit/hyperactivity disorder	Deficits in executive function and response inhibition [27]
Specific developmental disorders	
Dyslexia	Phonological deficit [34]
Dyscalculia	Deficient number module [34]
Mathematics learning disorder	Difficulties in multiplication, learning to solve math word problems and automatized memory of basic facts [35]
Non-verbal learning disorder	Poor psychomotor coordination, arithmetic skills and drawing activities. Impaired social judgement and social problem-solving [36]
Other	
Foetal alcohol spectrum disorder	Deficits in executive functioning, particularly in tasks that involve holding and manipulating information in working memory [28]
Fragile X syndrome	Impaired mental status, intelligence, executive functioning, working memory, remote recall of information, declarative learning and memory, information processing speed, and temporal sequencing, visuospatial functioning [29]
Velocardiofacial syndrome	Weaknesses in the areas of visuospatial memory and arithmetic; morphological changes in the frontal cortex [30]
Prader–Willi syndrome	Impaired executive memory, and visuospatial tasks [31]
Williams syndrome	Variable cognitive profile with participants performing better on language and face recognition tasks, compared with visuospatial and number tasks [32]

Adapted from Salvador-Carrulla et al. [7]

mental disorders found lower rates of psychosis and other severe mental disorders in the BIF group, but that the BIF group had higher rates of PTSD. Just over half of the patients with BIF were diagnosed with a personality disorder, most commonly personality disorder not otherwise specified followed by borderline personality disorder [44].

Other studies suggest that limited communication or verbal ability may lead to longer inpatient admissions and more coercive care approaches for those patients [21]. The authors examined records of current psychiatric admissions and of admissions which had taken place in the preceding 5 years to check for evidence of seclusion, other restraint and enforced medication. They found that 44% of the inpatients screened positive for BIF or mild ID and that this group was almost 3 times as likely to have had involuntary admissions currently or in the past 5 years and almost 4 times more likely to have experienced any type of coercive treatment.

Another research found that BIF is likely to remain unidentified in mental health care and substance use services [45], which may hinder necessary treatment adjustments and worse the odds of positive outcome in both the short and long terms [21, 46–48]. Several papers deriving from the Adult Psychiatric Morbidity Surveys [49] in the UK indicate that adults with BIF and mental health issues

are younger, male and of lower socioeconomic status [12]. The BIF group was identified from those responding to the survey who had all completed the National Adult Reading Test (NART) which provides an estimation of current verbal IQ which is highly correlated with overall IQ. The BIF group had higher rates of common mental disorders, substance misuse and personality disorders. Further analyses [50] showed that the BIF group is also more likely to have made suicidal acts though not intending to take their own lives and are least likely to recover from them [51]. This is in accord with findings from epidemiological studies that indicate that low IQ is a risk factor for common mental disorders especially depression [52].

The BIF group is also found to have higher odds for problem gambling [53] and are less happy than their peers without BIF [54]. A recent report on symptoms of psychotic disorders in the BIF group showed that while community-dwelling respondents have twice the rates of psychosis compared to peers without BIF, they are also more likely to report auditory hallucinations but not delusions and that depression is a contributor in the pathway to developing and expressing those symptoms [55].

The most recent Adult Psychiatric Morbidity Survey [56] confirms previous findings and shows that all disorders are increased in the BIF groups compared to their peers of average intelligence. Details are shown in Table 4.2.

One potential reason as to why rates of emotional disorders are higher in this population is the possibility of being chronically frustrated of not meeting expectations set by family, education or the wider society [57]. In adolescents with either BIF or ID, emotional–behavioural difficulties are reported to be worse than for those with neither disability nor BIF [58]. Feelings of unworthiness may be compounded by not being eligible for supports due to arbitrary service cut-offs.

Recently, the relationship between BIF and adult psychiatric morbidity has been found to be partially mediated by exposure to Adverse Childhood Experiences (ACEs) [59].

Persons with BIF show higher rates of all mental disorders, especially depressive disorders, than their peers of average intelligence.

4.5 Social and Legal Aspects of BIF

Most individuals with BIF lead fulfilling lives. However, many face a number of difficulties in lack of prospects or close relationships and may feel that they have not reached their potential. Greenspan [4] argues that BIF is seen "as the poverty disorder" given that many of the individuals labelled as having BIF may also belong to lower socioeconomic and minority ethnic groups which are already vulnerable and discriminated against.

As many individuals will not be detected as suffering from BIF even during their school years, they may end up leaving the education system without skills that will lead to unemployment and possibly being unable to live independently. Many will have supportive families and may be employed in menial jobs without reasonable adjustments in the workplace and may not even know of their rights given that there is no specific statement to mandate input by social or healthcare professionals. This is a significant gap that does not afford people with BIF the same rights as those with disabilities and therefore, may be unable to access services.

Offenders with cognitive limitations should be supported by "appropriate adults". An appropriate adult in English law is a parent, guardian or social worker; or if no person matching this is available, any responsible person over 18. The term was introduced as part of the policing reforms in the Police and Criminal Evidence Act 1984 and applies in England and Wales [60].

People with cognitive limitations are often impulsive and lack social judgement which may also contribute to their criminal behaviour. Rather worryingly, in the USA, given the outlawed application of the death penalty to individuals with intellectual disability, inmates who have committed capital offences may be diagnosed as having BIF so the death penalty could be imposed upon them.

Table 4.2 Psychiatric morbidity by IQ range in a sample of community living adults in England

Predicted verbal IQ [a]						
Mental health conditions [b]	**70**	**71–79**	**80–89**	**90–109**	**110+**	**All**
Men	%	%	%	%	%	%
Any common mental disorder (CMD)	23.5	17.4	16.9	12.7	10.5	13.2
PTSD screen positive	4.9	7.2	5.3	3.4	2.7	3.7
Probable psychotic disorder	4.5	2.4	1.3	0.8	0.5	1.0
Autism	–	5.2	0.7	0.6	1.4	1.1
Personality disorder (SAPAS)	16.0	26.5	11.8	12.2	11.2	13.2
ADHD screen positive	10.9	17.8	14.3	10.1	7.7	10.0
Bipolar disorder screen	4.3	4.9	1.1	2.1	1.8	2.1
Alcohol: AUDIT score 16+	7.1	7.5	5.5	3.3	5.0	4.4
Alcohol AUDIT score 8+	17.8	21.4	26.9	29.2	29.8	26.3
Drug dependence signs	3.0	9.4	7.0	4.3	2.2	4.3
Suicide attempt (lifetime)	10.6	6.9	8.0	5.2	4.2	5.4
Women						
Any CMD	27.9	31.4	26.2	21.1	16.2	20.7
PTSD screen positive	12.4	15.2	8.8	5.0	2.4	5.1
Probable psychotic disorder	2.1	4.0	1.3	1.1	0.5	1.1
Autism	[–]	[–]	[–]	[0.1]	[0.6]	[0.2]
Personality disorder (SAPAS)	27.9	22.0	18.6	15.6	8.6	14.0
ADHD screen positive	20.2	14.6	7.6	10.4	7.5	9.5
Bipolar disorder screen	4.7	3.3	1.7	2.2	1.4	1.8
Alcohol AUDIT score 16+	5.0	1.1	2.2	2.5	1.3	1.8
Alcohol: AUDIT score 8+	15.2	15.6	16.4	14.8	13.1	13.4
Drug dependence signs	4.3	1.8	5.6	1.8	1.0	1.9
Suicide attempt (lifetime)	8.4	15.3	12.5	8.3	5.9	8.0

Reproduced from the report, 2016 [56]
[a]Based on the National Adult Reading Test
[b]Compared with peers without cognitive impairment

4.6 Organisational Issues and Service Delivery

Research has shown that people with BIF have a range of difficulties in adaptive functioning and undoubtedly suffer from mental ill-health including a number of mental disorders.

Findings from epidemiological and clinical studies indicate that they receive more medication, have less access to psychological therapies and self-report poorer health [12]. These issues seem to stem from inadequacy of mental health services, which lack the expertise to handle these patients, fail to meet their mental health needs and consequent development of more complex psycho-physical health problems [46].

Study of BIF can support the adaptation or development of treatments to address some of those conditions, for example,

trained staff in mental health services or primary care will ensure that at least BIF status is ascertained and appropriate pathways for assessment, treatment and management are formulated. Public Health initiatives can be inclusive of this population group and ensure that reasonable adjustments are adopted by generic services to improve healthcare and help-seeking behaviour. Researchers in the Netherlands have focused on interventions for substance misuse in this population; Van Duijvenbode and collaborators [61] investigated the standardisation of pictorial materials to address cognitive biases in adults with BIF and alcohol misuse. Van Duijvenbode and colleagues [62] have also examined tests that can be used in clinical practice to aid the detection of cognitive biases in young people and adults with BIF who misuse substances; they suggest that word association tasks are better at identifying high-risk conditions under which alcohol/other substances maybe used.

Education authorities also have a role to play in ensuring that children who are poor academic performers are referred for further assessment and are given the tools to manage their limitations with additional supports. Teachers can also work with families of BIF children to equip parents to support them effectively and seek the right support for the child.

A BIF consensus group from Catalonia, Spain, has posed a number of objectives in improving the lives of people with BIF [7]. This initiative was followed by the creation of an international group of experts (The Borderline Intellectual Functioning Consensus Group) including members of the World Psychiatric Association Section on Intellectual Disability and the Fogarty/NIMH NCD-LIFESPAN Programme, which produced a declaration, named "Girona declaration", with calls for action to promote policies and practices for improving health and quality of life of people with this condition [63].

Those include improvements in early detection based on awareness of warning signs and using appropriate psychometric instruments as well as measures of adaptive functioning in assessment. This process should be instigated as early as possible and no later than the 6 years of life to ensure that early intervention is implemented. Confirming that the individual has a diagnosis of BIF will enable him/her to seek help and be made aware of any rights under disability legislation. Good practice dictates that individualised intervention plans are developed early on with the input from the individual and his/her family carers. The plan should detail any health monitoring required and other provisions to be made regarding periods of transition, reaching adulthood and finding suitable employment.

Box 4.1 Types of Instruments Used in Assessments of Individuals Suspected as Having BIF

Screening assessment with KBIT-2 (Kaufman Brief Intelligence Test-second edition [65]; quick to carry out in routine care; children).

Full psychometric assessment (any established measure of cognitive functioning standardised for the population, e.g. WAIS - Wechsler Adult Intelligence Scale [66]; WISH - Wechsler Intelligence Scale for Children) [67].

Functional assessment and adaptive behaviour (any measure that can provide standardised assessment of abilities and needs, e.g. the Camberwell Assessment of Need for Adults with Developmental and Intellectual Disabilities; CANDID) [68].

Mental ill-health (any validated scales of psychopathology, e.g. Developmental Behaviour Checklist (DBC) [69]-children; Psychiatric Assessment Schedule for Adults with Developmental Disabilities (PAS-ADD)-checklist or mini versions) [70].

Adapted from Recommendations for caring for people with borderline intellectual functioning, Generalitat de Catalunya [64].

As has been mentioned already in the chapter, many of the individuals with BIF face service gaps as well as being turned down by services as they are often found to not be eligible for specialist ID community services but also find generic services difficult to navigate.

Patients with BIF are reported to be underserved by mental health programmes, which are unable to fulfil their mental health needs, leading to the emergence of more complex psycho-physical health issues.

4.7 Concluding Considerations

BIF as a diagnostic entity is highly contested, and current classification approaches are unsatisfactory as they tend to be too narrow in being based on IQ in the main without taking into consideration adaptive functioning. Individuals with BIF across the lifespan have a number of neurocognitive deficits [71] and are likely to also suffer from significant physical and mental health comorbidities for which they receive little targeted support. The mediating risks for further mental ill-health span the environmental and genetic spectrum, which could be the focus of novel research to better understand the biological substrate of intelligence and adaptive functioning but also how education and public (mental) health policies can support disadvantaged communities and children born within those households. BIF also raises important ethical dilemmas in the practice of – dare we say – medicine where forensic aspects are evident, and therefore it merits an overhaul of current conceptual frameworks given that individual lives are at stake.

Tip

The definition of BIF as well as its position within classification systems should be given further thought. A well-defined classification will help patients with BIF be identified more easily and gain access to mental health services, bridging the current gap between high prevalence and low recognition.

Future research and practice should focus more on the mediating risk of BIF for mental ill-health, especially in terms of environmental and genetic factors.

Special attention should also be given to the way through which education, social and mental health policies can adequately support persons with BIF.

Key Points

- There are significant issues in the definition of borderline intellectual functioning which may add to the difficulty of patients accessing support.
- There is little awareness of the complex needs of people with BIF across the lifespan.
- People with BIF have several mental health comorbidities.
- Individuals often fall through services due to artificial constructs about upper IQ limits denoting eligibility for services.

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Diagnostic Issues

Marco O. Bertelli and Steve Moss

Contents

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Learning Objectives

The primary objective of this chapter is to provide an overview of the peculiarities of presentation of psychopathological symptoms and syndromes in persons with intellectual disability (ID) and/or low-functioning autism spectrum disorder (LF-ASD) as well as the factors that impact on the diagnostic process. Adaptations of ICD and DSM diagnostic criteria to ID/LF-ASD are described. The chapter also provides indication on how to carry out a psychopathological evaluation in an appropriate setting.

5.1 Introduction

Why do we want to conduct mental health assessments? This might seem an obvious question, but in fact the answer is not at all straightforward. In clinical terms, we presumably wish to conduct an assessment in order to help someone, but this is a broad concept which could include multiple different approaches. Making a precise diagnosis can suggest we are getting closer to understanding the person's problems, but this may not necessarily be the case. For instance, there are probably at least 200 different psychiatric diagnoses, but only a small number of possible treatments.

Consider another question: why do people get mentally ill? We can identify at least four fundamental possible reasons:

- The person's biological/physical status.
- Incongruence between the person's hopes, dreams and aspirations, and the realities of their life.
- Psychodynamic issues: bonding, attachment and the complex development of the person's internal world that determines their interactions and relationships for the rest of their life.
- The environments and ecologies in which they live.

Each of these fundamental causes has particular issues for people with ID. Most people with ID have physical issues of one kind or another, including genetic problems, brain damage, hormone imbalances, etc.

The incongruence between a person's hopes and aspirations and the reality of their lives is probably as great in people with ID as any other sector of the population. One need only reflects on the number of people with ID who have seen their brothers and sisters grow up, get boyfriends and girlfriends, good jobs and become independent. Compare that with the lives of most people with ID.

In psychodynamic terms, the development of an individual is an immensely complex process. The relationship between a child and its parents starts even before the child is born; in the later stages of pregnancy, the foetus can hear what is going on, as well as having the other sensory modalities operating. Once the child has been born, the process of bonding is crucial to successful development. Beyond this, the child develops a mental world that is partly a response to do things they experience. Throughout this whole process, it is clear that the presence of ID will have a fundamental impact.

The ecologies of people with ID are typically very limited compared with members of the general population. Most of us have got a wide range of ecological supports that protect us against mental illness. These typically include parents who care for us, partners, friends, meaningful occupation that gives us a sense of self-esteem, and good health. Crucially, our ecologies include a wide range of problem-solving skills that enable us to seek help, and also to adapt our lives to fit our capabilities. Compare this to the ecology of a typical person with ID, who probably has a very small social network, very few opportunities for self-esteem and autonomy and possibly poor physical health. A range of problem-solving skills is typically very impoverished.

5.2 Developing Assessment Protocols

What do these four fundamental dimensions tell us about mental health assessment? Firstly, it will never be possible to find physical

reasons for every mental health problem, simply because they are not entirely generated by physical reasons. Secondly, we need to think carefully about our models of mental health assessment, and what we would like them to achieve. First of all, what should be the focus of our assessment? It is worth noting that we typically have very different emphasis on the assessment of children compared with that of adults. If the patient is a child, it is very often the case that they have been referred because of management problems. One of the first questions that is often asked is whether there are problems in the family. In other words, the context within which the child lives is considered to be a primary focus for evaluation. By the time a person has reached adulthood, it is more likely that a practitioner will say 'he has got agoraphobia or 'she has got a psychotic disorder'. In other words, the perceived locus of the problem has moved from the ecology to some kind of internal pathological state. Logically, there seems no reason for making this shift. Rather it would seem appropriate to view all mental health disorders as probably having contributions from multiple dimensions. Sometimes, the influence of the environment, for example, problems of housing, income, physical health and social disadvantage, can outweigh the impact of any physical causes.

What this means in practical terms is that we need always to assess these four fundamental dimensions whenever considering the mental health needs of an individual. We often talk about multi-disciplinary assessment, but this may actually mean that different professionals continue to work within their own perspectives, rather than coming together around a common framework. It is the latter approach that hold out greater promise for understanding people's needs.

5.3 Differences from the General Population

In persons with ID, especially in the lower part of the severity range and/or LF-ASD the presentation of psychopathological symptoms and syndromes can considerably vary from that of the general population for a number of reasons, including cognitive and communicative impairments, developmental peculiarities and neuroautonomic vulnerability. Also many factors related to carers' and clinicians' consideration of the psychic suffering of these persons can impact on the definition and the identification of psychiatric disorders (PD).

5.4 Cultural Factors

Many clinicians believe that the structural and functional alterations of the central nervous system, detected or hypothesized in many forms of ID and LF-ASD, do not allow the development of a psychic suffering similar to that described by individuals of the general population and upon which the current psychopathological semeiology has been developed. There is, however, no current evidence to support this. Other clinicians and researchers argue that a further problem in the conceptualization of PD in people with ID and LF-ASD consists of the difficulty of ascertaining the impairment of functioning and the degree of subjective distress that they determine, which are fundamental for defining a psychiatric disorder, since in their absence no set of symptoms would have clinical significance. In ID and LF-ASD, these aspects are often already present at a high level for the developmental condition and may impede to identify any increase due to co-occurrent PD. In particular, subjective distress is often higher than in the general population due to the several psychological and relational factors, such as reduced opportunities for participation in community life, greater exposure to traumatic experiences or lower coping skills [1, 2].

This difficulty of distinguishing those manifestations that are a function of the ID or LF-ASD, and those related to additional PD, has been called 'diagnostic over-shadowing' [1] and refers not only to the impairment of functioning and to the subjective distress but also to psychopathological symptoms and the way they cluster into syndromes. A part of such a shadowing seems to be linked to

the above-mentioned clinicians' viewpoint, that severe neurodevelopmental disorders do not allow any further psychic suffering, which depends in turn on the level of specific knowledge and experience in the field. The two pivotal experiments in this regard are those of Reiss and collaborators, in which they showed that groups of randomly selected clinicians tended to formulate fewer psychiatric diagnoses in persons who were known to have an ID compared to those who were known to have IQ within the normal range [3]. Psychiatrists who have not received specific training in ID and LF-ASD also tend to make more diagnosis of psychotic disorder and less diagnosis of mood disorders or anxiety disorders compared to colleagues with higher specific knowledge [4–6].

5.5 Factors Linked to Cognitive and Developmental Peculiarities

In people with ID and/or LF-ASD, the psychiatric symptomatology can be strongly altered by the cognitive, communicative and behavioural characteristics resulting from these conditions, which can make it difficult to disentangle ID from psychiatric problems [7–9]. Sometimes even the nuclear elements of some syndromes, such as anhedonia or delirium, are not identifiable [10], especially in people with pre-verbal communication. Symptoms related to subjective experience are expressed as alterations in basic behaviours or behaviours usually associated with individual well-being [1, 11, 12]. These alterations can be both qualitative and quantitative; for the latter, the expression 'baseline exaggeration' has been proposed [1]. Behavioural problems that already exist at a low rate and low intensity may increase dramatically when a person has stress or mental ill-health.

Other factors that increase the difficulty in the diagnostic process are 'developmental inappropriateness', that is, the mismatch between the level of individual development expected for the chronological age and the level of actual individual development [13]; 'intellectual distortion', which refers to the conditioning that cognitive difficulties may have in understanding and communicating one's own experiences [1, 11, 14, 15]; and 'cognitive disintegration' related to excessively concrete or immature way of coping with emotional stress, as well as cognitive impediment to recognize one's own emotional states [1]. Even the peculiarities in interpersonal, cultural and environmental influences can have a significant impact on the presentation of symptoms, which is called 'psychosocial masking' [1].

Also, neurovegetative vulnerability is common among persons with ID and LF-ASD, it consists of a strong tendency to manifest psychic suffering with dystonias of the autonomic nervous system, somatic symptoms, organic dysfunctions or changes in circadian rhythms [11].

In addition to the aspects treated so far, the identification of psychopathological symptoms is also complicated by problems in communication. As mentioned above, people with ID and/or LF-ASD often have poor verbal expression skills, severe difficulties in understanding what they are experiencing, and a lack of problem-solving skills that could enable them to seek help. Thus, the possibility that they may describe their psychological distress and ask for the necessary treatment is very low.

Even those who are verbally competent and can express their own thoughts and emotions, such as those with mild ID or borderline intellectual functioning, may present many limitations in referring psychopathological symptoms to a clinician. In fact, they can be susceptible, suggestible, prone to acquiescence, they can have a cloak of competence or difficulty in describing lived experience due to attention deficits or space-time disorientation (see also paragraph 5.5) (■ Table 5.1).

In persons with ID and/or LF-ASD, the presentation of psychopathological symptoms and syndromes can considerably vary from that of the general population, for a number of reasons, such as cultural factors, developmental peculiarities and communicative impairments.

Table 5.1 Factors affecting the presentation of psychopathological symptoms in persons with ID and/or LF-ASD

Intellectual distortion	Level of cognitive, communicative, physical and social functioning
Cloak of competence	Problems with self-acceptance
Acquiescence	Problems with being interviewed and receiving questions
Developmental inappropriateness	Individual developmental level
Psychosocial masking	Interpersonal, cultural and environmental influences
Diagnostic overshadowing	Distinction between psychopathological symptoms and expressions of the neurodevelopmental disorder
Atypical or masked presentation	Aggressive behaviour, screams, maladaptive behaviours, etc.
Neurovegetative vulnerability	Somatic symptoms, changes in circadian rhythm and dystonias of the autonomic nervous system
Cognitive disintegration	Coping impairment, low stress threshold and difficulties in recognizing one's own emotional states

5.6 Behavioural Equivalence of Psychopathological Symptoms

In people with ID and/or LF-ASD, problem behaviours (PB) represent one of the factors with the greatest negative impact on the implementation of rehabilitative interventions, on the management of services' costs and on the quality of life of the disabled person and his family [16]. The term PB refer to culturally abnormal behaviours of intensity, frequency and duration such as to put at risk the safety of the person acting on them or others, or behaviour that severely limits or prevents access to the community's ordinary services. The most frequent PB are physical or verbal aggression towards self, other persons or objects, tantrum, escape, oppositional, provocative or impulsive behaviour and screams.

The data in the scientific literature indicate that PB are very frequent in people with ID and/or LF-ASD, even if findings of different studies are extremely variable due to relevant limits and methodological differences, such as inclusion and exclusion criteria, sample characteristics, time interval and evaluation procedures. For example, the reported prevalence of self-injurious behaviour ranges from 1.7% [17] to 41.2% [18], and that of aggression towards others from 7% [19] to 51.8% [20]. Some of the largest studies in this area found that about 25% of people with ID have at least one problem behaviour, 5% show self-injury and 10% show physical outwardly directed aggression, the latter further divided into aggression towards people (6.5%) and objects (3.5%). The verbal aggression is instead acted by 7.5% [21–23]. When considered as a psychopathological category in persons with severe and profound ID, PB account for the 75% of all the diagnoses [24].

The persistence over time of PB also varies consistently in different epidemiological studies. For self-injurious behaviour, in particular, the range of remission rate is very wide, between 3.7% and 96% [25, 26]. However, the surveys conducted at precise time intervals agree on very low remission rates: 79% of PB in general and 65% of stereotypies are still present after 10 years from the onset [25], self-injury disappears only in 38% of cases after 2 years from onset [20] and in 29% after 8 years [17] and outwardly directed aggressive behaviour disappears after 2 years only in 27% [23] and in 30% after 11 years [27]. For many individuals, PB onset in childhood and persist until middle age [28].

It is often difficult to determine whether PB are expression of physical pain, organic problems, drugs, psychological, environmental, socio-relational factors or whether they represent psychiatric symptoms, or even a combination of some of these aspects. The same behaviour can be interpreted in a very different way by the various professionals, even within the same multidisciplinary team,

with consequences that are sometimes relevant for the interventions.

Some PB are very frequent in certain genetic syndromes, such as self-mutilation in Lesch-Nyhan syndrome, hyperphagia in Prader-Willy, introduction of objects into the body orifices in Smith-Magenis, hand rubbing in Rett syndrome or hyperactivity in Fragile X. In these cases, PB are called behavioural phenotypes [29].

Numerous studies have shown a relationship between PB and PD [30–35], especially in persons with low functioning and adaptive skills [31]. Some PB have been identified as symptoms, or groups of symptoms, specific to some PD, taking the name of 'behavioural equivalents' (BE); they are characteristic for onset, development, maintenance and extinction, especially with respect to other concurrent possible symptoms of a PD [36–38]. Other studies have revealed no evidence regarding the possibility that PB can be considered as BE of an underlying psychiatric disorder [39, 40], determining a more aporetic position in a part of the scientific community, for which the PB would be an expression of non-specific emotional stress (see paragraph 5.4).

One of the most recent research in this field focused on the relationship between PB and symptoms of depression, as codified in the DC-LD (Diagnostic Criteria for Learning Disability) [41], and used statistical methods significantly superior to all those used in previous studies. Here, PB were found to be associated more with a dimension of general emotional dysregulation than with a dimension of depression [42]. An in-depth analysis of this research shows that its main value does not concern the level of correspondence between behaviours and symptoms, but the way through which clinical data have to be detected and interpreted in people with ID and/or LF-ASD. A first indication of great utility is the need for specific adaptations for each individual based on the various characteristics, cognitive, communicative, sensorial, adaptive and physical; a second indication is that a dimensional symptomatological model can have a higher predictive validity than a categorical one; a third indication confirms what has already been mentioned about the importance of performing a careful evaluation of the onset, course and above all the co-presence of other symptoms, for every possible BE.

Some behaviours, especially problem behaviours, have been identified as symptoms specific to some psychiatric disorders (PD), taking the name of 'behavioural equivalents' (BE). These BE are characteristic for onset, development, maintenance, extinction and relationship with other symptoms.

5.7 Presentation of Symptoms and Diagnostic Criteria

The complexity and difficulty of the psychopathological evaluation in people with ID and/or LF-ASD also depend on the impossibility of using the diagnostic criteria that define the different psychiatric syndromes in the general population, such as those in the Diagnostic and Statistic Manual of Mental Disorders (DSM) [43] or in the International Classification of Diseases (ICD) [38, 44, 45]. Some authors have suggested that these criteria can be adopted only in case of very mild ID [46], while their validity falls for more severe forms [1, 47]. The reason for this inapplicability is their prevalent reference to the patient's reports, which pre-supposes good communication skills and, albeit to a lesser extent, awareness of intrapsychic experiences. Psychiatric semeiology has always been concerned with the internal mechanisms and the way through which they are expressed, spontaneously or solicited.

As already mentioned, people with ID and LF-ASD often have great difficulties in communication and conceptualization, sometimes they are incapable. Therefore, the evaluation of their psychopathological suffering must be based on the direct observation of behaviours in the context of habitual life as well as on the descriptions made by third parties. Obviously, among the latter, the most relevant are those provided by family members and other persons more closely connected with the person under evaluation.

In addition to not being designed to refer primarily to behaviours and other observable aspects, the diagnostic criteria for the

general population present other important characteristics that make them difficult to use with people with ID/LF-ASD, such as the specifications of quality and duration of symptoms [48].

Some researchers have found that, in respect to schizophrenia, the only first rank symptom which can be detected with a good frequency is auditory hallucination, even in persons with more verbal skills [49]. Symptoms of mania or hypomania are more difficult to detect than depressive symptoms, and in some cases, the latter can mask the former, such as in the presence of irritability [50].

To solve these criticalities, some adaptations of the standard diagnostic criteria have been proposed, for both the DSM system and the ICD. The first attempt dates back to the late 1970s and concerns the Research Diagnostic Criteria (RDC) [51] in the area of schizophrenic spectrum disorders [52], but the most consistent implementations were made in the last 25 years. In the 1990s, an adaptation of the tenth version of the Diagnostic Criteria for Research of the World Health Organization was produced [53] (DCR-10 Modified), with recommendations for clinicians more detailed than those of the RDC but still limited to some major disturbances. The multi-axial approach of these criteria allowed to confirm the communicative and interventional importance of an organized collection of different types of information: acute PD, neurodevelopmental and personality disorders, physical disorders, psychosocial problems, environmental problems and global functioning. The most consistent adaptations for ID were implemented in the criteria for schizophrenia, other psychotic disorders and mood disorders [53, 54]. Interesting data on the presentation of bipolar disorder were produced by Sovner [55], Cain and collaborators [50], Szymanski and King [56], who found a high frequency of BE, such as aggression, self-injurious behaviour, provocation and intrusiveness.

In this period it was also found that psychiatric symptoms could present differently in the various levels of severity of ID. Using a checklist of 30 ICD-10 [45] symptoms associated with depressive disorder (Clinical Behavior Checklist for Persons with Intellectual Disabilities; CBCPID [57]), Marston and collaborators found that the only symptoms common to all people with ID likely to suffer from a depressive disorder were depressed mood and sleep disturbances, all the other symptoms appeared in an almost specific way in the different levels of severity [55, 57] (Table 5.2).

In 1999, the American Academy of Child and Adolescent Psychiatry produced a list of parameters for the adaptation of psychiatric diagnoses for people with ID [56].

As early as the mid-1990s, the scientific community of the field realized that the adaptations produced up to that time were too many and too different from one another to be able to advance knowledge on the presentation and the epidemiology of PD. In fact, the results of the various studies were strongly discordant and the criteria used to obtain them were difficult to compare. Thus, the creation of standard international ICD adaptations was encouraged. The first attempt in this direction was represented by the diagnostic

Table 5.2 Most common depressive symptoms in the different severity levels of ID

ID level	ICD-10 symptoms of depressive disorder
Mild	Crying frequently for no apparent reason
	Significant diurnal mood variation
	Lack of energy
	Loss of interest
	Low self-esteem
Moderate	Social withdrawal
	Self-injurious behaviour (medium intensity and severity)
	Weight loss
Severe and profound	Tendency to scream
	Aggression
	Self-injurious behaviour (high intensity and severity)

Adapted from Marston et al. [57, 55]

criteria Kettering/Leicester [58], for which a group of UK psychiatrists combined their extensive clinical experience with data from all the literature produced up to that point. These criteria showed a good face validity, that is, they were appropriate to the situation, the population and the subject of the investigation and were presented to the Royal College of Psychiatrists as a starting point for the production of that complete and articulated system that would come out 5 years later with the name of Diagnostic Criteria for Psychiatric Disorders for use with Adults with Learning Disabilities [41] (DC-LD).

The DC-LD included different adaptations for the different degrees of severity of ID and used a hierarchical approach in the interpretation of symptoms, so as to support the clinician in the semeiological evaluation of PB, with clear instructions on organic disorders and behavioural phenotypes. Some items were formulated so as to give space to the pathoplastic effect of most severe ID and many symptoms based on the patient's reports, and introspective skills were replaced with observable aspects [59]. Guilt feelings, feelings of unworthiness, loss of anticipation of future positive events or suicidal thoughts were removed in favour of the onset or the increase in maladaptive, self-injurious, withdrawn or oppositional behaviours and physical symptoms. Anhedonia, or the loss of interest and pleasure for most of the activities of daily life, was expressed as a clear reduction in participation in activities, personal skills, collaboration in self-care or an increase in phobic behaviour or seeking reassurance.

A few years before the release of the DC-LD, the Americans of the NADD (National Association for the Dually Diagnosed) have started a process of adaptation of the DSM-IV-TR [60] criteria, with the contribution of an international advisory board. The work was completed in 2007 and was called Diagnostic Manual – Intellectual Disability (DM-ID) [61]. The main authors called it a textbook for the diagnosis of mental disorders in people with ID. In fact, it included a wide introduction on the subject, a detailed description of all the main disorders of DSM-IV-TR and the peculiarities in persons with ID, a summary of the standard criteria and a detailed list of their adaptation to the various degrees of ID. Everything was supported by a careful review of the literature, with an indication of the level of evidence for each conclusion.

The DM-ID also offered easy-to-read tables and clear examples of how the criteria have to be interpreted.

Also, this manual, like the DC-LD, emphasized the need for change compared to what was normally followed in psychiatric semiotics for the general population. A particular recommendation concerned the psychopathological interpretation of the onset or intensification of PB such as aggression, self-injurious behaviour, destructive or severely disturbing behaviours, stereotypies and ritualistic behaviour.

The DM-ID was designed in accordance with an evolutionary perspective that is helping clinicians to identify PD in all ages of life in children as in elderlies, specifying each time the main peculiarities in the presentation of symptoms.

The DM-ID has already been adapted also to DSM-5 [43], with the name of DM-ID 2 [62], again with the participation of an international panel of experts in the field. This latest edition has further detailed the number of possible BE and observable symptoms, for the most severe degrees of ID and LF-ASD, both to improve diagnostic sensitivity and to develop research activity and discussion throughout the international scientific community. However, confirmations of validity and reliability from field trials are limited [63] and many of the symptom adaptations have been proposed as interpretative notes and not as definitive criteria.

The validity of diagnostic criteria refers to their ability to effectively define a given clinical condition, while reliability only indicates the degree to which different operators are able, using them, to formulate the same diagnosis. Validity does not imply reliability and vice versa. It is relatively easy to design a diagnostic system with high reliability, but it is much more difficult to give validity to the proposed diagnoses, especially with respect to ID and ASD, which represent groups of many different syndromes, each of them with

a probable specific way to experience mental suffering and to express it.

The first version of the DM-ID was subjected to a single field study involving 63 clinicians from 11 different countries and around 850 patients. The opinions of the clinicians were positive: 67.9% of those who completed the study indicated that the system was 'easy' or 'very easy' to use, 83.1% that it had allowed to achieve an appropriate diagnosis and 36.5% that this diagnosis was much more valid and precise than that which would have been obtained using DSM-IV-TR [60].

Compared to the previous systems, the DM-ID-2 [62] has substantially increased the reliability of psychiatric diagnoses in people with ID and LF-ASD, while its validity is only partially increased.

Some adaptations to ID/LF-ASD of the diagnostic criteria for the general population have been proposed for both the DSM and the ICD systems. The first attempt, called Research Diagnostic Criteria, dates back to the late 1970s, but the most comprehensive, the DM-ID-2, came out on 2016 with reference to the DSM-5.

5.8 Assessor, Informant and Setting

As mentioned earlier, the psychopathological evaluation of the person with ID and/or LF-ASD implies particular attention, adaptations and abilities. The most important ones related to the evaluation, the information sources and the setting are discussed here.

Assessor

The main difficulty that the assessor may encounter is the obtaining of information directly from the person with ID and/or ASD, who, as already described, may have limitations in their ability to provide information, as well as in understanding and communicating their own suffering. There are persons who neither speak nor understand verbal language; others who would be able to speak but refuse to do it with a stranger and do not do anything more than answering the questions with laconism, sometimes only 'yes' or 'no'; others who need time to answer the questions, sometimes quite a lot of time; others who hide the answers in a stream of stereotyped sentences; others who do not want to be touched or even get close; others who do not interact but are open to any type of manipulation; others who do not accept to maintain a position suitable for the interview; others who want to touch all the objects in the room; others who do not tolerate sensory stimuli that you barely perceive; and others who instead have serious hearing or visual problems.

The difficulties faced by clinicians when interviewing people with ID frequently leads either to a rejection of the case, or an almost total reliance on information from third parties. This can lead to major errors in the evaluation and diagnostic processes (Table 5.3).

It is very important to maintain a welcoming and reassuring attitude, especially in the early stages of the evaluation session. Too formal attitudes should be avoided. People with ASD stay calm and are more willing to cooperate when they are informed precisely about the length of the interview and about the distances (in meters or number of steps) between the room in which they are located and the

Table 5.3 Communication skills in persons with ID

Communication skill		ID level
Preverbal	The person has no cognitive ability to understand words. They can be helped to understand through combinations or sequences of stimuli, tone of voice, objects and images	Severe to profound
Nonverbal	The person has the ability to understand the words but not to use them to express themselves. It uses alternative communication methods, such as gestures or images	Moderate to severe
Verbal	The person has a more or less wide range of comprehension and word-use skills	Mild to moderate

stairs, the elevator, the exit of the building, their car or the bus stop, up to their home. Even the repetition of information often has an anxiolytic effect, especially for people with greater cognitive difficulties; in fact, they often search for it or induce it by asking the same questions for quite a lot of time.

The language has to be simple and much effort has to be made to avoid abstractions, inferences (such as 'what would you do if ...?'), ambiguous expressions, jargon and discrepancies between what is said and the tone of voice, as well as mimicry and gestures.

The assessor should give many examples. A great advantage could also be drawn from the use of expressions known to the person with ID/ASD and a style of making questions that implies the possibility to provide many possible answers.

Box 5.1 Common Causes of Behavioural Alterations in Persons with LF-ASD

- Confusion or fear caused by an extraordinary event or an unfamiliar situation
- Interference with daily routines or repetitive activities
- Difficulty or inability to understand explanations or instructions
- Lack of knowledge on how to behave to be appropriate
- Difficulty or inability to communicate needs and emotions
- Hypersensitivity
- Fear of specific situations or objects
- Pressure to perform tasks that are too difficult

For people with more pronounced linguistic difficulties, the use of images and symbols is quite useful. Also mimicry can help, but be sure to use gestures and actions that the person knows the meaning of.

In detecting the presence of symptoms through the interview, it is useful to consider that some persons with ID attempt to hide their disabilities and adopt a 'cloak of competence' [64], tend to be less forthcoming with respect to self-descriptions, and try to please the interviewer by answering falsely or in a manner that is inaccurate ('acquiescence bias'). Thus, too direct questions can easily condition the answer.

Communicating appropriately represents not only an advantage for the evaluation purposes and for any future therapeutic alliance but also a deontological duty and a legal obligation, aligned with the rights of the person with disability to receive medical acts appropriate to their

Box 5.2 Suggestions for Appropriate Communication

Verbal language

- Active listening
- Check if you understood correctly through frequent summaries of what you heard
- Use easy, straightforward language and short sentences
- Avoid jargon
- Avoid abstractions and complex concepts
- Avoid complex sentence constructions such as conditional tenses
- Articulate clearly with the right tone of voice
- Repeat and rephrase questions
- Check if the person understands
- Use open questions as far as possible

Non-verbal communication

- Look at the person you are communicating with (not your PC monitor or case notes)
- Be smiling and reassuring
- Use intonation, gesture, body language and posture to support the person in their expression
- Use pictorial and symbolized material (e.g. books beyond words series)
- Some people may use signs (individual or standard)
- If necessary, ask for high-tech communication devices, and specialist speech and language therapy support

condition and respectful of their dignity and autonomy (see art. 25 of the UN Convention on the Rights of Persons with Disabilities) [65].
There are some tools that can help the assessor to know the ways in which a non-verbal person with ID/LF-ASD usually expresses distress in response to different factors. One of these is the Disability Distress Assessment Tool (DisDAT) [66].

Being able to guide the informant to reflect on the symptomatology and on the occurrence of behavioural alterations is an operation that requires in-depth training, knowledge of interview techniques and familiarity with the compilation of assessment tools, as well as good interpersonal skills.

It is also necessary to consider the possible discordance between different evaluators, particularly frequent for professionals with different background, who did not receive specific training on mental health of neurodevelopmental disorders or on the use of psychometric tools.

Informant

Family members or other carers represent a valuable source of information in the diagnostic process, especially when the person with ID and/or ASD has serious communication limits or lack of language [67]. Parents and other close relatives can allow to understand how the person developed through their childhood, the adverse experiences he/she encountered, and the achievements and relationships that can have influenced most their current psychological set (see Box 5.3 on usefulness of developmental history). Sometimes, however, emotional involvement and negative attitudes towards assessment services, prejudices or lack of knowledge about co-occurrence of PD can limit the supply of help or even constitute a further obstacle to diagnosis [68, 69]. The false belief that all people with ID and/or ASD have unchangeable abnormal behaviours is widespread [7, 70], as well as the tendency to underestimate or deny the presence of psychiatric symptoms and additional mental health problems.

Healthcare assistants and other professional operators are often subjected to a rapid turnover and, therefore, have a limited time frame of knowledge of the behavioural repertories and relational attitudes of the person with ID/ASD. Thus, they can give wrong information or even be unable to report significant behavioural changes.

The need to obtain accurate information with respect to the entire lifespan requires in some cases the involvement of multiple sources of information.

The reliability of the assessments made through interviews with informants also varies according to the cultural level, the personological characteristics, the nature of the relationship with the person with ID, the level of emotional involvement and effective knowledge of the person under evaluation [71].

In order to maximize the validity of the data collected during the psychopathological evaluation, it is therefore advisable to involve various professionals as evaluators, as well as the participation of different informants. Multi-perspective interdisciplinary assessment, supported by an adequate psychodiagnostic tool, is particularly effective in identifying

Box 5.3 Usefulness of Developmental His tory

- Enables appropriate interpretation of complex epiphenomena with possible psychopathological nature
- Favours the understanding of the pathoplastic effect of ID/LF-ASD on psychopathological features
- Supports (through the identification of usual pattern of behaviours) the distinction between symptoms of mental ill-health from long-standing traits and personological characteristics
- In persons with ID, allows a differential/adjunctive diagnosis for other neurodevelopmental disorders, such as ASD or attention deficit hyperactivity disorder (ADHD)
- Reduces the likelihood of diagnostic overshadowing
- May contribute to understand the aetiology of PB (some behaviours are associated with a specific developmental phase)
- Plans of treatment/interventions/supports need to be designed in a way that is developmentally appropriate

qualitative or quantitative changes in basic behaviours and in determining their character of symptomatological equivalence [72].

Setting

The context of psychopathological evaluation also requires numerous special attention when dealing with a person with ID and/or LF-ASD. First of all, the maximum flexibility should be observed, both in the place, duration and modalities. People with ID and/or ASD are very sensitive to changes and may be frightened by a context that does not include anything familiar.

A standard room of an outpatient clinic can elicit emotional and behavioural alterations, making it very difficult to carry out the psychopathological assessment and increasing the risk of misdiagnosis. It is preferable that the evaluation is performed in one of the places usually attended by the person. If this is not possible, a cosy, silent room with limited sensory stimuli should be preferred. Computer monitors and other screens should be standby or turned off, the room lighting should be soft and the phone silenced, especially in front of persons with probable sense-perception alterations.

Environments that are too 'medicalized' can arouse fear or discomfort, so even when it is unavoidable to carry out the interview in a health clinic, it is better to free it as much as possible from equipment and technical instruments (e.g. syringes, needles, sphygmomanometers and phonendoscopes). For the same reason, it is advisable to avoid wearing a white coat or other typical medical clothing.

> Appropriate setting adaptation represents not only a clinical advantage but also a deontological duty and a legal obligation, aligned with the rights of the person with disability. This includes adequate consideration of family members.

5.9 Focusing on the Individual

Diagnosis and Need

Overall, we need to think carefully about why we are making a diagnosis and what we hope to achieve. One of the problems of diagnoses, particularly those that imply a lifelong condition, is that they tend to become labels attached to the individual. One of the authors of the present chapter (SM) has seen numerous examples where attempts to understand the patient's needs are channelled almost completely by the presence of a diagnosis that someone has made in the past. To illustrate this, one example used in training involves a person in his early 30s with a diagnosis of Asperger's disorder. Without giving specific details (this being a real case), trainees will typically interpret the patient's problems entirely in relation to his diagnosis, rather than the fact that he is now having to live alone, has no friends and has no partner. Even without the diagnosis, the behaviours described in the example could be entirely interpreted in terms of his life circumstances.

To illustrate this even more forcefully, consider two people with symptoms of major depression. Each of them has the same symptoms and severity but one has a biological tendency to get depressed, and this depressed mood has a corresponding impact on the people around them. The other person is depressed *because of* the circumstances in which they live. Each of these people gets the same diagnosis, but the formulation of their problem will be entirely different.

Focusing on the Positive

The above paragraph in no way seeks to minimize the importance of making diagnoses. Diagnoses are an important pathway for clinicians to understand the needs of patients and how they experience the world. They also give important information about the likely time course. At the same time, it can be seen that diagnoses should be one component of a wider investigation. Usually, these investigations tend to focus on the negative factors, for example, the persons symptoms, the impact on their life, the circumstances of their life, etc. At the same time, there is nothing to stop us from focusing on the positive aspects; indeed, this could be a good way to bring

Fig. 5.1 The THRIVE model

together mental health practitioners from a wide range of disciplines.

One thing we can all agree on as professionals is that we want people to *thrive*. We want them to have satisfaction in their lives, and gain the necessary support to help them do this if they have problems. The THRIVE framework (see Fig. 5.1) provides a way of helping people who get away from an 'escalator' model of increasing severity or complexity to one that focuses on a wish to build on individual and community strengths wherever possible. The model was developed by the Anna Freud National Centre for Children & Families and the Tavistock & Portman NHS Foundation Trust [73]. It was designed in the context of mental health services to parents and families, but is applicable to mental health services in general. It aims to talk about mental health and mental health support in a common language that everyone understands.

The framework is needs led. This means that mental health needs are defined by patients and families themselves, alongside professionals, through shared decision-making. Needs are not based on severity, diagnosis or healthcare pathways. The model has the advantage that it also recognizes there are groups of people within society who, though have mental health needs, may not be able to access them for one reason or another. 'Getting risk support' is in this category. Even if we are not in a position to provide services, we should make every effort to monitor the situation.

People with ID typically have multiple problems and needs, many of which are best provided through community resources. Diagnostic issues, seen in this perspective, can perhaps be viewed in a new light, and lead to new ways of working in close collaboration across multiple statutory and voluntary services, as well as with the families themselves.

Tip

Psychopathological evaluation often tends to focus only on negative factors, such as symptoms, their impact on functioning or negative life events. It should also include positive aspects and information from other mental health practitioners instead, in accordance with a person-centred interdisciplinary approach.

Key Points

- In persons with ID and/or LF-ASD, the presentation of psychopathological symptoms and syndromes can considerably vary from that of the general population.
- Factors with a major impact on the identification of psychiatric disorders are diagnostic overshadowing, intellectual distortion, cloak of competence, acquiescence, developmental inappropriateness or psychosocial masking.
- Some behaviours, especially problem behaviours, have been identified as symptoms specific to some psychiatric disorders, taking the name of 'behavioural equivalents'.
- Main adaptations to ID/LF-ASD of the diagnostic criteria for the general population are the DC-LD (Diagnostic Criteria for Psychiatric Disorders for use with Adults with Learning Disabilities) and the DM-ID 2 (Diagnostic Manual – Intellectual Disability, second edition), with reference to the ICD-10 and the DSM-5 respectively.
- Appropriate setting adaptation and adequate consideration of family members represent not only a clinical advantage but also a deontological duty and a legal obligation.

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Psychopathology and Mental Status Examination

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Contents

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Learning Objectives

Descriptive evaluation of psychopathology and the present state examination represent fundamental phases of the psychiatric assessment of the person with ID and/or ASD, although scarce attention received to date in clinical practice and research. The present chapter describes the three parts into which the human psyche is traditionally divided (cognition, affection, and volition/conation) and addresses all the specific aspects and symptoms to be considered during the present state exam such as availability, accessibility, appearance, behavior and motor activity, speech, cognitive functions, consciousness, insight, judgment skills, orientation, attention, memory, sense perception, mood, anxiety, will, and thought.

Current and future clinical care of individuals with ID/ASD needs a substantial reevaluation and expansion of the psychopathological approach in order to improve professional knowledge, practice, and contact with patients' experience. It is to be hoped that the content of this chapter will be significantly extended and detailed in the forthcoming years.

6.1 Introduction

The term "psychopathology" derives from the Greek ψυχή (psyche) for "soul" or "mind," πάθος (pathos) for "suffering," and λόγος (logos) for "discourse," "reason," or "cause search," and is roughly translated into "the study of the sufferings of the soul." The term was coined by the German psychiatrist Hermann Emminghaus in 1878 [1], but it became a scientific discipline only in 1913, with the publication of Karl Jaspers' book "Allgemeine Psychopathologie," [2] who aimed at describing "the individual as a whole in his illness, as far as it is a mental and psychogenic illness" within the complexity of "the soul of the individual." [3] From that moment on psychopathology had increasingly become "the subject matter of psychiatry" [4] and clinical psychology, successfully guiding clinical and scientific progress.

Currently, the term "psychopathology" is employed in a number of different ways, which are commonly grouped into explanatory and descriptive. The former refers to assumed explanations according to theoretical constructs such as psychodynamics or cognitive behavioral models, while the latter consists only of descriptions of any individual behavior or experience which causes impairment, distress, or disability, with limited absent arguments of causation. In the present chapter, the term psychopathology is used in a descriptive way, with specific reference to defining, understanding, and categorizing symptoms as reported by people with intellectual disability (ID) and/or autism spectrum disorder (ASD) and observed through their behavior.

These groups of people have been excluded from research and clinical approaches based on descriptive psychopathology, mostly because they have been considered unable to provide data rich enough to reflect their experiences, or because of ethical reasons, such as being easily coerced or having low coping with distress factors associated with complex assessment and repeated interviews. Thus, authentic voices of persons with intellectual disabilities and autism are often unheard, their behaviors are inadequately observed, and accommodations to ensure their inclusion are rarely made [5–7]. In fact, descriptive psychopathology is important for persons with ID and ASD as it is to the general population, since it provides the fundamental elements for understanding their condition and suffering, as well as making a psychiatric diagnosis.

People with intellectual disability and/or autism spectrum disorder have been excluded from research and clinical approaches based on descriptive psychopathology, mostly because they have been considered unable to provide data rich enough to reflect their experiences, or because of ethical reasons, such as being easily coerced or having low coping with distress factors associated with complex assessment and repeated interviews.

6.2 General Psychopathology

For 250 years, many psychopathologists took for granted that the study of the mind could be divided into three parts: cognition, affection, and volition/conation [8]. The persistence of these areas of human psyche as major taxonomy references for mental phenomena suggests that there may be a natural utility of this scheme for clinical and research purposes, although various organizational models have been proposed [9], in which some of these areas, especially conation, have been merged or eliminated [10, 11].

6.2.1 Cognition

Traditionally, the term cognition refers to mental processes through which information from the environment is acquired and elaborated as knowledge. Cognition is said to include specific mental functions such as attention, memory, understanding of language, and intelligence. Intelligence is often further broken down into learning, reasoning, problem solving, and decision making.

Intelligence is often defined as the ability to solve problems, whether it is to understand how a toy works, to solve a question of trigonometry, or to guess stock market movements.

The Diagnostic and Statistical Manual's fifth edition of the American Psychiatric association (DSM-5) [12] diagnostic criteria for intellectual disability also includes impairments in reasoning, planning, abstract thinking, making judgments, and learning from both practical experience and from education. Intelligence is often expressed as the intelligence quotient or IQ that compares an individual's test score to the average score of a population. By convention, most, but not every, intelligence test converts raw scores to a distribution with a mean of 100 and a standard deviation of 15. Thus, IQs between 85 and 115 are classified as average. IQs above 130 are unusually high and IQs below 70 are unusually low. IQs between 70 and 85 are below average and sometimes regarded as borderline intellectual functioning. Finally, definitions of ID use IQ scores of below 70 combined with other diagnostic criteria, such as significant deficits in adaptive behavior, age of onset during the developmental period, typically defined as before 18–22 years, exclusion of other explanations, and professional judgment (see ▶ Chap. 1).

DSM-5 definition of adaptive behavior includes four domains: (a) communication, such as conveying information to others and understanding information from others; (b) social skills, such as interacting effectively with others, following social conventions, and responding to nonverbal cues from others; (c) personal independence at home is in the community, including bathing, doing the laundry, and using public transportation; and (d) school or work functioning, such as conforming to social standards, and learning and behaving independently at school or work. Adaptive behavior is usually assessed using appropriately normed psychometric instruments such as the *Vineland Adaptive Behavior Scales* [13] or the *Diagnostic Adaptive Behavior Scale* [14]. The nature of intelligence may be more complicated as it may be better to talk about a complex of correlated but distinct mental functions and abilities. The scientific community is still debating whether these mental functions depend on a single, general intellectual capacity, the so-called *g* for general intelligence as the psychologist Charles Spearman called it in the last century, or by distinguishable individual components or facets of intelligence, such as Howard Gardner's multiple intelligences (e.g., musical-rhythmic, visual-spatial, verbal-linguistic, logical-mathematical, bodily-kinesthetic, interpersonal, and intrapersonal intelligences) that are combined differently from person to person. Other models of intelligence have proposed a hierarchical structure with a generic ability at the apex of a pyramid and more specific skills being progressively differentiated in lower levels of the pyramid model. A common distinction is between crystallized and fluid intelligence. Crystalized intelligence refers to learned knowledge and skills which often increases with age, whereas fluid intelligence refers to the ability to perceive new relationships which is independent of prior experience and is reflected in global capacity to reason

and learn and declines with age. Some have suggested that the evaluation of intelligence according to a unitary model is insufficient. Rather it may be better to describe people's cognitive difficulties and specific disabilities and to understand how cognitive difficulties and specific disabilities are linked to psychopathology [15]. It may also be useful to describe the role of highly specific cognitive functions, such as the orientation of attention, the shift of attention, or some reductions in working memory [16, 17] (see ► Chap. 1).

Traditionally, the term cognition refers to mental processes through which information from the environment is acquired and elaborated as knowledge.

The current model of intelligence, based on IQ, is of limited utility for intellectual disability and autism spectrum disorders, given the wide range and variability of cognitive functions and adaptive capacities.

6.2.2 Affectivity/Affection

Affectivity is the area of human psyche related to the ability to experience moods and affects, such as feelings and emotions. The meaning of everyday terms such as sadness, irritation, joy, etc. may seem self-evident, whereas unusual experiences such as delusions, obsessions, and hallucinations require some careful definition. Yet, this is not the case as feelings and emotions are seemingly obscure and changeable events which are difficult to describe. Affectivity seems to be something that is difficult to define as most people report affect as an internal experience available only to the person who is experiencing it, although observable affective behavior can be identified in people with ID/ASD [18].

The terminology with which problematic feelings is dealt with is often ambiguous and controversial. For some authors feelings are simply excited states due to physiological changes in response to some event [19], while for others they are characterized by conscious perception [20]; affects and mood are terms sometimes used antithetically. For example, Taylor [21] stated that affect is "the emotional tone underlying each behavior," while others have opined that the mood is only a part of an affection of the individual, which is a more global function, when, for most authors, almost the inverse is true [19, 22]. For others, these terms are synonyms [23].

For some, emotions arise from archaic nervous center. Emotions apparently arise, grow, diminish, and disappear without active mental participation so that it may appear that there is a complete disconnection between affective and rational life. Emotions are essential for the adaptation of the organism as they provide the driving force for behavior [24].

An alternative, environmentalist view of emotions as the causes of behavior comes from Skinner [25], who considered emotions to be "an excellent example of the fictional causes to which we commonly attribute behavior" [25]. Rather, emotions refer to predispositions to behave in certain ways and to responses that covary which are under environmental control. Thus, the angry person, in response to their supervisor's criticism, turns red and sweats, takes on a facial expression of anger, slams doors, kicks the cat, and watches fights with unusual interest, suggesting that in response to criticism, observing harm holds this class of behavior together.

The emotional aspect of mental illness is characterized by diffuse and impalpable constellations of fleeting sensations and long-lasting dispositions that are difficult to describe. Contemporary research on emotions suffers from a serious conceptual confusion. The first thing to do is to get a hold on the terminology describing emotions [26]. Two concepts seem to dominate the current debate, namely feelings and emotions. *Feelings* are understood as perceptions of bodily changes and affective states such as, for example, discomfort, pleasure, pain, exaltedness, tiredness and sadness. *Emotions* are mostly considered as intentionally or rationally structured experiences such as anger with somebody, surprise at an event, love of someone, pride in one's own behavior, shame at being caught doing something wrong, guilt about one's previous actions, and so on.

We propose a framework for understanding emotional experience that is grounded in

two key points: (1) the definition of "emotion" as felt motivation to move, (2) the distinction between "affect" and "mood" according to their intentional structure. As for the first point, the word "emotion" derives from the Latin *ex movere*. Emotions are the *lived motivation for movement*. Emotions are kinetic, dynamic forces that drive us in our ongoing interactions with the environment. They are *functional* states which may produce movements and *motivational* conditions that project the person into the future providing a felt readiness for action. The connection between emotion and movement can be illustrated as follows. For instance, in sadness I flow downwards in a slow, sinking manner as things appear to be forlornly sinking and sagging downwards. In joy, I flow upwards in a radiated manner as things around me have an uplifted momentum. In retaliatory anger, I feel driven forward, violently attacking as the object of anger grows larger and occupies the foreground. In love, I flow forward in a gently binding way as the loved person flows forward, toward me. In pride, I go upwards in an inflated rising as things grow smaller compared to me. In humiliation, I flow downwards in a plummeting, quick, and violent drop as persons around me grow larger and look at me. In repugnance, I flow backwards as things flow forward toward me. In awe, I flow backwards and downwards in a shuddering manner as things flow forward and upwards, towering above me. In fear, I move backwards in a shrinking and cringing manner as things flow forward, toward me in a looming and menacing manner. In anxiety, I feel suspended in air in a quavering manner which is felt in a menacing manner. Thus, "emotion" is an umbrella term denoting the multifarious phenomena that make up our emotional experience.

As for the second point, at the opposite ends of our emotional experience we find two very different kinds of experiences: affects and moods. The basic difference lies in the fact that affects are focused and possess an apparently clear intentionality or a specific directedness, are experienced as externally motivated with relatively clear environmental determinants, and thus, are more determined and more articulated than moods. Moods, on the contrary, are characterized by a lack of a clear, if any, intentional structure or goal. They are unfocused, and thus, do not possess a specific directedness and *aboutness*.[1] They are felt as undetermined, and there are no specific identified causes for them. They are more indefinite and indeterminate than affects and are often inarticulate. Moods usually manifest themselves as prolonged constellations of feelings such as vague feelings that permeate the person's whole field of awareness. Examples of affects are fear, grief, joy, anger, and boredom. For example, one is fearful because of the imminent threat of negative social validation when one sees one's boss walking toward oneself with a stormy expression on her face. Examples of moods are anxiety, depression, euphoria, dysphoria, and tedium. For example, anxious mood is shown when over an extended period of time there is a sense of impending threat with no obvious source.

> Affectivity is the area of human psyche related to the ability to experience moods and affects, such as feelings and emotions. Emotions are functional states which may produce movements and motivational states that project a person into the future providing a felt readiness for action. Affects and moods are at the opposite ends of human emotional experience: affects are focused and possess an apparent clear intentionality or a specific directedness, while moods represent prolonged constellations of feelings that permeate a person's whole field of awareness.

6.2.3 Volition/Will

Whether volition can be considered an autonomous psychic function is still under discussion. Contemporary classifications of mental disorders do not include it, considering it a behavioral reduction of affectivity (impulse, desire, and motivation) and to a lesser extent

1 Aboutness is a term used in philosophy of mind, often considered synonymous with intentionality.

of cognition (intention, decision, choice, and control). Yet, until the nineteenth century, volition was the most commonly cited concept to explain the ability to initiate or inhibit behavior. It is still commonly used in the forensic field in the sense of personal responsibility for one's own actions and in social and health policies for people with ID and/or ASD in the sense of self-determination.

In everyday language, volition refers to the ability to choose and carry out behavior to achieve certain aims. Scharfetter [27] defined it as a completed, planned activity based on a motivation or a primary need such as hunger or thirst, or a secondary or acquired need like smoking, reading, etc. With the term "motivation," he referred to a state characterized by emotional and cognitive aspects, often linked to a need. In contrast, Jaspers [28] described volition/will as a function welded to the awareness of a goal, the means to achieve it, and its consequences. In this way, he blurred the boundaries between volition/will and judgment.

Various other terms have been used across time to represent alternative interpretations or specific aspect of volition, including intrinsic motivation, goal orientation, conation, self-direction, self-regulation, and self-determination. From the higher faculty that it was, volition/will has been transformed first into a dark ambiguous force including a multitude of drives, longings, and impulses, and then it has been wiped out and substituted in recent decades by notions such as those of "instinct," "drive," "motivation," and "ability to decide."

Whether Volition can be considered an autonomous psychic function is still under discussion. Contemporary classifications of mental disorders do not include it, considering it a behavioral reduction of affectivity and to a lesser extent of cognition. Various other terms have been used across time to represent alternative interpretations or specific aspect of volition, including intrinsic motivation, goal orientation, conation, self-direction, self-regulation, self-determination, and more recently, instinct, drive, motivation, and ability to decide.

6.3 Present State Examination

The present state examination (PSE) of the person with ID and/or ASD is a fundamental clinical moment in the direct evaluation of psychopathological symptoms. Without it, psychodiagnostic tests and questionnaires are of limited value. Unlike a typical conversation between two people, the PSE has specific techniques and rules, some of which apply to all clinical interviews and concern the professional-patient relationship, and others which are much more specific to the PSE. As already noted, people with ID and/or ASD have many relational, communicative, and expressive characteristics, which the interviewer must take into account. These aspects are treated in the ▶ Chap. 5, the one on assessment of psychopathology.

6.3.1 Availability

The PSE begins with the observation of the availability for evaluation. It is an aspect of primary importance, which is not limited to the reaction to the request for information and private experiences, but extends to the whole interpersonal contact that the person tends to take with the evaluator. In persons with ID and/or ASD, availability is often limited by the individual's cognitive and relational characteristics, but this may not effect more experienced and able clinicians' abilities to detect co-occurrence of psychopathology and ID/ASD. The clinician should record if the individual is sympathetic, cooperative, motivated, appropriate to the situation, and willingly collaborates or if the individual is passive, acquiescent, querulous, ironic, arrogant, oppositional, hostile, aggressive, demanding, or if she/he presents behavioral disturbances that make interaction very difficult.

6.3.2 Accessibility

Accessibility refers to the extent to which the persons under examination let the clinician access their intrapsychic experiences. Compared to availability to which it is often

mistakenly equated, accessibility represents a subsequent moment of the PSE. In fact, a patient may be available to a doctor's examination but unable to communicate his or her mental states. In people with ID and/or ASD, accessibility should be assessed in a less rigid way than in the general population and after collecting information on the usual communication attitude and skill from clinical records and habitual caregivers.

6.3.3 Appearance

This part of the PSE assesses the individual's overall exterior aspect. The individual may show a lively, flamboyant attire, typical of manic states, or be unkempt and poorly treated, as seen in schizophrenic psychoses or major depressive episodes. The interviewer must assess appearance in the light of the characteristics of the person with ID and/or ASD, with particular reference to the usual modes of expression, basic skills, life context, sociocultural background, and motor activity. In addition, the interviewer should be sensitive to whether the individual's appearance reflects caregivers' practices and standards, as much as those of the individual.

6.3.4 Behavior and Motor Activity

Persons with ID and/or ASD may feel more anxious than the general population, especially in unfamiliar contexts. During the PSE, anxiety can manifest itself in many forms of inappropriate or challenging behaviors such as nonresponsiveness, shyness, avoidance, opposition, hyperactivity, stereotypies, or aggression. These behaviors can also be the expression of psychopathological conditions, such as anxiety disorders, manic phases, or alcohol intoxication. In these cases, the interviewer may observe severe crisis of psychomotor agitation or destructive behavior toward everything. Conversely, during depressive episodes, the individual's behavior can be slower and more withdrawn than his/her usual behavior.

6.3.5 Speech

According to traditional psychopathology, the evaluation of speech substantially coincides with that of thought, since thoughts are usually expressed in words; however, since in fact the modes of expression of a thought are not limited to speech and speech cannot be considered a direct expression of thought, this is not necessarily true. Indeed, individuals with ID often have difficulties in expressing their thoughts through speech. When evaluating speech, the interviewer should note the form and content of the individual's speech and nonverbal communication. Evaluation of the form of speech should include both qualitative and quantitative characteristics such as the person being talkative, taciturn, or the speech being articulate, accelerated, mumbled, spontaneous, etc. The most common formal alterations of the speech are represented by poverty or wealth of speech, acceleration, slowing down, fragmentation, or lack of speech.

Speech acceleration is associated with an excess in quantity (logorrea), which may appear sometimes apparently unstoppable and sometimes musical, and may include play on word, puns, and jokes as is frequently seen in hypomania and mania, alcohol intoxication, and psychostimulant usage. In manic episodes, extremely high rates of speech may result in incomplete sentences, incomplete words, word repetition (palilalia), echolalia, repetitive sounds, and quickly switching from one topic to another. Sometimes incoherent speech results in so-called word salad most commonly in psychotic disorders, sometimes referred to as "schizophasia."

Marked increases in rhythm and content can be seen in people with pathological anxiety, although lack of speech, stuttering, and incomplete words may also occur. Individuals with ID/ASD and severe language difficulties including those who are nonverbal may show anxiety through intensification of stereotypic behavior, such as rocking, rubbing, and repetitive hand movements, or self-injurious behavior, such as hitting the face or other body parts, hair pulling (trichotilloma-

nia), or biting nails (onychophagia), cuticles, or the skin of the fingers (dermatophagia). Some individuals may speak coherently, but very slowly which may be an expression of psychomotor retardation as in depressive episodes, disorders of consciousness or memory, and organic brain syndromes.

6.3.6 Cognitive Functions

The assessment of cognitive functions is another key component of the PSE in people with ID or ASD, especially when no precise neuropsychological reports are available. Some researchers assert that cognitive functions can be evaluated through their practical, applicative expression, or executive functions. For example, Miyake et al. [29] have suggested a subdivision into three basic executive processes that, although they have some common elements, must be evaluated through the execution of differentiated tasks. These processes include: (a) *Shifting,* the ability to change, alternate behaviors based on the analysis of the results obtained or predictable; (b) *Updating,* the ability to monitor incoming information, their relevance to a current task, and to update the information content replacing the older or irrelevant information with a more recent relevant information; and (c) *Inhibition,* the ability to consciously inhibit excessive, automatic, or predominant responses.

6.3.6.1 Consciousness

The investigation of the state of consciousness focuses on the awareness of oneself and external, objective world. This is particularly difficult in people with ID/ASD with poor or absent verbal abilities. Consciousness can be altered in a quantitative sense, such as in the clouding of consciousness, as in the twilight state, where awareness is restricted to a limited amount of content, or in the dream-like states where the ability to distinguish between the imaginary and the real is compromised.

In certain psychiatric disorders, such as psychotic episodes due to medical illness or a substance, the alteration of consciousness may be particularly marked and be variously associated with anxiety, agitation, hallucinations, language alteration, insomnia, fever, loss of appetite, neurovegetative symptoms, suicidal thoughts, and impulse dyscontrol. Psychotic episodes due to medical illness or a substance is a rare complication of infectious diseases, intoxications, or brain injuries, although Theodor Hermann Meynert (1833–1892), the German psychiatrist who coined the term, related it to acute episodes of bipolar disorders, catatonic schizophrenia, and dissociative syndromes.

A severe loss of consciousness occurs in delirium, where it is expressed mainly as inability to maintain attention and disorganized thought. Persons with more severe ID/ASD can show significant worsening of their underlying attention difficulties and disorganized behavior. During delirium, disorientation can be observed with respect to places of everyday life, astonishment or perplexity can appear even for most familiar objects and people, and hyperresponsiveness to sensory stimuli can determine impulsive glances in the direction of the source where they came from. More rarely in dementia, there is hyporeactivity, reduced motor activity (hypokinesia), somnolence, and even stupor.

To assess the severity of the inability to maintain attention, the patient can be asked to perform a reordering of objects or data, which he would normally handle easily, for example, by placing the largest or smallest cubes or listing the months of year.

Delirium tremens, a particular form of dementia, can occur in chronic alcoholics after 2–3 days of sudden abstinence. It is characterized by the presence of motor alterations, especially tremor, and visual hallucinations which may sometimes include small animals or objects that move quickly in the room or on their body (microzoopsies). In rare cases of delirium tremens, hyperthermia or epileptic seizures may occur.

Other characteristic reductions of consciousness may occur in parasomnias, such behavioral disturbance of REM sleep, night terrors (*pavor nocturnus*), and somnambulism. Since consciousness is already reduced during sleep, it is difficult characterize them clearly. Parasomnias and sleep-related rhythmic

movement disorders are common in persons with ID/ASD [30–35] (see ▶ Chap. 26). The latter typically involve the head and neck (so-called *jactatio capitis nocturna*), occur at sleep onset, during the Non-REM sleep, and are sustained into light sleep.

6.3.6.2 Insight

The term "insight" refers to the degree of understanding of the impairment of one's psychic functioning and of the factors that determine it.

In persons with ID and/or ASD, the lack of insight in the psychiatric disorders that affect them must be carefully distinguished from underlying cognitive difficulty and atypicality in understanding and judging one's own psychic activity.

6.3.6.3 Judgment Skills

The term "judgment skills" refers to the ability to predict the consequences of one's behavior. This can include both practical and moral consequences. In ID/ASD, it is often reduced, especially due to the co-occurrence of a wide range of other mental disorders.

6.3.6.4 Orientation

The term "orientation" refers to the awareness of one's own being, time, and space. In other words, being oriented means knowing who we are, what day it is, the time of day, and where and in what situation we are at the moment. In persons with ID/ASD, orientation must be evaluated in proportion to premorbid functioning. Some authors also include derealization and depersonalization among the alterations of self-consciousness.

Derealization is an altered perception of the environment, in which the environment appears as unreal, dreamlike, foggy, blurred, or flattened. The person who experience derealization describes him/herself as being far away from the real situation or as seeing everything through a filter. Other persons can be perceived as automatons or robots or otherwise without affective coloring. Derealization can be distinguished from psychotic states for that the person maintains awareness of and insight into the strangeness of the experience. Persons with ID/ASD, who have adequate verbal competence, often use the expression "as if." This phenomenon can occur not only during acute episodes of anxiety and mood disorders, especially depressive ones, but also in those related to substance abuse.

In depersonalization, the feeling of detachment does not concern so much the surrounding environment as in derealization, but rather detachment from one's body or mental states. The person who experiences depersonalization describes herself as an external observer of their own existence. In some cases, these experiences intensify so much that the person believes they are on the verge of insanity. Depersonalization differs from disintegration of the self that occurs in psychotic disorders in that the capacity to discriminate between reality and unreality is maintained [36]. Depersonalization is often triggered by a traumatic or intensely stressful event. It is frequently found in posttraumatic stress disorder; anxiety disorders, particularly during panic attacks; major depressive disorder; substances-related disorders, such as excessive marijuana, cannabis, ketamine, and ecstasy use; seizures; and strokes [37]. Depersonalization seems to represent a negative prognostic factor in mood disorders, since it is often associated with comorbidity, chronicity, and lower response to pharmacological treatments [38]. In the DSM-5, the persistent presence of depersonalization and/or derealization represents the first criterion of a homonymous disorder, which is part of the meta-structure of dissociative disorders [12].

In people with ID/ASD, depersonalization and derealization can be expressed with an acute loss of cognitive abilities, especially attention skills, with chaotic, oppositional, and aggressive behaviors, or more rarely social withdrawal.

6.3.6.5 Attention

Attention is an important cognitive function and should be evaluated carefully. It serves to direct the conscience toward specific contents including the outside world, for example,

toward an image, sound, or the inner world, such as a thought, sensation, or memory. Alcoholic and other substance intoxication, psychosis, obsessive-compulsive disorder, and attention-deficit and hyperactivity disorder include a reduction in attention. Some authors use the term "hypoprosexia" to define an attention reduction associated with narrowing of interests, particularly frequent in depression and dementia. Attentional deficits are common in various syndromes including ID and/or ASD, so the identification of additional qualitative or quantitative dysfunctions due to co-occurrent psychiatric problems requires great experience and competence. Unfortunately, to date, knowledge on attention alterations in ID of different origin is limited, as well as their impact on "higher-order" executive functioning abilities. Deficits of disengaging and set shifting have been identified in Fragile-X syndrome, deficits of selective attention in Williams syndrome, and deficits of attention switching in Prader-Willi syndrome [39, 17].

In individuals with ASD, most frequently reported deficits refer to selective visual attention to social targets and selective visual tactile attention [40–42].

6.3.6.6 Memory

Memory can be evaluated informally with a series of simple questions. For example, for short-term memory, by asking the individual to recall information provided shortly before recall; or, for long-term memory, asking facts related to person history and checking confirmation with documents, family members, or staff.

Memory problems can be divided into qualitative and quantitative problems. Amnesia is obviously the most frequent quantitative memory problem, which can further be characterized by the type and chronology of lost memories. The term "total amnesia" is used when there apparently is no memory, even for one's own identity. The term "partial amnesia" is used when the loss of memory is restricted to only some aspects of their existence. Finally, the term "selective amnesia" is used to refer to specific aspects that are mutually linked. Selective amnesia is further divided into: (a) retrograde amnesia for data related to events preceding the onset of amnesia; (b) anterograde amnesia for data concerning events after the amnesia; (c) amnesic lacunae for data related to a specific period of time; and (d) sense-specific amnesia for data concerning a specific sense organ.

It is also useful to differentiate the causes and prognosis of amnesia. Thus, we speak of: (a) organic amnesia, when physical causes have been identified, such as cranial traumas, cerebral circulation disorders, metabolic disorders, and central nervous system degenerative processes; and (b) psychogenic amnesia in relation to psychological trauma or mental disorders, especially mental disorders such as dissociative, anxious, or depressive disorders. Transient amnesia refers to an amnesia which is limited in duration and may involve a return to premorbid functioning. It may occur in association with a moderate head injury. A stable amnesia occurs when there is no recovery of memory loss. Stable amnesias may occur in association with ischemic damage. Finally, progressive amnesia refers to a gradual worsening of memory. This may occur in some degenerative diseases.

Korsakoff's syndrome is associated with amnesia, impairment in learning, and confabulation (see below) and can occur in alcoholism, vitamin B1 deficiency, and some dementias.

The term hypomnesia refers to a reduction in the memory capacity which can be observed in depression, some psychoses, typical aging, and in some stress-related conditions, such as sleep deprivation. In contrast, the term hypermnesia indicates an increased ability to remember that hinders cognitive performance as it includes memories not useful for the task being performed. It is observed especially in manic excitement and in some twilight states.

The qualitative alterations of the memory are categorized as allomnesies and pseudomnesias. Allomnesias consist of distorted reenactments of actual memories while

pseudomnesias are memories of events that never have been experienced, but which are subjectively considered to be real.

Dejà-vu consists of the sensation of having already previously seen an object, person, or place and déjà-vécu refers to the feeling of having already lived a situation that is actually seen or experienced for the first time. In sporadic and transitory forms, they represent very frequent qualitative changes in memory, both in the general population and in people with ID, without a clear pathological significance. On the other hand, if they are persistent, they may be a symptom of psychiatric and neurological disorders, such as epilepsy, alcoholism, depersonalization, and derealization. In jamais-vu and jamais-vécu, the feeling is the opposite of the previous ones; that is to say, to live a completely new experience while in reality we are faced with a habitual, familiar situation. This is most often seen in temporal epilepsy, some anxiety disorders, and psychoses. These alterations can be confused with ecmnesia, where memories are experienced as actual experiences and more often occur in hallucinogens, intoxications, and in some forms of posttraumatic stress disorder. It is called cryptomnesia when a memory appears to the person as an original creation of his or her own mind.

Many qualitative alterations of memory must be distinguished from the fantastic pseudology (or mythomania or pathological lie), which may occur in people with mild-to-moderate ID and ASD. Here, the person refers as experiences things that he has invented from scratch or that he constantly changes to obtain a material or social advantage, often to increase his self-esteem or protect himself from others' negative judgment. In some cases, especially in older people, the fantastic pseudology requires a further effort to differentiate from confabulation, in which the production of fancy data serves to conceal a memory deficit.

People with ID/ASD may present with various memory problem, although it is challenging to assess as changes must be evaluated relative to baseline functioning, which may be difficult to assess formally. When assessing memory with people with ID/ASD, it is possible to resort to combinations of images or objects related to personal memory; for example, one can verify the ability to connect the image of a familiar car with that of its owner or usual driver. Such idiosyncratic assessments are very simple, but lack normative data and standardization; hence, standardized psychometric tests of intelligence and memory are helpful in determining if a person with ID/ASD has a specific memory problem. Many individuals with ID and ASD also show hypomnesia. This makes it more difficult to identify it as a symptom of a co-occurring psychiatric or neurological disorder.

Studies indicate that memory deficits are not homogeneous across all individuals with ID/ASD, but they are related to the specific etiology of ID and subgroups of ASD. That is, in Down syndrome a widespread deficit in the explicit domain of long-term memory (LTM) compared to the implicit one was demonstrated, while studies on Williams syndrome have showed more mixed results. There is evidence of opposite profiles for the implicit LTM, with individuals with Down syndrome relatively preserved and with Williams syndrome relatively impaired [43].

In persons with ASD memory deficits are related more to retrieval than to encoding and many individuals find it hard to remember information if they need a cognitive organizing strategy to aid recall or if they have to detect such an organizing element in the information itself [44, 45]. Other common memory reductions in ASD concern visuospatial and phonological working memory, especially when tasks impose heavier demands on working memory, attaching context to memories, and information that involves social aspects [46]. Problems with prospective memory, which is the ability to remember to carry out a planned intention at an appropriate moment in the future, can also represent a challenge for people with ASD in everyday life [47].

Some persons with ASD show specific memory skills that are much greater than the general population, as those with savantism, although their nature is still to be defined [48].

Memory problems can be divided into qualitative and quantitative. Amnesia is the most frequent quantitative problem, which can further be characterized by the type and chronology of lost memories. Qualitative alterations are categorized as allomnesies and pseudomnesias.

Differential patterns of memory deficits are documented across different etiological and clinical groups of individuals with ID and/or ASD.

6.3.7 Sense Perception

This expression refers to the neurological organization of sensory experience, that is, to the reaction to internal and external stimuli received by the sense organs. In order to transform them into perceptions, sensations must be integrated with a system of products of central neuropsychological activities, such as memory (e.g., past experiences), emotions (e.g., fear or interest), or volition (e.g., motivation).

Perceptual processing can change sensory information, for example, illusions are contrasting or undefined sensory stimuli which blend with perceptive patterns so tightly that it makes it impossible to distinguish the former from the latter. Illusions can involve all the senses, but the visual illusions are the most characteristic and well known. Pareidolia is the tendency to perceive familiar forms in disordered stimuli, such as seeing faces in the moon or animals in the clouds. This may be reduced in people with ASD, especially when related to social stimuli [49, 50].

The alterations of the sense perception of major clinical interest are hallucinations which are perceptions in absence of an object or an external stimulus to be perceived. They can occur for each of the sensory modalities, including visual, auditory, gustatory, olfactory, and tactile systems. Hallucinations may be coenaesthetic, enteroceptive, and proprioceptive. Coenaesthetic hallucinations are hallucinations related to one's body; they are neurologically and physically impossible, such as perceiving a scratching on the inside of one's skull. Enteroceptive hallucinations stimuli are hallucinations regarding stimuli from inside the body, such as proprioception to detect pressure and internal mechanical stimuli that assist in the correct performance of the neuromotor functions such as standing upright and coordination of movements. Auditory hallucinations are found in psychotic disorders, substance poisoning, and, less frequently, in bipolar and depressive disorders. Visual and olfactory hallucinations are more common in organic disorders.

Simple hallucinations relate to a single sensory modality and do not involve complex cognitive elaborations, for example, whistles, points or bright areolas, and flashes of light or color, whereas complex hallucinations involve more senses and integration by brain areas other than primary sensory ones, for example, seeing a lion and hearing and feeling it roar. In most cases, hallucinations are interpolated or inserted in the typical sense-perceptive context, but rarely involve changes in the whole perceptual field. The most frequent hallucinations are auditory, followed by visual hallucinations.

In people with ID/ASD, who have limited verbal communication skills, hallucinations may be indicated by significant changes in behavior. For example, a person with some visual hallucinations may continue to look at an area of the environment where there does not seem to be any relevant stimuli and another person with auditory hallucinations can show sudden communication gestures or plug their ears. A person with coenaesthetic hallucinations may appear strangely intent on removing something from their skin or inspecting a particular area of their body. But in people with ID, they often experience their own thoughts as real voices. The clinician has to distinguish whether the reported auditory hallucination is experienced by the patient as a real perception of the outside environment or if the patient experiences his/her own thoughts aloud. A list of observable aspects and behaviors with possible hallucinatory equivalents is reported in Table 6.1. These equivalents must be carefully distinguished from certain behaviors typical of ID/ASD, such as speaking in a stereotypical way, speaking to themselves, even asking questions and

Table 6.1 Examples of observable/behavioral equivalents of hallucinations in people with ID/low-functioning ASD

Hallucination (sensorial type)	Behavioral observable equivalents
Visual	Fix an area of the environment, in which there does not seem to be anything particularly to be seen (even considering any attention and special features) Suddenly turn to an area of the environment Nod Make gestures with the hands with apparent reactive or communicative value Move as if he were defending himself or fighting Move as if he were loving Cover the eyes with hands or various bandages Look bad or even angrily at people first appreciated or strangers Avoid or hide from family members or with whom he normally has a good relationship Inspect objects, food, or beverages with exaggerated and unusual intensity
Auditory	Nod Make gestures with the hands with apparent reactive or communicative value Suddenly turn to an area of the environment Cover the ears with hands or fingers, or cover them with various materials
Tactile	Rub or make gestures as if he wanted to take something off his skin
Olfactory	Smell objects, food, or drinks with exaggerated and unusual intensity Curl the nose and grimace as if he perceived unbearable odors Sniff the air as if to check for the presence of gas or other contaminants
Somatosensory Coenesthetic	Make gestures as if he wanted to take something off his body Wear heavy, close-fitting clothing or clothes with many layers of clothing (clothes are interpreted as containers to prevent the escape of parts of the body, perceived as unstable) Bandage the ankles or the wrists with various materials Wear hats, bandanas, or foulards, inadequate to the rest of clothing and context
Multisensorial	Move as if he were loving Move as if he were defending himself or fighting

giving answers or using unusual inflections and tone of voice, shouting or speaking with a loud voice, imitating others, or behaving in ways induced by others. Hallucinations must also be distinguished from hallucinosis, in which the awareness of sense perceptions is not shared by others. In most cases, they are simple changes often in the course of organic pathologies, such as intoxications, neoplasms of the encephalic trunk, angiopathy, cranial trauma, and epilepsy.

6.3.8 Mood

Mood alterations are present in many psychiatric disorders. For example, "low" mood or apathy (from Greek without pathos, i.e., feeling), that is, the lack of emotional resonance, is typical of major depression, but is also found in other disorders, including schizophrenic, bipolar, substance-related, neurocognitive, anxiety, and stress-related disorders. Periods of apathy may also occur in situations of extinction of adaptive behavior, such as losses related to depression, like unemployment, loss of family members and friends, and moving place of living or work. Irritability is sometimes defined as a proneness to anger [51] and excessive responsiveness to stimuli. It is more common in mania, premenstrual dysphoric disorder, borderline personality disorder, and some forms of depression. It is also observed in elevated or

expanded mood which characterizes the hypomanic and manic phases of bipolar disorders, but is also found in substance-related disorders, neurocognitive disorders, and pathological gambling.

A severely altered mood can also be expressed in patients with suicidal ideation and behavior, which are in fact frequently found in depressive and bipolar disorders. Suicidal thoughts and attempts may sometimes have a more complex relationship to psychopathological conditions, as demonstrated by the repeated association with delirium, neurocognitive, dramatic personality disorders, behavioral disturbances, and stress-related disorders. In schizophrenia, paradoxical affective responses may occur such as hilarity when listening to sad topics. In patients with ASD, empathy and emotional reciprocity are lacking (see ▶ Chap. 16). In some dissociative or conversion disorders, emotional detachment may occur, for example, a patient may talk about the severity of his/her own symptoms with absolute coldness (*belle indifférénce*).

During the PSE interview, mood may be observable through the individual's behavior, facial expressions, gestures, speed of movement, response latency to stimuli, and general reactivity to context. Emotional behavior during the PSE may differ in quantity, for example, as it is the case in reduced psychomotor activity, or be too intense. Emotional behavior may also differ in quality, for example, when the person shows detached or paradoxical emotions.

Mood alterations may occur in individuals with ID/ASD in whom it may often replace depressed affect. In the person with ID/ASD, mood changes can also be observed through striking variations in behavior, both in quantitative terms, as reduction or increase in their activities, and qualitative, as an appearance of aggressiveness toward others or to oneself, noncompliance, or provocation (see ◘ Table 6.2). A systematic description of the presentations of emotional changes related to psychiatric disorders in individuals with ID can be found in Vannucchi [52]. It should finally be noted that persons with ID/ASD, due to their insight and language problems, may have serious difficulties in distinguishing bodily feelings related to emotions from other bodily feelings, such as pain, in expressing their emotions, especially their moods, and focusing on the event or situation which has caused them. Sometimes observational measures of mood-related behavior may be helpful [18].

In individuals with ID/ASD, mood alterations may often replace depressed affect. Mood changes can be observed through striking variations in behavior, both in quantitative terms, as reduction or increase in their activities, and qualitative, as an appearance of aggressiveness toward others or to oneself, noncompliance, or provocation.

6.3.9 Anxiety

The term anxiety refers to an unpleasant mood, characterized by strong apprehension, due to uncertainty or expectation of something of great subjective importance which cannot be clearly focused. It is often related to physical or social threats or the possibility of such threats and is often highly adaptive in maintaining the integrity of the organism by avoiding or minimizing harm. In serious cases, it involves an overstimulation of the vegetative nervous system including tachycardia, thoracic constriction, rapid breathing, sweating, and abdominal spasms, which may also exacerbate the emotional response itself. Anxiety also disrupts ongoing adaptive behavior [53] and may imply maladaptive conducts like fight, flight, or freezing (see Kretschmer) [54], so that the anxious person may freeze, does not talks, and no longer engages in adaptive responses to threats such as effective avoidance of the feared stimuli.

Anxiety can also imply sensory-perceptual changes, such as impaired vision (reduced visual fields or objects appearing more distant than they actually are), a feeling of unreality, dying or going insane, and to cognitive problems, such as difficulty in concentration,

Table 6.2 Examples of observable/behavioral equivalents of mood changes in people with ID/low-functioning ASD

Mood alteration	Behavioral/observable equivalent
Deflection	Psychomotor slowdown Reduction of participation in activities or opposition Aggression, both physical and verbal, toward objects or other people Self-harming or self-mutilating behavior Reduction of resistance and/or frequency in carrying out usual activities Reduction of sexual activity or sexually oriented behavior Reduction of a range of emotional responses Pervasive tendency to cry
Elevation or expansion	Psychomotor agitation and/or restlessness Increase in stereotypies Self-harming or self-mutilating behavior Excessive search for interpersonal contact and communication Excessive commitment to activities or occupations Aggression, both physical and verbal, toward other people or objects Excessive increase in sexual activity or sexually oriented behavior Excessive self-esteem and/or behaviors sustained by the feeling of being capable of extraordinary or unrealistic performances Increase in phonation or speech
Irritability	Easy access to anger, even for stimuli and normally well-tolerated situations Aggression, both physical and verbal, toward objects or other people Self-harming or self-mutilating behavior Increase in stereotypies
Lability	Rapid changes in mime and gesture Rapid switches from laughing to crying or vice versa

Hurley [55]; Pary et al. [60]; Charlot et al. [61]; Vannucchi et al. [52]

memory, or hypervigilance. Pathological anxiety is a symptom of anxiety disorders such as panic disorder, agoraphobia, specific phobia, separation anxiety disorder, generalized anxiety disorder, and obsessive-compulsive disorders. It may also occur in delirium, intoxication or substance withdrawal, neurocognitive disorders, somatic symptoms disorder, psychotic disorders, body dysmorphophobia, and stress-related and mood disorders. Sometimes anxiety symptoms occur in response to concerns related to other psychiatric disorders.

In the PSE with individuals with ID/ASD, some anxiety disorders, such as simple phobias, are easily recognized. Other anxiety disorders, such as generalized anxiety disorder, may not be so readily apparent. Some have suggested that anxiety in individuals with ID/ASD may express itself in increased motor activity, intensification of stereotypies, or a tendency to cry [55].

6.3.10 Will

The alterations of the will can be quantitative, such as abulia, hyperbulia, and motor inertia, or may be qualitative like dysbulia, automatism, negativism, and impulse control. Abulia (from the Greek "without will") is expressed by the inability to make decisions independently, take initiative, or to initiate an action,

even if the action is trivial or knowing that it is necessary. Hyperbulia is the opposite of abulia. It manifests itself through an excess of decision-making and initiative, often with negative consequences.

Negativism consists of an energetic and persistent opposition to the movements that one wants to make (passive negativism) or in the execution of actions opposed to those actions that are required (active negativism). The term automatism means acts performed mechanically and without participation of will. Autonomous automatisms are automatic and determined by spontaneous activation of the motor centers. They are automatic but originally were voluntary, yet made subconscious by the great repetition. An example of autochthonous automatism is postepileptic automatisms, consisting of a repetition of complex actions following a seizure and of which the individual does not recall. Abulia is seen in major depressive episodes and some forms of psychoses. Hyperbulia is more common in manic and hypomanic episodes, but it can also occur in mixed-mood episodes and anxiety disorders, and may be associated with hyperkinesis. In some psychotic disorders like schizophrenia, abulia may present as catatonia.

Qualitative alterations in will are also found in disruptive behavior disorders, impulse control disorders, kleptomania, pyromania, antisocial and borderline personality disorders, and some forms of suggestibility, for example, in histrionic or dependent personality disorder, eating disorders, and addiction and substance disorders. Lack of will also characterizes disorders of craving (uncontrollable desire to consume a substance or to behave), loss of control, and underrating of consequences.

Alterations of will may occur in people with ID/ASD without being a symptom of an additional psychiatric disorder. This must, therefore, be carefully defined on the basis of changes from premorbid behavior.

Stereotypies are repetitive behaviors with no immediately apparent purpose. Examples include body rocking, head hitting, and hand flapping. Stereotypies often appear relatively invariant in form. At first glance, they are similar to other repetitive and unchanging behavior, such as compulsions and tics. Compulsions, however, differ in that they typically involve cognitive components, such as fear of contamination or disaster, and typical topographies include hand washing and checking. They mostly serve to get rid of anxiety in response to contamination or an intrusive thought. Whereas stereotypies appear in early development, including the development of typical children, compulsive behaviors rarely appear before the third or fourth year of life and tend to begin in teenage years.

Tics are usually sudden, rapid, ballistic movements and are often topographically simpler movements than stereotypes, although motorically more complex forms of tics exist. Tics usually affect facial muscles of the face (as in blinking, stretching of the mouth, and curling of the nose), vocal apparatus (as in coughing and making noises), and upper body skeletal muscles (shoulders and limbs) and sometimes the trunk. In DSM-5 [12], tic disorders are part of movement disorders together with the disturbance of coordination development and stereotyped movement disorder. Tourette syndrome is characterized by multiple, often more complex and energetic tics associated with the emission of inarticulate sounds, coprolalia (cursing), and behavioral and anxiety disorders. Tourette's syndrome may start as early as 5 years, although many individuals develop related symptoms earlier.

Stereotypies are common in individuals with ID/ASD, especially those with more severe ID. Some individuals with ID/ASD also show tics and Tourettes syndrome that appear broadly similar to these disorders in the general population. Relatively little has been published on this topic.

Alterations of will may occur in people with ID/ASD without being a symptom of an additional psychiatric disorder. This must, therefore, be carefully defined on the basis of changes from premorbid behavior.

6.3.11 Thought

Thought is defined as mental activity that allows us to evaluate reality and formulate judgments through processes of association, correlation, integration, abstraction, and symbolization. Traditionally, for the evaluation of thought, clinicians must refer to both form and content [56]. Quantitative alterations in thinking consist of acceleration and slowing down. Acceleration is characterized by the difficulty in focusing on content, superficial thinking connected with shifting from topic to topic, and consequently constructing associations based on uncommon or idiosyncratic features, sometimes resulting in so-called flight of ideas (idiorrhea). Flight of ideas can be so severe that the patient's spoken words cannot keep up with the rapidly shifting content of his/her thinking such that the outcome can be mutism or motor blocking, which is sometimes seen in very severe manic episodes. In slowing down, thought runs painfully slowly. The patient responds to questions with hesitation and delay (retarded thinking), often laconically. It may be seen in major depression, catatonic schizophrenia, and organic syndromes. A particular type of slowing down is viscous or ponderous thinking, which appears to contribute to alterations in the quality of thought. Viscous thinking is characterized by affirming a concept or acting behavior and failing to abandon it, despite the fact that the context requires it.

Qualitative alterations are much more complex than quantitative ones. Circumstantial thought is characterized by the indirect answers to questions or the conversation's goal. It may include the insertion of irrelevant content, long windedness, or prolixity. Perseveration consists of the continuous repetition of a content of thought, even when it is extraneous to the context. Perseveration can be difficult to distinguish from viscous thinking, which is characterized by the inability to give up a point of view. In perseveration, uncritical use of an idea prevails. Perseveration is often mistakenly equated to intrusive or obsessive thoughts, which instead differ for not being evoked, but rather arising against the person's will who indeed recognizes them as inappropriate.

Perseveration is common in individuals with ID/ASD, while viscosity is more frequent in those with co-occurrence of seizure epileptic disorders and obsessions in those with co-occurrence of obsessive-compulsive disorder.

Dissociation consists of a loss of logical links between individual ideas. It is seen in fragmented and bizarre speech. Frequent derivations of dissociation are represented by fusion, derailment and block (or barrage). Magical thinking consists in the attribution of cause-effect links to stimuli, objects, or situations that did not in fact cause something. In nonpathological forms, beliefs change in the face of criticism. It is widespread in the general population in the form of superstition. Pathological forms are associated with psychoses and obsessive-compulsive disorder with little insight (► Chap. 20).

Primitive or predicate thinking [57] is a form of unlogical thinking which results in illogical conclusions. It equates persons and objects merely on the basis of their predicates or functions, for example, a person who thinks of being a doctor only because he is wearing a white coat or because he is using a stethoscope.

Among the qualitative alterations of thought, qualitative alterations of abstraction deserve special consideration. Abstraction consists of the ability to pass from the particular to the general and to replace with symbols the concrete. It is considered by some to be a cognitive function. Many people with ASD are very concrete thinkers. They tend to misunderstand easily or not understand metaphors, analogies, euphemisms, double meanings, jokes, and puns, and references to emotions and feelings. To support them with this difficulty it may be useful to constantly refer to images, objects, or situations that they have combined with various abstractions. Problems with abstract thinking can occur in the case of co-occurrence of psychotic disorders, severe mood disorders, dementias, and brain lesions of the frontal areas or their connections with other cortical structures. To evaluate abstraction skills, you can informally

ask the individual with ID/ASD to sort sets of objects, blocks, tokens, etc., choosing those that "go well together" or "that have something in common." As a counter-test, you can then ask the person to group the same objects on the basis of a new criterion. The behavior manifested by people in these tests also provides information on the ability to identify different plausible criteria for a task and to flexibly change them.

The most serious and most striking alteration in the quality of thinking is seen in delusions, commonly defined as an error in belief that does not recede to criticism or to the contrary evidence of the facts. Jaspers [28] distinguished primary delusions (or delusions proper) from delusion-like ideas on the basis of their comprehensibility or incomprehensibility. Delusion proper is specific to schizophrenia patients and the most characteristic of them are delusional perceptions in which a person correctly perceives a given worldly object but realizes that the object is there to mean something personal, of extraordinary relevance to him, and typically disconnected from common sense. Jaspers identified the peculiarities of delusion proper in the certainty of the belief, the resistance to influence, and in the absurdity of its content. Delusion-like ideas are those in which any relationship with external circumstances or other psychic alterations is detectable. Holotimic delusions (from Greek involving an overall change in mood) are emblematic, such as delusions of guilt, ruin, and illness. Paradigmatically nihilistic delusions such as the patient is convinced he/she is dead and the world is also dead are linked to serious mood swings. Delusions of grandeur, invention, and wealth are associated with marked euphoria.

In people with ID/ASD and psychotic or mood disorders, delusions tend to be less bizarre and complex than in persons of the general population with the same psychopathological conditions. In those with serious communication difficulties, they must be evaluated through significant variations in the quality of behavior, perhaps through avoidance, anger, or aggression toward objects, environments, people, or situations that were previously well tolerated or even appreciated, sudden refusal of assistance and treatment, and social withdrawal.

> In persons with ID/ASD, delusions tend to be less bizarre and complex than in persons of the general population. In those with serious communication difficulties, the presence of delusion must be evaluated through significant variations in behavior, such as the onset or exaggeration of avoidance, anger, or aggression toward objects, environments, people, or situations that were previously well tolerated or even appreciated, sudden refusal of assistance and treatment, and social withdrawal.

> The present state examination of the person with ID and/or ASD is a fundamental clinical moment in the direct evaluation of psychopathological symptoms. Without it, psychodiagnostic tests and questionnaires are of limited value. It must include the assessment of all the following functions: availability, accessibility, appearance, behavior and motor activity, speech, cognitive functions, consciousness, insight, judgment skills, orientation, attention, memory, sense perception, mood, anxiety, will, and thought.

6.4 Final Considerations and Future Directions

Research and practice has made progress in the assessment of psychopathology as shown by the development of numerous psychometric measures and structured diagnostic interviews, although much is left to be done especially with reference to persons with greater impairment of communication and insight. This has been fostered by social pressure for clinical casualness and health costs containment as well as research investments, which have increasingly focused on neuroimaging, genetic, and neuropsychology. The skill to precisely and carefully assess mental states and symptoms of mental suffering was a core attribute of mental health professionals, but today's curricula pay increasingly less atten-

tion to its training [58], thus, blurring the border between co-occurrence of psychiatric disorders and variants of the underlying neurodevelopmental disorder.

Current and future clinical care of individuals with ID/ASD needs a substantial reevaluation and expansion of the psychopathological approach in order to improve professional knowledge, practice, and contact with patients' experience. It is to be hoped that the ideas of this chapter will be significantly extended and detailed in the forthcoming years. In fact, despite the prophecy from the end of the last century that psychopathology would be doomed by neurobiological parameters, psychiatric diagnosis continues to rely exclusively on psychopathology, even in the very latest classificatory systems, such as DSM-5 [12] and ICD-11 [59], and there is a growing demand for personalized symptom-based approaches and precision psychiatry.

Tip

Current and future clinical care of individuals with ID/ASD needs a substantial reevaluation and expansion of the psychopathological approach in order to improve professional knowledge, practice, and contact with patients' experience. It is to be hoped that the ideas of this chapter will be significantly extended and detailed in the forthcoming years.

Key Points

- The Present State Examination of the person with ID and/or ASD is a fundamental clinical moment in the direct evaluation of psychopathological symptoms. Without it, psychodiagnostic tests and questionnaires are of limited value.
- The Present State Examination must include the assessment of all the following functions: availability, accessibility, appearance, behavior and motor activity, speech, cognitive functions, consciousness, insight, judgment skills, orientation, attention, memory, sense perception, mood, anxiety, will, and thought.
- Differential patterns of memory deficits are documented across different etiological and clinical groups of individuals with ID and/or ASD.
- In individuals with ID/ASD, mood alterations may often replace depressed affect. Mood changes can be observed through striking variations in behavior, both in quantitative terms, as reduction or increase in their activities, and qualitative, as an appearance of aggressiveness to others or to oneself, noncompliance, or provocation.
- Alterations of will may occur in people with ID/ASD without being a symptom of an additional psychiatric disorder. This must, therefore, be carefully defined on the basis of changes from premorbid behavior.
- In persons with ID/ASD, delusions tend to be less bizarre and complex than in persons with other clinical conditions and may manifest through significant variations in behavior, especially in those with serious communication difficulties.
- Current and future clinical care of individuals with ID/ASD needs a substantial reevaluation and expansion of the psychopathological approach in order to improve professional knowledge, practice, and contact with patients' experience.

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Problem Behaviour

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Contents

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Learning Objectives

- Prevalence of problem behaviour in adults with intellectual disabilities
- Risk factors for problem behaviour
- Assessment of problem behaviour
- Rating Scales for problem behaviour

- A higher proportion of people with intellectual disabilities compared with the general population who do not have intellectual disabilities display problem behaviour.
- Various factors such as age, gender, level of intellectual disabilities, level of communication, co-morbidities such as psychiatric disorder, other neurodevelopmental disorders such as autism spectrum disorder (ASD) and attention deficit hyperactivity disorders (ADHD) may influence the rate and severity of problem behaviour.
- Problem behaviour, in general, and aggression, in particular, poses a major management problem and is a major barrier to social integration, leads to caregiver stress, community placement breakdown and use of restrictive practices.
- A thorough person-centred assessment is vital for a successful management.
- The assessment should take a bio-psycho-social approach by incorporating assessments of the behaviour, the person showing the behaviour, medical, psychiatric/psychological and social/environmental issues.
- A number of instruments are available to rate different types of problem behaviour and assess their impact. Some of these are generic and some are specific to problem behaviour.
- Some functional assessment scales that assess the environmental effect are also available.
- The aetiology of problem behaviour is complex and often is the outcome of an interaction between internal factors within the person and external factors such as the environment.
- Therefore, the management of problem behaviour requires input from a multitude of professionals with relevant and specific skills.
- The ultimate aim of the management of problem behaviours should be to improve the quality of life of the person with intellectual disabilities and their family members.

Tip

Problem behaviour is more prevalent in people with intellectual disabilities than without and is multi-factorial in aetiology.

Therefore, a multi-professional, person-centred, bio-psycho-social approach is needed for a thorough assessment and management of problem behaviours.

7.1 Introduction

Individuals with intellectual disabilities (ID) have an increased risk for the development of problem behaviour (PB) such as aggressive behaviour (AB). AB is common in this population, and it is often targeted for intervention [1, 2]. AB continues to be a major concern for families, caregivers, clinicians and other professionals [3] acting as a major barrier to social integration/inclusion and limiting access to certain residential settings, education, occupational opportunities or social acceptability [4–6]. Furthermore, it has been reported that AB predicts admission to psychiatric inpatient units [7], is the most common reason for referral to mental health services or specialist behavioural support teams [8], is related to use of psychotropic medication [9, 10], is associated with placement breakdowns [11], use of restraint and restrictive practices [12] and higher cost of service provision [13, 14].

7.2 Terminology

Different terminology is currently used in the field to describe PB. The term ‘challenging behaviour’ [15] has received a wide acceptance replacing other terms such as maladaptive, abnormal or aberrant behaviour or

behaviour disorder or behaviour problem. Other terms such as 'behaviour that challenges' or 'behaviour of concern' have also been used recently [16]. Emerson [15] defined challenging behaviour as 'culturally abnormal behaviour(s) of such intensity, frequency or duration that the physical safety of the person or others is likely to be placed in serious jeopardy, or behaviour which is likely to seriously limit use of, or result in the person being denied access to, ordinary community facilities'. The term PB is used by many organisations such as the Royal College of Psychiatrists in the UK [17], and the World Psychiatric Association [18, 19]. PB is seen as a type of behaviour rather than behaviour being a 'problem' as such. PB has been defined as 'socially unacceptable behaviour that causes distress, harm or disadvantage to the persons themselves or to other people, and usually requires some intervention' [19].

The closest categories for PB in the Diagnostic and Statistical Manual-5 (DSM5) are 'Disruptive, Impulse-control, and conduct disorders' and personality disorder [20]. However, given strict criteria for diagnosis, it seems that in a large proportion of people with ID this classification is difficult if not impossible to apply [21]. There are similar problems in categorising PB in ID using the International Classification of Diseases tenth version (ICD-10) criteria [22]. It is not only difficult to define PB, but it is also not clear which behaviours should be classified as PB. For example, the following behaviours may or may not be seen as types of PB; pica, disruptive and oppositional behaviours, hyperactivity, stereotype, social withdrawal, wandering aimlessly, night-time disturbance, inappropriate sexual behaviour (including aggression), antisocial behaviour, screaming/shouting aimlessly (not particularly directed to anyone), scattering objects indiscriminately, socially unacceptable personal habits such as hoarding, smearing faeces and taking others' possessions. However, some of these behaviours could be the manifestation of other pathology. For example, hyperactivity could be part of attention deficit hyperactivity disorder (ADHD) or mania, stereotype could be part of an organic disorder or autism spectrum disorder (ASD) or adverse effects of medication, and social withdrawal could be due to depression or dementia. It is possible that some PB such as antisocial behaviour and aggression are defined as personality disorder when exhibited by people with mild ID [23].

AB is the predominant form of PB [24, 25]. In this chapter, we have presented data interchangeably for both PB and AB. AB can include verbal aggression, physical aggression towards others, property destruction and self-injurious behaviour (SIB; physical aggression towards the self). Outwardly directed aggressive behaviour is often distinguished from SIB as different aetiopathology may be responsible for these two different types of behaviour [26]. For example, outwardly directed aggression is often perceived by many as part of a communication process, whereas SIB has been associated with many organic factors such as genetic syndromes [27]. SIB has also been linked to a dysregulated internal opioid system [26, 28–30]. Some SIB has been shown to be part of an obsessive compulsive disorder or stereotyped behaviour or dysregulation of arousal [26]. The operant model of the development of SIB has received some compelling support in the literature, especially with relation to the strengthening of SIB, however, studies do not support the notion that 'social reinforcement processes are responsible for shaping stereotypy into SIB' [26, 30, 31].

7.3 Prevalence of Problem Behaviour Among People with Intellectual Disabilities

There have been many attempts to determine the rate of PB and AB in people with ID. However, difficulties remain in case detection (finding all people with ID in a representative sample) and case definition (difficulty in defining PB and AB and also use of different methods to record prevalence) [32–34]. Similarly, prevalence rates vary due to differ-

ent populations (e.g. in ages and location of study), time scales and forms of 'prevalence' (e.g. point prevalence, life-time occurrence or time-limited incidence) used in different studies [35]. Epidemiological studies report a very varied rate from 2% to 60% for PB (see Tables 7.1 and 7.2) [5, 33, 36–39]. Prevalence of AB tends to fall somewhere in between and, among all four types of AB reported, verbal aggression tends to be the most common type. Often, adults with ID exhibit multiple forms of AB and other types of PB [6, 40–43]. However, there is little research that has explored the topography of AB in adults with ID in detail, and the field remains dominated by retrospective research lacking detailed descriptions of behaviours in specific settings [44].

7.4 The Impact of the Behaviour

Those who exhibit AB are perceived to be less satisfied with their life situation than those who do not and tend to have a poorer quality of life [45] with a reduction in behaviour being linked with improved quality of life [46, 47]. Presence of AB inevitably increases the risk of injury to the person and those around them. Frequency of AB has been reported to be the best single predictor of restraint-related injury, with one in three emergency restraints resulting in personal injury [48]. Apart from the people with ID, AB also impacts paid caregivers, informal caregivers and family members, including siblings as AB is a common source of stress leading to negative emotions [49, 50]. Paid carers, working in community group homes with residents who exhibit AB, report lower job satisfaction and more anxiety compared with similar workers in homes without such residents [51] and a significant relationship between AB and staff burnout has been reported [52, 53]. In addition, 42% of all caregivers of adults who exhibit physical aggression towards others reported that they felt unable to cope with the behaviour, rising to 58% for family carers of those known to services [54] with fear of assault also relating to emotional exhaustion [55].

7.5 Natural History of Problem Behaviour

PB is commonly considered as a chronic and enduring condition, with aggression during childhood being a major predictor of aggression later in life [56, 57]. Few studies have investigated the age at which such behaviour tends to first emerge, however, available studies suggest that behaviours often appear in childhood [58]. Cross-sectional research reports peak in adolescence/early adulthood and decline from around 30–35 years [25, 33, 54, 59]. The pattern reported in cross-sectional research suggests that remission with age occurs for some individuals. Cooper and colleagues [42, 43] reported 2-year remission rates of 29–38%, and onset of episodes in other individuals, thus, suggested PBs are relapsing-remitting conditions. Other research suggests persistent rates of 50–90% at different time, with rates varying according to type of behaviour and time period studied [60, 61].

7.6 Risk Factors Associated with Problem Behaviour

Epidemiological research has sought to identify factors, both within the person and, less commonly, in the environment, that are associated with PB and AB. Identifying such correlates aids the understanding of the processes involved in a person displaying PB, as well as helping to develop effective management strategies and inform service provision [42, 43, 62]. We presented here an overview of the most commonly studied risk factors for PB. Tables 7.1 and 7.2 present details of community-based studies that have investigated the prevalence of, and factors associated with, AB in adults with ID.

7.6.1 Age

It is difficult to ascertain a true association between age and the rate and type of PB in the absence of any long-term longitudinal cohort study. The current knowledge is based on

Table 7.1 Prevalence of and factors associated with outwardly directed aggressive behaviour in community studies of adults with intellectual disability

Authors	*N*	Population	Assessments	Diagnostic criteria	Findings (% rounded up to closest decimals)
Eyman and Call[a] [127]	5243	Administrative database of service users with ID aged 13 and over, USA	Selected items of the ABS completed by direct-care workers and social workers	The behaviour was rated as occurring frequently or occasionally	13+: [d]28% physically aggressive, 22% destructive and 19% verbally aggressive. People with physical aggression were older, male, of lower ability and institution residents. People with destructiveness were of lower ability, male and institution residents. People with verbal aggression were older, male, of higher ability and institution residents
Jacobson [38]	32,112	Administrative database of service users (children and adults) with ID in one state, USA	42-item questionnaire routinely completed by staff	The general frequency of up to three problem behaviours could be reported	All ages: 11% physically aggressive, 4% destructive and 6% verbally aggressive Aged 22+: 'Psychiatric and developmental disabilities' group: 18% physically aggressive, 7% destructive and 12% verbally aggressive. 'Developmental disabilities only' group: 11% physically aggressive, 4% destructive and 6% verbally aggressive. Adults with physical aggression: 17% profound, 14% severe, 8% moderate and 5% mild ID. Adults with destructiveness: 7% profound, 5% severe, 3% moderate and 2% mild ID. Adults with verbal aggression: 3% profound, 8% severe, 10% moderate and 9% mild ID
Qureshi and Alborz [128] and Emerson et al. [68]	695[b] (4200)	Children and adults using ID services in seven Health Authority areas in England, UK	All ID services were asked to identify people with problem behaviour. Key informants were then asked to return a questionnaire on each person they identified	Interview with staff for each person who had been identified as having aggressive behaviour, on its type, intensity and frequency	All ages: [e,d]11% for all categories, or [f,d]7% for serious/serious but controlled physical aggression, [e,d]9% all categories or [f,d]5% for serious/serious but controlled destructive behaviour
Sigafoos et al. [25]	261	Children and adults using ID services in one state in Australia	Managers were asked to distribute surveys to a senior staff in each service, asking for a questionnaire completed on each person with aggressive behaviour	A two-page questionnaire listed 14 topographies of aggressive behaviour and sought frequency as never (0) through to more than 15 times a day [7]	All ages: 261 people engaged in aggressive behaviour, cited as 11% of 2412 service users; but the non-response rate was unclear

Harris [6]	168[b]	Children and adults using ID services in one health district in England, UK	Staff were asked in writing to identify people with aggressive behaviour; a face-to-face interview was then conducted with the staff of each person so identified	People with serious problems like biting, kicking, scratching, etc. resulting in injury to others, e.g. bruising, bleeding and other tissue damage. Also actions, e.g. shouting/ screaming at others, or violence towards objects, presenting serious management difficulties because of threat of injury	All ages: estimated 18% had aggressive behaviour. The highest prevalence was in the group living in hospital. In day centres, prevalence was higher for men than women, but there was no gender difference in schools or in the hospital population
Borthwick-Duffy [59]	91,164	Administrative database of service users (children and adults) with ID in one state in USA	Routine annual report on adaptive behaviour and diagnoses completed by staff	More than one violent episode causing serious physical injury to others (requiring immediate medical attention) in the past year. Serious property destruction and/or minor property damage on more than six occasions within the past year	All ages: 2% physically aggressive, 7% destructive. 4% of the profound, 3% of severe, 2% of moderate and 1% of the mild ID showed aggression; 1% of females and 3% of males; 3% of non-verbal and 2% with verbal skills showed aggression. Persons destructive to property were 15% of the profound, 10% of severe, 7% of moderate and 4% of mild ID; 5% of females and 8% of males; 7% of non-verbal and 8% with verbal skills 21+: 3% were aggressive and 8% destructive
Smith et al.[a] [37]	2202	Adults (aged 20 years and over) included in an ID register for one county in England	Face-to-face interview with each person supported by their carer	Items from the DAS (aggressive behaviour rated as absent, severe and frequent {more than 3 times a week}, less severe but frequent, severe but less frequent or lesser management problem)	[d]22% physically aggressive and 17% destructive. Both aggressive behaviours were individually associated with more severe disabilities. The strength of other associations was not reported

(continued)

Table 7.1 (continued)

Authors	*N*	Population	Assessments	Diagnostic criteria	Findings (% rounded up to closest decimals)
Emerson et al. [68]	264[b] (2189)	Children and adults with ID in two Health Authority areas in England	All services supporting people with ID were asked to identify people with problem behaviour. Key informants were then asked to return a questionnaire on each person so identified	Individual schedule incorporating a measure of aggressive behaviour (serious, serious but controlled, moderate, lesser or none)	All ages: [d]7% physically aggressive
Deb et al.[a] [33]	101	Random sample of 16- to 64-year-olds with ID known to a social services department in Wales, UK	Face-to-face interview with each person supported by their carer	Items from the DAS (aggressive and destructive behaviour rated as absent, severe and frequent {more than 3 times a week}, less severe but frequent, severe but less frequent)	23% physically aggressive, 12% destructive and 29% screaming/shouting behaviour. Physical aggression: 65% were female; 22% severe, 39% moderate and 39% mild ID [30% of women and 16% of men were physically aggressive; 45% with severe, 22% with moderate and 19% with mild ID were physically aggressive]. Only taking psychotropic drugs was associated with physical aggression
Tyrer et al.[c] [54]	3065	Adults (aged 20 years and over) on an ID register in one county in England	Face-to-face interview with each person supported by their carer	Questions from the DAS. Carer report of frequency (more than 3 times per week) and/or severe physical aggression towards others	14% physically aggressive. Higher prevalence for men, younger adults, more severe ID and in institutional settings; lower prevalence for people with Down syndrome. No relationship with epilepsy or autistic symptoms
Crocker et al.[a] [5]	3165	Adults (aged 18 years and over) receiving ID services in Canada	Educators in participating agencies were sent a survey form to fill in about aggressive behaviour over the past 12 months	MOAS. verbal and/or motor behaviour towards oneself, environment or others, directly or indirectly and more or less planned. Could potentially cause physical/psychological harm to others, and may have management difficulties {0 = no behaviour, 4 = highest score of such behaviour}	52% aggressive in the previous 12 months. 24% physically aggressive, 24% aggressive to property and 38% verbally aggressive. No gender difference for physical or verbal aggression; men more aggressive to property (Mann-Whitney U). Mild/moderate ID more verbally aggressive (41% vs. 29%). Profound/severe ID more physically aggressive (32% vs. 21%) and aggressive to property. Age was negatively correlated with physical and property aggression (Spearman correlation) for men, but not women. Highest in group homes. Physical, property and verbal aggression were highly correlated

Lowe et al. [41]	901[b]; 705 adults +196 children (5395)	Children (aged 5 years or over) or adults with ID using services in a defined area, Wales	All services supporting people with ID were asked to identify people with problem behaviour. A face-to-face interview was then conducted with the primary carer of identified persons	Individual schedule incorporating a measure of aggressive behaviour (serious, serious but controlled, moderate, lesser or none)	All ages: [d]4% had serious, 2% serious but controlled, 5% moderate, 3% lesser and 86% no physical aggression; 2% had serious, 1% serious but controlled, 4% moderate, 3% lesser and 90% no destructive behaviour
Jones et al. [77] and Cooper et al.[c] [43]	1023	Adults (aged 16 years or over) receiving social care, specialist health services or known to GPs in a defined area in Scotland in UK	All identified adults with ID invited to participate. Face-to-face interviews with specialist nurse. Those potentially with problem behaviour, mental illness or ASD further assessed by a specialist psychiatrist	C21st Health check to screen for aggressive behaviour. Operationalised DC-LD definition of aggressive behaviour (excluded problem behaviour with a known cause such as ASD, mental illness or physical illness). Risk factors assessed: personal factors, lifestyle and supports, health and disabilities	[g]Point prevalence: 23% with problem behaviour (including SIB; psychiatrist's opinion); 18.7% with problem behaviour (including SIB; DC-LD). [g]7% verbally aggressive, 3% property destruction and 6% physically aggressive to others. [h]10% with outwardly directed aggression. [g]Higher prevalence of problem behaviour (including SIB) for females, lower ability level, living in a congregate care setting, or with paid carer support (rather than living with a family carer), having ADHD, urinary incontinence, visual impairment, not having Down syndrome and not having severe physical disabilities. [h]Higher prevalence of outwardly directed aggression for men, more severe ID, not living with family, presence of ADHD, absence of Down syndrome and presence of urinary incontinence
Lundqvist et al.[c] [36]	915	Adults (aged 18 years and over) receiving care from Local Health Authorities in a county in Sweden	Staff members judged to be in a good position to provide information about each participant were interviewed by country council workers, specialising in working with adults with ID and BPs using an inventory. Data collection took place at daily activity centres and community homes	BPI to determine presence and severity of aggressive destructive behaviour. Risk factors assessed: Personal factors, disability/disorder, body functions, social activity/participation and services/treatment factors	62% with problem behaviour (including SIB); 18.7% identified as having challenging behaviour (with at least one behaviour rated as severe). 30.4% with aggressive destructive behaviour and 11.9% with severe aggressive destructive behaviour. Higher prevalence for women, older ages, presence of autism, longer night sleep duration (negative correlation), presence of auditory hypersensitivity, using signs to communicate, absence of group functioning, presence of initiation of social interaction and in contact with psychiatry. These variables accounted for 23% of the variance in aggressive destructive behaviour

(continued)

Table 7.1 (continued)

Authors	*N*	Population	Assessments	Diagnostic criteria	Findings (% rounded up to closest decimals)
Tsiouris et al.[c] [24]	4069 (9894)	Adults (ages 18 years and over) receiving care services in New York State in USA	Anonymised data collected by chief psychologists from information in participants' files	Modified version of MOAS to determine frequency of verbal aggression towards others, verbal aggression towards self, aggression towards objects and physical aggression towards others, each in the last year. Risk factors assessed: basic demographics and psychiatric conditions	83% with aggression (including SIB) Frequency of verbal aggression towards others was independently associated with less severe ID, older age (positive correlation) and presence of depression, bipolar disorder, psychosis, impulse control disorder and personality disorder Frequency of verbal aggression towards self independently associated with less severe ID, younger age (negative correlation), female gender and presence of anxiety, depression, bipolar disorder, psychosis, impulse control disorder and personality disorder Frequency of physical aggression towards others independently associated with more severe ID, younger ages (negative correlation), male gender and presence of autism, anxiety disorder, bipolar disorder, psychosis, impulse control disorder and personality disorder Frequency of physical aggression towards objects independently associated with less severe ID, younger ages, male gender and presence autism, anxiety disorder, bipolar disorder, OCD, psychosis, impulse disorder and personality disorder

Adapted from Cooper et al. [43]

ID intellectual disabilities, *ASD* autism spectrum disorder, *OCD* obsessive compulsive disorder, *SIB* self-injurious behaviour, *ADHD* attention deficit hyperactivity disorder, *DAS* Disability Assessment Schedule [63], *ABS* Adaptive Behaviour Scales, *MOAS* Modified Overt Aggression Scale, *DC-LD* Diagnostic Criteria-Learning Disability, *PAS-ADD* Psychiatric Assessment Schedule for Adults with Developmental Disabilities, *BPI* Behaviour Problem Inventory.

[a]Study using Chi-square analysis to model behaviour from multiple variables

[b]The number of persons on whom data were collected, that is, who had problem behaviour (the estimated denominator of number of persons with ID in the population)

[c]Study using multiple regression analysis to model behaviour from multiple variables

[d]Prevalence not reported in the paper, but calculated on the basis of other data presented

[e]From Qureshi and Alborz [128]

[f]From Emerson [68]

[g]From Jones et al. [77]

[h]From Cooper et al. [43]

Table 7.2 Prevalence of and factors associated with SIB in community studies of adults with ID

Authors	*N*	Population	Assessments	Diagnostic criteria	Findings (% rounded up to close decimals)
Eyman and Call[a] [127]	5243	Administrative database of service users with ID aged 13 and over, USA	Selected items of the ABS were completed by direct-care workers and social workers	SIB rated as occurring frequently or occasionally	13+: [d]24% had SIB, ranging from 5% of mild-moderate ID living with relatives to 47% of profound ID in hospitals. People with SIB were older, Caucasian rather than Mexican American, of lower ability and in institutions. Equal gender distribution
Jacobson [38]	32,112	Administrative database of service users (children and adults) with ID in one state, USA	42-item questionnaire routinely completed by staff	The general frequency of up to three problem behaviours could be reported	All ages: 8% had SIB Aged 22+: 11% with 'psychiatric and developmental disabilities' had SIB and 9% with developmental disabilities only had SIB. Of adults with SIB, 18% profound, 6% severe, 3% moderate and 3% mild ID
Kebbon and Windahl [129]	28,215	Services for people with ID in 22 of 25 counties, Sweden	Staff completed a structured questionnaire	SIB including an overt motor component, such as head hitting, head banging, hitting other parts of the body, hair pulling and scratching; at frequency of daily, weekly, monthly or once or twice in 3 months	All ages: 4% had SIB; 11% in institutions, 2% in integrated accommodation. People with SIB were 46% female [compared with 44% female for whole ID population], 40% profound, 48% severe, 12% moderate and 1% mild ID [13% with profound, 7% severe, 1% moderate and 1% mild ID have SIB]. Highest frequency of SIB at ages 22–51
Rojahn [130]	431[b] (25,872)	Children and adults with ID using 294 services (excluding hospitals), in Germany	Postal survey for staff to complete on persons with SIB; second postal survey completed on three users of the same service who did not have SIB	SIB that causes (or potentially causes) damage; 15 topographies, with 3-point severity scale and 4-point frequency scale	All ages: 2% had SIB; 7% in schools/training centres, 2% in sheltered workshops and 8% in group homes. Of people with SIB, 48% female, 16% profound, 43% severe, 28% moderate and 13% mild ID

(continued)

Table 7.2 (continued)

Authors	*N*	Population	Assessments	Diagnostic criteria	Findings (% rounded up to close decimals)
Oliver et al. [131]	596[b] (not reported)	All service users (children and adults) with ID in one health region, England	Staff/carers were asked in writing to identify people with SIB; a face-to-face screening interview was then conducted with the staff of each identified person	Repeated, self-inflicted, non-accidental injury, producing bruising/bleeding/other tissue damage + such SIB prevented by protective measures/interventions	All ages: People with SIB were 42% female; 40% profound, 49% severe and 12% mild ID Adults: 3% who used a social education centre had SIB; 12% in a long-stay hospital had SIB
Qureshi and Alborz [128], Emerson et al. [68]	695[b] (4200)	Children and adults using ID services, in seven Health Authority areas, England	All services supporting people with ID were asked to identify people with challenging behaviour. Key informants were then asked to return a questionnaire on each person they had so identified	Interview with staff for each person who had been identified as having challenging behaviour, on its type, intensity and frequency	All ages: [e,d]8% had SIB, or [f,d]4% had serious/serious but controlled SIB
Borthwick-Duffy [59]	91,164	Administrative database of service users (children and adults) with ID in one state, USA	Routine annual report on adaptive behaviour and diagnoses completed by staff	Client Development Evaluation Report (1. severe SIB requiring medical attention once per month and/or minor SIB requiring first aid weekly, or 2. weekly SIB)	All ages: 2% had type 1 SIB and 9% had type 2 SIB. Of persons with SIB, 25% profound, 15% severe, 7% moderate and 3% mild ID; 9% females and 10% males; 17% non-verbal and 9% with verbal skills had frequent SIB
Hillery and Mulcahy[a] [132]	429	IQ < 50, living in a single community care area (children and adults), Ireland	Carers completed a record sheet daily for 4 weeks, indicating whether or not SIB had occurred	Self-injurious and non-accidental behaviour, producing bruising, bleeding or other tissue damage, + such SIB prevented by protective devices or restraints	All ages: 62 (14%) have SIB; of whom 42% female (NS); 5% profound, 53% severe and 42% moderate ID; no association between age and SIB (t-test) or accommodation type and SIB

Collacott et al.[c] [75]	2101	Adults (aged 20 years and over) with ID in a defined geographical area, England	Face-to-face interview with each person supported by their carer	Items from the DAS (SIB rated as absent, severe and frequent {more than 3 times a week}, less severe but frequent, severe but less frequent or lesser management problem)	17% had SIB + 3% with past history of SIB in remission; SIB was severe and frequent for 2%, frequent but less severe for 3%, severe but less frequent for 3% and lesser management problem for 9%. Lower age, lower ability, impaired hearing, impaired mobility and 'number of autistic symptoms' were independently associated with SIB, but epilepsy, gender and impaired vision were not
Emerson et al. [68]	264[b] (2189)	Children and adults with ID in two Health Authority areas, England	All services supporting people with ID were asked to identify people with challenging behaviour. Key informants were then asked to return a questionnaire on each person they had so identified	Individual schedule incorporating a measure of SIB (SIB defined as serious, serious but controlled, moderate, lesser or none)	All ages: [d]3% had SIB
Deb et al.[a] [33]	101	Random sample of 16–64-year olds with ID known to a social services department, Wales	Face-to-face interview with each person supported by their carer	Items from the DAS (SIB rated as absent, severe and frequent {more than 3 times a week}, less severe but frequent and severe but less frequent. 'Lesser management problem' was omitted)	24% have SIB; of whom 67% were female; 33% severe, 33% moderate, 33% mild ID [36% of women and 12% of men have severe and/or frequent SIB; 73% of sample with severe, 19% with moderate and 17% with mild ID have SIB]. Female gender, more severe ID, impaired communication skills and psychotropic drugs were associated with SIB
Lowe et al. [41]	901[b]; 705 adults +196 children (5395)	Children (aged 5 years or over) or adults with ID using services in a defined area, Wales	All services supporting people with ID were asked to identify people with challenging behaviour. A face-to-face interview was then conducted with the primary carer of each identified person	Individual schedule incorporating a measure of SIB previously designed by Alborz et al. [64] (SIB defined as serious, serious but controlled, moderate, lesser or none)	All ages: [d]3% have serious, 1% serious but controlled, 4% moderate, 3% lesser, 90% no SIB

(continued)

Table 7.2 (continued)

Authors	*N*	Population	Assessments	Diagnostic criteria	Findings (% rounded up to close decimals)
Jones et al.[c] [77], Cooper et al.[c] [42]	1023	Adults (aged 16 years or over) receiving social care, specialist health services or known to GPs in a defined area in Scotland in UK	All identified adults with ID invited to participate. Face-to-face interviews with specialist nurse. Those potentially with problem behaviour, mental illness or ASD further assessed by a specialist psychiatrist	C21st Health check to screen for aggressive behaviour. Operationalised DC-LD definition of aggressive behaviour (excluded problem behaviour with a known cause such as ASD, mental illness or physical illness) Risk factors assessed: personal factors, lifestyle and supports, health and disabilities	[g]5% with SIB. [h]Higher prevalence for more severe ID, not living with family, presence of ADHD, absence of Down syndrome and presence of visual impairment
Danquah et al.[c] [70]	94	Adults (aged 18 years or over) receiving ID services in Manchester in UK	Information was gathered on each participant using a purpose designed, structured interview with service managers	Risk factors assessed: opiate release/topography related, communication, medical/ diagnoses-related behaviours, boredom/frustration and habitual	Higher continued prevalence (after 2–4 years) for those with self-biting and lower levels of verbal ability
Tsiouris et al.[c] [24]	4069 (9894)	Adults (ages 18 years and over) receiving care services in New York State in USA	Anonymised data collected by chief psychologists from information in participants' files	Modified version of MOAS to determine frequency of verbal aggression towards others, verbal aggression towards self, aggression towards objects and physical aggression towards others, each in the last year. Risk factors assessed: basic demographics and psychiatric conditions	83% with aggression (including SIB). Frequency of SIB independently associated with more severe ID, younger ages (negative correlation) and presence of autism, anxiety disorder, bipolar disorder, psychosis, impulse control disorder and personality disorder

Lundqvist et al.[c] [36]	915	Adults (aged 18 years and over) receiving care from Local Health Authorities in a county in Sweden	Staff members judged to be in a good position to provide information about each participant were interviewed by country council workers, specialising in working with adults with ID and BPs using an inventory. Data collection took place at daily activity centres and community homes	BPI to determine presence and severity of SIB Risk factors assessed: Personal factors, disability/disorder, body functions, social activity/participation and services/treatment factors	30.9% with SIB and 8.4% with at least one SIB rated as severe. Higher prevalence for more severe ID, presence of autism, absence of Down syndrome, presence of night-time sleep disturbances, presence of tactile hypersensitivity, presence of communicating in pictures and taking psychotropic medication. These variables accounted for 24% of the variance in SIB

Adapted from Cooper et al. [42]

ID intellectual disabilities, *ASD* autism spectrum disorder, *OCD* obsessive compulsive disorder, *SIB* self-injurious behaviour, *ADHD* attention deficit hyperactivity disorder, *DAS* Disability Assessment Schedule, *ABS* Adaptive Behaviour Scale, *DC-LD* Diagnostic Criteria-Learning Disability, *PAS-ADD* Psychiatric Assessment Schedule for Adults with Developmental Disabilities, *BPI* Behaviour Problem Inventory

[a]Study using Chi-square analysis to model behaviour from multiple variables

[b]The number of persons on whom data were collected, that is, who had challenging behaviour (the estimated denominator of number of persons with intellectual disabilities in the population)

[c]Study using multiple regression analysis to model behaviour from multiple variables

[d]Prevalence not reported in the paper, but calculated on the basis of other data presented

[e]From Qureshi and Alborz [128]

[f]From Emerson [68]

[g]From Jones et al. [77]

[h]From Cooper et al. [42]

cross-sectional or retrospective case studies. AB is commonly considered to increase with age until mid-adulthood [65]; however, studies have reported mixed results in terms of an overall relationship with age [34, 66]. Some studies did not find any associations with age, most consistently with SIB [36, 42, 43, 67]. The difference among studies may be explained by a shift in topography with age. For example, the severity of verbal aggression towards others increased in the study by Tsiouris and colleagues [24], whereas all other types decreased with age. Tyrer and colleagues [54] reported a decreased prevalence of physical aggression towards others with age. Lundqvist [36] reported decreasing prevalence with age for outwardly directed aggression with a second peak among those aged 70 or older.

7.6.2 Gender

It is a commonly held assertion that AB is associated with the male gender [68], although some studies have reported SIB to be more common in females [5, 33]. Crocker and colleagues [5] highlight from their study of individual associations that SIB was also more severe in those with more severe ID and more females have a severe ID [21, 66] which could act as a confounder, accentuating the actual gender difference. This highlights the importance of controlling for other variables through multivariate analyses. Three studies did not locate any gender associations with SIB, however, females were more likely to exhibit outwardly directed aggression (including verbal aggression) [36, 42, 43]. Drieschner and colleagues [67] report more severe total and individual types of aggression in females in inpatient treatment facilities. This is in contrast to other studies that have reported more severe/prevalent aggression in males, including a meta-analysis [5, 6, 24, 54, 59, 62].

7.6.3 Severity of Intellectual Impairment

Increasing severity of ID has commonly been associated with PB [5, 33, 59]. Tsiouris and colleagues [24] reported increased frequency of AB associated with increased severity of ID for two types of AB, namely physical aggression towards others (Tyrer and colleagues also reported this relationship [54]) and SIB, whereas verbal aggression and property destruction demonstrated the opposite effect. Cooper and colleagues [42, 43] also found SIB and outwardly directed aggression to be more common among those with more severe ID. Lundqvist [36] reported the same in relation to SIB only. Aman and colleagues [69] did not find any relationship with severity of overall aggression. However, Emerson and colleagues [68] reported an association between mild ID and verbal aggression, meanness and cruelty, and between severe ID and physical aggression and temper tantrums.

7.6.4 Verbal Communication

Lower levels of language skill have been associated with a higher prevalence and severity of aggressive behaviours, especially SIB [5, 33, 41, 66, 70, 71]. A meta-analysis and systematic review also assert that SIB is associated with lower levels of expressive or receptive verbal communication [26, 62], with no association with aggression towards others [62]. However, other studies have failed to locate individual associations when accounting for potentially confounding variables [42, 43].

7.6.5 Physical Health

Increased prevalence of PB has been linked to various physical conditions, although they remain rarely studied. An early study found that individuals with ID and poor physical health were more likely to exhibit PB than those with better health, regardless of health issue [72]. A systematic review synthesised evidence published between 1990 and 2008 on the relationship between physical health problems and challenging behaviour in people with ID [73]. Eleven well-conducted studies were reviewed that reported significant and independent associations between pain related to cerebral palsy, chronic sleep problems, uri-

nary incontinence and visual impairment. Results were also suggestive of associations with gastrointestinal problems, menstrual cycle and sub-types of epilepsy. However, the authors commented that for some physical health conditions, no articles were identified. Furthermore, as the majority of studies were cross-sectional or retrospective, the authors specified a need for further longitudinal research that can delineate cause and effect. Other authors have reported few associations with physical health conditions [33, 42, 43, 74]. Some have reported associations with visual or hearing impairments [6, 42, 75], immobility [76] and urinary incontinence [43] and found mobility problems to be associated with the presence of SIB [68, 71, 74]. On the other hand, it seems that PB is less common in those with physical disability [77].

PB could be associated with pain, which is often under-recognised in people with ID, largely due to communication difficulties and diagnostic overshadowing [78, 79]. This, along with the increased prevalence of health conditions [73, 78, 80], may indicate that PB is the manifestation of an uncomfortable/aberrant internal state, with pain reducing the capacity for adaptive behaviour [81]. Chronic pain and behavioural expressions of pain have been linked to increased levels of AB in adults with ID [79, 82]. Furthermore, some authors have suggested that pain regulation may be disturbed in some people with ID [3]. For example, ASD, which has a high co-morbidity with ID [83], has been associated with sensory processing abnormalities, including hypersensitivity and hyposensitivity to pain [84].

7.6.6 Psychiatric Disorders

The extent of overlap between mental health problems and PB has been a matter of ongoing debate. There have been many studies showing that psychiatric morbidity including anxiety disorders, psychotic disorders and mood disorders among people with ID is associated with higher levels of PB [85, 86, 87]. Conversely, other studies have not found any such association [41, 66, 88]. It is now accepted by the majority that mental health and PB interplay in complex ways and that there are multiple reasons for PB with mental illness being only one of many possible causes or outcomes. It is likely that mental health problems, when defined as diagnosable mental disorders from standard classification systems, do not play a major role in the vast majority of PB. However, it is worth keeping in mind that in many prevalence studies PB is classified as a psychiatric disorder. The previous labelling of PB as the so-called behavioural 'equivalents' of psychiatric symptoms is not supported by the evidence [88–91]. Perhaps it would be better to describe PB as sometimes being behavioural 'correlates' of psychiatric symptoms in people with ID [34, 66, 89, 92] (for further description of the concept of behavioural equivalence and the cases in which it can be considered as such refers to ▶ Chap. 5).

There are obvious difficulties in diagnosing psychiatric disorders in people with ID, especially those with severe and profound ID, limited communication skills and co-morbid ASD [21, 33, 66], which may lead to both under-diagnosis and over-diagnosis of psychiatric illness in people with ID, in general, and ID and PB, in particular. The under-diagnosis may happen because of the so-called 'diagnostic/behavioural overshadowing', whereby the behaviour is perceived as part of the developmental disability repertoire rather than a symptom of an underlying psychiatric illness [33, 66, 93]. Over-diagnosis may also occur, influenced by co-morbid physical health problems, lack of knowledge of carers, medication adverse effects [33, 66], health insurance systems or used as a justification for psychotropic medication prescription. This has led to wide-ranging prevalence estimates [94] and difficulties in consistently identifying psychiatric risk factors. For example, some people with ID may look as if they are hallucinating when they speak to themselves, speak or look at an imaginary person, speak to an object such as a tree or a table or talk about an imaginary world/friend, whereas, in fact, these are manifestations of neurodevelopmental disorders such as ASD and/or ID.

Among studies using multiple regression analyses, varied results relating to psychiatric conditions were reported. Cooper and colleagues [42, 43] did not locate any association other than with ADHD. Similarly, Drieschner and colleagues [67] only reported an association between ADHD/disruptive behaviour disorder and frequency of aggression, while Lundqvist [36] and Tyrer and colleagues [54] reported no independent associations. Conversely, Tsiouris and colleagues [24] who specifically studied the relationships between aggressive behaviour and psychiatric conditions reported independent associations for all psychiatric conditions under investigation.

Melville and colleagues [95] argued that identifying dimensions of psychopathology has a greater validity than diagnoses of psychiatric disorder using a categorical classification system in the general population. In a subsequent paper, Melville and colleagues [88] using an exploratory and confirmatory factor analysis of psychopathology in two large samples of adults with ID identified PB within an emotion dysregulation problem behaviour dimension that was distinct from the depressive, anxiety, organic and psychosis dimensions. The authors found that the dimensional model had better predictive validity than a categorical diagnosis.

7.6.7 Neurodevelopmental Disorders (ASD and ADHD)

Both ASD and ADHD are common comorbidities of ID [83, 96–99] and both ASD and ADHD have commonly been implicated as a risk factor for PB [100, 101]. Tsiouris and colleagues [24] report independent associations between diagnosis of autism, and physical aggression against others and property destruction. Conversely, others have reported no relationships with outwardly directed aggression [36, 43, 54, 67]. Coe and Matson [102, 103] found a higher rate of some PB in children with ADHD and ID when compared with children with ID who did not have ADHD. Similarly, Melville and colleagues [98] found a higher rate of PB in people with ID and autism compared with those who have just ID but no autism. However, this difference no longer existed once the groups were individually matched.

7.6.8 Epilepsy

Mental health symptoms could be observed in the pre-ictal, ictal, post-ictal and inter-ictal phases. In certain types of seizures, particularly associated with complex partial seizures, certain psychological and behavioural symptoms could be manifested during the 'ictal' phase. The inter-ictal psychopathology is the subject of research over many years and the findings in this area to some extent still remain controversial [104]. There have been six controlled studies of PB and psychopathology among adults with ID and with and without epilepsy [66, 105]. Overall, no significant difference is observed in the rate of PB between epilepsy and the non-epilepsy group. Further, non-controlled studies have supported these findings [66, 105]. Deb and Hunter [106] reported a significantly higher rate of PB among the subgroups of individuals with epilepsy who showed generalised epileptiform EEG changes, who received one anti-epileptic drug at a time and those who received carbamazepine. Some studies have shown an increased rate of psychopathology, in general, but not necessarily PB among children with epilepsy in the general population and also with ID [107, 108]. However, some have shown that PB started in children before the onset of epilepsy [109].

A recent systematic review and meta-analysis has found no association between epilepsy and PB in adults with ID, and hypothesised that although multi-factorial in eatiology, the underlying brain damage in the case of severe ID and psychosocial advantages in the case of mild ID are the stronger determinants of parapsychology in this population than the epilepsy *per se* [110, 111].

Factors that affect behaviour in the presence of epilepsy are related to (a) an underlying brain damage such as the location and severity of any deformity or space occupying lesion or abnormal electrical discharge in the brain; (b) epilepsy-related factors such as certain epileptic syndromes are prone to develop more PB; (c) seizure-related factors such as the severity,

type and frequency of seizures; (d) anti-epileptic–related factors such as the adverse effects of certain anti-epileptic medicine such as topiramate, levetiracetam and perampanel and (e) psychosocial factors such as loss of occupation, financial problems, lack of support, locus of control being outside the person so the person does not have any control over when the seizure is going to occur, etc. [105]

7.6.9 Life Events

In a systematic review, Wigham and colleagues [112] summarised findings from 15 studies. There is also a further publication from the same authors on this subject [113]. The majority of studies reported an association between life events and PB. The types of life events described in the summarised studies included sexual and other abuses, bereavement, and other life events. The description of PB included aggression, SIB, irritability, 'acting out', hyperactivity, destructive behaviour, conduct disorders, adjustment disorders, stereotypical behaviour and inappropriate speech.

7.7 Environmental/Contextual Risk Factors for Problem Behaviour

Carr [114] categorised functions of PB in terms of social positive reinforcement, social negative reinforcement and sensory (i.e. automatic) reinforcement. Environmental factors for PB have most commonly been studied through functional assessment [115], which seeks to identify relationships between antecedents and reinforcing consequences [116] and to identify contextual variables and events that elicit PB [117]. Emerson and Einfeld [3] included these features in their operant model for PB (see Figs. 7.1 and 7.2). Researchers have suggested that people with ID and subsequent limited communication skills 'rely primarily on expressive behaviour to communicate their wants and needs…these limited skills can lead to communication in the form of… behaviour' [1, 16, 114].

7.8 Behavioural Phenotypes

Certain genetic syndromes that cause ID are also shown to be strongly associated with certain features of PB and SIB [118, 119]. For example, Cri du Chat, Cornelia de Lange, Angelman, Fragile X, Prader-Willi, Lowe, Smith Magenis, Lesch-Nyhan, Rett and Brachmann de-Lange syndromes are known to carry a higher risk for certain types of behaviours such as physical aggression and SIB [27, 119, 120]. Conversely, research has demonstrated that those with Down syndrome are less likely to exhibit physically aggressive behaviour [54, 76]. In Table 7.3, we have presented both physical and behavioural phenotypes of some of the relevant syndromes.

7.9 Assessment of Problem Behaviour

In this section, we present a bio-psycho-social assessment model for PB. Important aspects include an assessment of the (a) behaviour, (b) person showing the behaviour, (c) cause(s) of the behaviour, (d) reaction to and consequences of the behaviour and (e) environment and the risks involved with the behaviour (see Comprehensive Assessment of Triggers for behaviours of concern Scale; [121] ▶ https://spectrom.wixsite.com/project).

Using this model, the assessment may be carried out under four broad headings, namely *B*ehaviour, *M*edical, the Person, *P*sychological/Psychiatric and *S*ocial (*BMPPS*) (see Table 7.4). The importance of this schema is that it informs the intervention plan to reduce the PB, and to treat other conditions that might be presenting as PBs but actually need specific treatments to correct or ameliorate them.

An important area of assessment of PB includes the assessment of the individual themselves. This should include an assessment of (a) their strengths and abilities, opportunities and resources available to them; (b) their mental and physical health needs, educational/vocational needs and impact of disabilities, service and resource gaps in their lives; (c) their likes, dislikes and preferences and how they express these; (d) their history such as social, developmental, psy-

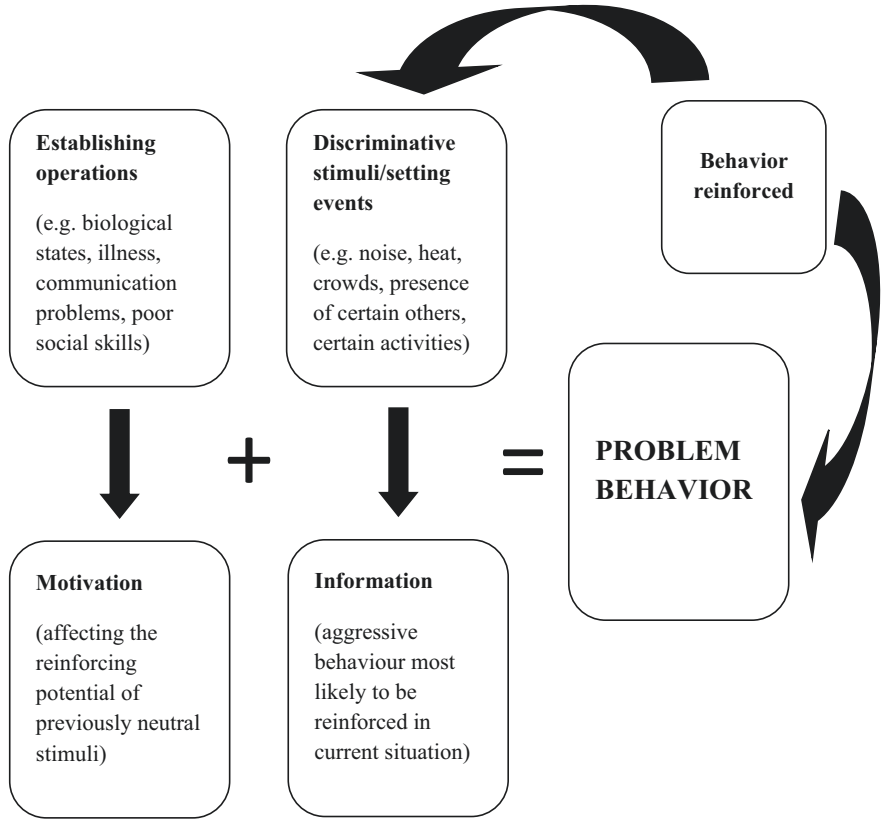

Fig. 7.1 Schematic representation of the Operant model of aggressive behaviour. (Adapted from Emerson and Einfeld [3])

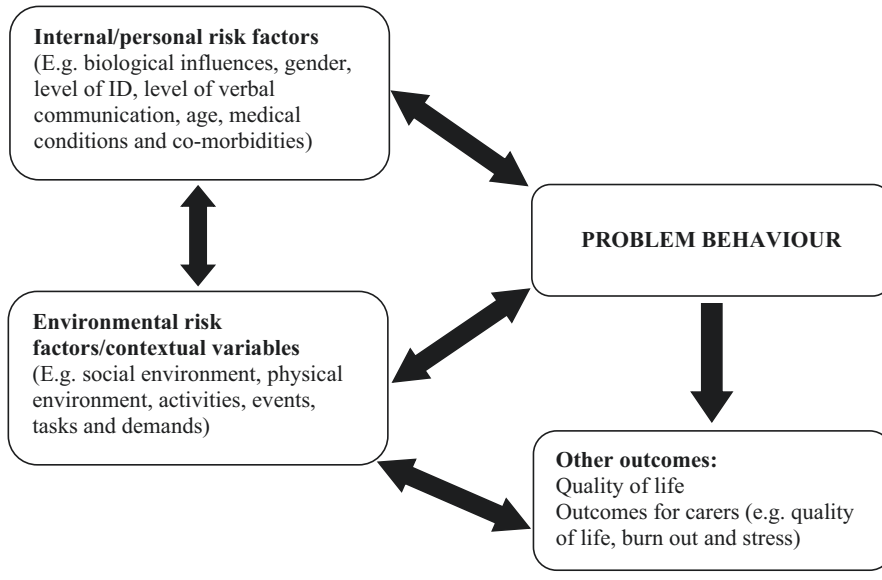

Fig. 7.2 Schema for problem behaviour in intellectual disabilities

Table 7.3 Physical and behavioural phenotypes of common genetic syndrome associated with intellectual disabilities [118]

Name of the syndrome	Genetic phenotype	Physical phenotype	Behavioural phenotype
Down's syndrome	Trisomy 21 (95%); Translocation (4%); Mosaic (1%); 1:600–1000 live births (average incidence); 1:1500 (maternal age <30); 1:800 (maternal age 30–34) and 1:100 (maternal age 40–44)	Short stature; small head; brachycephaly; high arched palate; eyes: upward, outward slant and epicanthic fold; protruded tongue; eyes: nystagmus, cataract and Brushfield spots; simian (single palmar) crease; fingers: syndactyly and toes: sandal gap	Moderate learning disability; usually of friendly nature but some may show problem behaviour; dementia is very common with early age of onset
Fragile X/ ataxia syndrome (FRAXT) (also FRAXA and FRAXE)	1 in 4000 men; 1 in 6000 women; Xq27.3; CGG repeat (up to 50 is normal; 43–200: carrier); CGG trinucleotide expansion; (>200: Fragile X syndrome) (90%); Fragile X Mental Retardation-1 (FMR-1 gene); absent/ low FMR protein (FMRP)	Subtle physical features and usually do not appear before adulthood; macro-orchidism (large testes); long face; long ear; prominent jaw; connective tissue disorders; epilepsy (25%; usually appears in adolescence) and cardiac abnormalities (mitral valve prolapse, post-ductal coarctation, aortic hypoplasia and dilatation)	Mild-to-moderate ID (30% severe ID); delay/abnormal language development; dysfunction in social play; verbal communication (speech volume, word/ phrase perseveration); non-verbal communication (gaze aversion); repetitive motor behaviour (hand flapping, hand biting and rocking); ADHD and ASD features; poor mathematical skills; abnormal sensitivity to external stimulus; mouthing/ smelling of objects; aggression (20–30%); hyperarousal and over activity
Foetal alcohol spectrum disorder (FASD)	0.2–3 per 100 live births; 10–20% mild ID; 8 or > drinks per day; bingeing/ poly drug use; newborn babies show features of alcohol withdrawal; microcephaly and epicanthic fold	Short palpebral fissure; optic nerve hypoplasia with poor visual acuity; hearing loss; ventricular septal defect (VSD), renal hypoplasia and bladder diverticula	Hyperactivity; sleep disturbance and language impairment
Rett syndrome	1 in 10,000–15,000; only girls are affected; gene at Xq28 (MECP2)	Develop normally up to age 18 months to 2 years; starts showing deterioration; epilepsy (59–72%); limb spasticity from age 5; a few years plateauing, further deterioration, increased spasticity/rigidity lower limb; poor balance, tremor and wide-based gait	Severe and profound ID; abnormal motor movement (hand flapping/wringing) and autistic features

(continued)

Table 7.3 (continued)

Name of the syndrome	Genetic phenotype	Physical phenotype	Behavioural phenotype
Prader-Willi syndrome	1.2–1.3 per 10,000; boy: girl:1.6:1.2; deletion of chromosome 15 (q11q13) of paternal origin (50–70% cases); FISH test; maternal disomy in chromosome 15; (some cases)	Obesity; diabetes; cryptorchidism; hypogenitalism and duodenal ulcer/ renal stone	Moderate ID; hyperphagia (excessive eating); sleep disturbances (excessive daytime sleepiness); psychotic features and skin picking
Lesch-Nyhan syndrome	X-linked recessive condition (Xq26q27); 1 in 10,000–380,000; inborn error of xanthine metabolism	Poor prognosis; spasticity and epilepsy	Severe/ profound ID; severe self-injurious behaviour common
Phenylketonuria	1 in 5000–10,000 live births; autosomal recessive (gene 12q22–24); detected by routine heel prick test of newborn babies and dietary treatment from infancy	Fair skin, blonde hair and blue eyes; seizure, eczema; tremor and musty odour urine (untreated cases)	Poor attention, hyperactive and aggression (untreated cases)
Velocardiofacial syndrome	Also known as Shprintzen/ Di George/ Cayler/ Takao/ 22q11-deletion/ CATCH 22 syndromes; autosomal dominant; 1 in 5000; majority microdeletion at 22q11.2	Cleft palate; cardiac abnormalities (ventricular septal defect); pulmonary stenosis; facial dysmorphology; seizures; hypocalcaemia	Common in late life: psychoses (schizophrenia); bipolar disorder; social withdrawal; anxiety
Angelman syndrome	Also known as happy puppet syndrome; 1:20–30,000; deletion of chromosome 15 (of maternal origin; as opposed to Prader-Willi syndrome, which is of paternal origin); 15q11-q13 (60–75% cases)	Severe epilepsy (86%); microcephaly (small skull); characteristic EEG pattern; blue eyes and blonde hair in two-thirds of cases; waddling gait (ataxia); prognathism	Severe ID; characteristic laughter; typically sociable and affectionate; over excitability; over activity and short attention span; get better with age
Williams syndrome	1 in 15,000 infants; autosomal dominant; elastin gene at 7q11.3	Idiopathic infantile hypercalcaemia; treat with a low-calcium diet and vitamin D restriction; 'Elfin-like' face (prominent cheeks, wide mouth and flat nasal bridge); kidney and heart lesions (supravalvular aortic stenosis and peripheral pulmonary artery stenosis); growth is usually retarded; life expectancy is related to metabolic and heart abnormalities; abnormally sensitive hearing	Moderate or severe ID; social disinhibition with abnormal friendliness to strangers; over activity; poor concentration; eating and sleeping abnormalities; abnormal anxiety; poor peer relationships; aggression (5–10% children) gets better with age

Table 7.3 (continued)

Name of the syndrome	Genetic phenotype	Physical phenotype	Behavioural phenotype
Rubenstein-Taybi syndrome	1 in 125,000; microdeletions at 16p13.3 (in some)	Short stature; small head; beaked or straight nose; downward slanting eyes; stiff gait; congenital heart defects; urinary tract abnormalities; severe constipation	Moderate ID; reduced attention span; rocking; spinning; hand flapping; friendly disposition; self-stimulatory activities such as rocking; intolerance of loud noises
Smith-Magenis syndrome	1 in 50,000; deletions at 17p11.2	Flattened mid-face; abnormally shaped upper lip; short hands and feet; single transverse palmar crease; abnormally shaped or placed ears and sometimes a high arched palate or protruding tongue; coarse facial features; otitis media and squint; speech delay is more pronounced than delay in motor achievement	Moderate ID is common; self-hugging and mid-line hand clapping; sleep disorders are common with some children waking repeatedly in a state of agitation; absence of rapid eye movement (REM) sleep
Smith-Lemli-Opitz syndrome	1 in 30,000; mildly affected people may be undiagnosed; one of the commonest autosomal recessive conditions; gene located at 11q12–13; deficiency of the enzyme 7-dehydrocholesterol reductase results in elevated levels of a cholesterol precursor	Small head; drooping eyelids; squint; forward-facing nostrils; small lower jaw; finger abnormalities such as extra fingers (polydactyly) and syndactyly (webbed fingers)	Intelligence varies from normal to severe ID; aggressive and self-injurious behaviours and autistic spectrum disorders
Tuberous Sclerosis Complex (TSC)	Autosomal dominant; 1 in 7000 newborns; chromosome 9 (9q34-TSC1): 50%; Chromosome 16 (16p13-TSC2): 50%; TSC2 is close to the gene for the adult polycystic kidney disorder (APKD-1)	Multi-system disorder; hamartomas in brain, retina, heart, lungs and kidney (skull X-ray will show calcified tubers); subependymal nodules in the brain (shown in MRI); adenoma sebaceum (angiofibroma and facial butterfly rash); depigmented skin area (hypomelanic macules) (best seen under the UV light); other skin lesions (Shagreen patches); 65–80% epilepsy; periungual fibroma; polycystic kidney in some	Learning disability (50%); language delay; restlessness; aggression; self-injurious behaviour; ASD/ADHD (up to 50%); also, unipolar or bipolar depression; anxiety

(continued)

Table 7.3 (continued)

Name of the syndrome	Genetic phenotype	Physical phenotype	Behavioural phenotype
Cornelia de Lange syndrome	1 in 60,000 live births; Nipped B-like or NIPBL gene (chromosome 5) (40%)	Growth retardation; distinctive facial features consisting of well-defined arched eyebrows which meet in the middle; long curled eyelashes; small nose with forward-facing nostrils and down-turned mouth with thin lips; limb abnormalities such as small or shortened limbs, especially arms; hearing impairments; gut malformations; congenital heart defects; early mortality is high because of feeding problems with regurgitation and vomiting leading to aspiration pneumonia in some cases	The degree of ID is usually severe, and speech is often very limited; however, some affected people have IQs within the normal range; self-injury, autistic features and pleasurable responses to vestibular stimulation, e.g. spinning in a chair have been reported as part of the behavioural repertoire; compulsive behaviour; self-injurious behaviour (not supported by recent studies)
XO (Turner's syndrome)	1 in 10,000 live female births; 45X karyotype (in some)	Short stature; ovarian failure; puberty does not usually occur naturally; webbed neck; low hairline; widely spaced nipples; multiple pigmented naevi; coarctation of the aorta or a ventricular septal defect (12%)	Usually of normal intelligence; hyperactivity and distractibility are common in childhood; poor social skills, with immature social relationships and low self-esteem in adolescence; specific cognitive abnormalities including deficits in spatial perception, visual motor integration, affect recognition, visual memory and attention
XYY syndrome	1 in 1000 live male births; 10% have mosaic 46, XY/47, XYY	Most are over 10 cm taller than their fathers as adults; sexual development and fertility are unaffected	Lower mean intelligence scores (with a large overlap with the normal range); poor social adaptation; distractibility; hyperactivity; temper tantrums; speech and language problems; little evidence to suggest a significant link with seriously aggressive criminal conduct in adult life

Table 7.4 Bio-psycho-social (*BMPPS*) model of assessment of problem behaviour in people with intellectual disability

Behaviour (*B*)	See list in ► Sect. 7.9.1
Medical and organic factors (*M*) (See also ► Sect. 7.9.2)	Physical symptoms (toothache, tummy ache, heartburn, headache, etc.) Medical conditions (cardiovascular, respiratory, endocrine, gastrointestinal, musculoskeletal, dental, skin and genito-urinary) Physical disabilities Problem with sleep, appetite, weight, bowel and bladder Epilepsy and other neurological conditions (spasticity, movement disorders, multiple sclerosis, brain tumour, etc.) Genetic conditions (Lesch-Nyhan syndrome, Prader-Willi syndrome, Fragile X syndrome, Smith-Magenis syndrome, etc.) Sensory impairment Communication/speech problems Drug- and alcohol-related factors Current medication, previous medication, polypharmacy and high-dose medication use and adverse effects including anti-cholinergic burden
Relevant histories (*Person*)	Developmental, social, family, occupational and relationship Current accommodation, daytime occupation, leisure activities family circumstances Patient's interests, strengths-abilities, likes, dislikes and preferences and how they express these opportunities, impact of disabilities, needs (including mental and physical health) and service and resource gaps Daily/weekly diary
Psychological/ psychiatric factors (*P*) (See also ► Sect. 7.9.3)	Psychiatric disorders: psychoses, bipolar disorders, dementia, depressive disorders, anxiety-related disorders, etc. Psychological symptoms: depression, anxiety, etc. Personality traits Dysregulated arousal and affect
Social/environmental factors (*S*) (See also ► Sect. 7.9.4)	Crowded/noisy/uncomfortable environment Demanding activities, lack of interesting activities, too many changes in the activities, etc. Personalities of other people/staff Change in the environment, activities of daily living at home (e.g. washing and cleaning), activities of daily living outside home (e.g. shopping), relationships, occupation and activities including leisure activities and financial situation and therapeutic interventions Changes required in the level of supervision and support, and major life events including abuse Adequate support for patients and also their caregivers (both family caregivers and paid care staff)

chological and history of use of services and (d) difficulties in developing fulfilling relationships, etc. Therefore, the formulation for the management of PB should be person centred. In this context, it is helpful to have a description of the individual's current and past weekly routine.

7.9.1 Behaviour

Assessment of the actual behaviour should include (a) past history of PB; (b) baseline behaviour prior to the onset of current PB; (c) the onset of the behaviour(s) to describe whether they appeared gradually over time or relatively abruptly perhaps precipitated by an acute event; (d) the frequency, severity and duration of the behaviour(s); (e) nature, content and the context of the behaviour as some behaviours may occur in certain circumstances/settings but not in others; (f) associated behaviours and (g) the impact of the behaviour(s) on the person's life, other's life and the environment. As for the consequences

of PB, the behaviour(s) may lead to (a) reduced quality of life for the individual and her/his caregivers; (b) reduced access to services including education, day service and employment opportunity and may lead to a threatened or actual loss of placement in a residential setting or day placement and (c) reduced social activities including leisure activities, access to friends, etc. In severe cases, the individual may end up being physically restrained, medicated or taken to a hospital or police station. All these scenarios may subsequently have their impact on the individuals, their behaviour and their caregivers. For example, some of these consequences may work as perpetuating factors for the ongoing behaviour(s). Some negative consequences of the behaviour such as physical restraint and use of medication may be seen as inappropriate or excessive and the individuals or their caregivers may perceive them as punishments.

Assessment of risks, including (a) risk to others, (b) risk to the individual, (c) risk to the environment and (d) other risks, forms an important part of the formulation. Clinicians should use the right methods of risk assessment, take note of previous risk assessment and review risks on a regular basis and (e) review the measures taken to reduce risks to assess their effectiveness.

The behaviour must be described in terms that are as follows: (a) clear, specific and unambiguous, (b) measurable (by observation or self-report or observer rating) and (c) capable of future replication.

7.9.2 Medical and Organic Factors

A full review of all bodily systems is essential, as both acute-onset and chronic conditions can present with PB. Assessment of medical and organic factors is an important part of both the differential diagnosis and assessment of contributory factors, and should include history taking, examination and investigations as indicted for:

(a) Chronic physical conditions such as headaches, toothaches, pain in other parts of the body and constipation may manifest as PB. It is important to assess individual's pain threshold as, for example, some individuals with ID, particularly those with autistic features, may have an impaired response to pain (either hypersensitive or hyposensitive) [84]. Gastro-oesophageal reflux disorder is common in people with ID and can present as PB, as can recurrent chest infection, heart/lungs/abdominal/GU/endocrine disease, etc.

(b) Epilepsy and other neurological conditions such as limb spasticity in cerebral palsy [105].

(c) Genetic conditions, such as Lesch-Nyhan syndrome, Fragile X syndrome, Prader-Willi syndrome and Smith Magenis syndrome, are associated with a higher prevalence of PB, particularly SIB [27].

(d) Sensory impairment such as hearing and visual impairment which may lead to PB through frustration but impairment in sensory processing as is known to affect individuals with ASD and ID may also lead to PB, particularly if caregivers are not aware of these difficulties.

(e) Communication problems as in some cases people with ID may communicate their distress through PB. However other forms of communication difficulties such as a discrepancy between expressive and comprehensive speech skills, and impaired social communication skills may also cause confusion and/or frustration leading to PB.

(f) Physical disabilities such as paralysis may cause frustration and lead to PB.

(g) Illicit drug and alcohol-related factors, although this is not known to be a major problem in individuals with ID, could affect a number of individuals with borderline intelligence.

(h) Some prescribed drugs can cause adverse events, which may lead to PB, and there are risks of PBs associated with polypharmacy and with excess anti-cholinergic burden due to polypharmacy, which is common for people with ID.

7.9.3 Psychological/Psychiatric Factors

This should include an assessment of psychiatric disorders as well as neuropsychological and other psychological factors. Psychiatric disorders such as depression, bipolar disorder, schizophrenia and other psychoses and anxiety-related disorders may lead to PB among individuals with ID [21, 66]. However, one has to be mindful about the common problems encountered in making a diagnosis of psychiatric disorders in this population [34]. As ASD and ADHD are common co-morbidities of ID, their influence on the manifestation of PB has to be assessed carefully. Therefore, it is important to investigate for these conditions because the management of PB may vary depending on the presence or absence of any psychiatric disorder. Additionally, clinicians should look for evidence of persistent enduring abnormalities of personality that are contributing to difficulties in interpersonal, occupational and social domains (e.g. paranoid and psychopathic). However, clinicians should be careful in using a diagnosis such as a personality disorder, which may simply be a way of describing PB in people with mild-to-moderate ID, whereas the same behaviour among individuals with severe and profound ID may not attract a diagnosis of a personality disorder.

Clinicians should also look for other contributory psychological and emotional factors such as (a) bereavement; (b) psychological trauma; (c) physical, sexual and emotional abuse; (d) new, ongoing or recurrent stress and life events and (e) relationship difficulty leading to loss of self-esteem and isolation that may perpetuate PB. There are important neuropsychological factors such as (a) impaired intelligence, (b) impaired memory, (c) impaired or abnormal communication skills, (d) impaired executive function, (e) impaired frontal lobe function such as lack of initiative and apathy and (f) lower threshold of stress tolerance, etc., which all should be considered when clinicians are assessing PB to formulate a management plan.

Impairment in executive function may take the form of a lack of planning ability, ability to think in abstract terms, multi-tasking abilities and sense of judgment. A lack or impairment of these functions is likely to cause stress and inability to address problems in day-to-day life and, as a result, may lead to PB. Many people with ID are likely to show impairment in their executive function. This impairment may remain subtle and undetected, and indeed there are practical difficulties in reliably assessing these aspects of cognitive function in most people with ID [122]. However, in the absence of an understanding of this process, the potential for PB may increase.

Some people with ID may appear to their caregivers as unwilling to take part in activities, and more and more demands may be placed on them in order to rectify this behaviour. However, the caregivers may not realise that this behaviour is caused by the impaired function of the pre-frontal cortex and the individual may not, therefore, have any control over their behaviour. This simple understanding on the part of the caregiver may help immensely in the management of such PB.

Individuals with ID may have a lowered tolerance of stress, and this may lead to PB. The lower threshold could be caused by many factors including (a) the cognitive factors that are discussed above, (b) lack of social support, (c) lack of self-esteem, (d) poor self-image, (e) learned maladaptive behaviour from the past, (f) lack of psychological and social reserve including the inability to use advanced psychological defences as opposed to using more primitive psychological defences and (g) an inability to think in abstract terms. All these areas need careful consideration in the assessment.

7.9.4 Social Factors

Assessment of environmental factors including a description of daily activities, educational activities, occupational and leisure activities, relationship and friendship with

others including family members and authority figures and change in the environment are all important factors for consideration in the formulation of management of PB. Such factors may play a major role in predisposing to, precipitating and perpetuating PB. Some individuals may perceive their environment as too demanding. For example, an individual may not want to carry out certain activities that are required of them and are seen as necessary for their personal welfare by the care staff. Similarly, some individuals may find the lack of space around them, other people around them and lots of activities/noise around them overwhelming and stressful. It is important to keep in mind that certain activities that may not seem demanding to a person who does not have ID may be perceived as demanding by some individuals with ID. Similarly, some individuals may perceive their environment as under-stimulating and lacking in appropriate activities. Both under- and over-stimulation in the environment may lead to PB. Structure and routine are also important for some people and lack of these can lead to misunderstandings, anxieties, confusion and so PBs.

Personalities of other people around the individual including those of care staff are likely to affect the individual's behaviour. Similarly, the way others interact with an individual with ID may also impact on her/his behaviour. Lack of respect for the individual with ID may be evident and can lead to PB. The lack of support from others including caregivers and other residents in a house may lead to PB. Similarly, a confrontational attitude on the part of the care staff may perpetuate PB. Although these are important causes for PB and must be considered during the assessment, these issues have to be handled with sensitivity and caution to avoid any conflict with the caregivers or the individual with ID.

Clinicians should also assess whether the individual has experienced any significant life events in the recent or distant past (e.g. loss and change). This should include a history of ongoing or past sexual, emotional and physical abuse. The individual may not feel part of an inclusive service or wider community or may experience stigmatisation and discrimination. He or she may also lack appropriate social exposure and support. All these should be considered carefully. A functional assessment of the behaviour is likely to include all the above elements including an assessment of antecedents and consequences of the behaviour.

An important area to assess is whether the caregivers are adequately supported (including having had adequate training) and whether they are fully involved in the formulation and provision of care. The need for support for stress and fatigue among caregivers should be assessed carefully. The absence of these supports may help perpetuate PB by creating a vicious cycle.

Other important areas to assess are the organisational settings, systems and processes in place to support the individual with ID and her/his caregivers, and the need to address the possible absence of adequate support.

Another useful schema (*H.E.L.P*) for assessment of PB in people with ID has been proposed by Bradley and colleagues (see ◘ Fig. 7.3) [16].

7.10 Assessment Methods/Tools

There is a large and growing number of behaviour rating instruments and it exceeds the scope of a book chapter like this to provide a comprehensive presentation and critical appraisal of all the available instruments. We have presented here some of the more prominent instruments (see ◘ Table 7.5). We have not presented scales that assess only psychiatric disorders or stereotyped behaviour. Further information on assessment scales is available from other sources including the UK NICE Guideline [123] and the European Guideline [124, 34].

7.11 Functional Assessment

The hallmark of behavioural interventions is to develop well-founded hypotheses about the contingencies of reinforcement of a given target behaviour in order to guide the development of rational customised behavioural

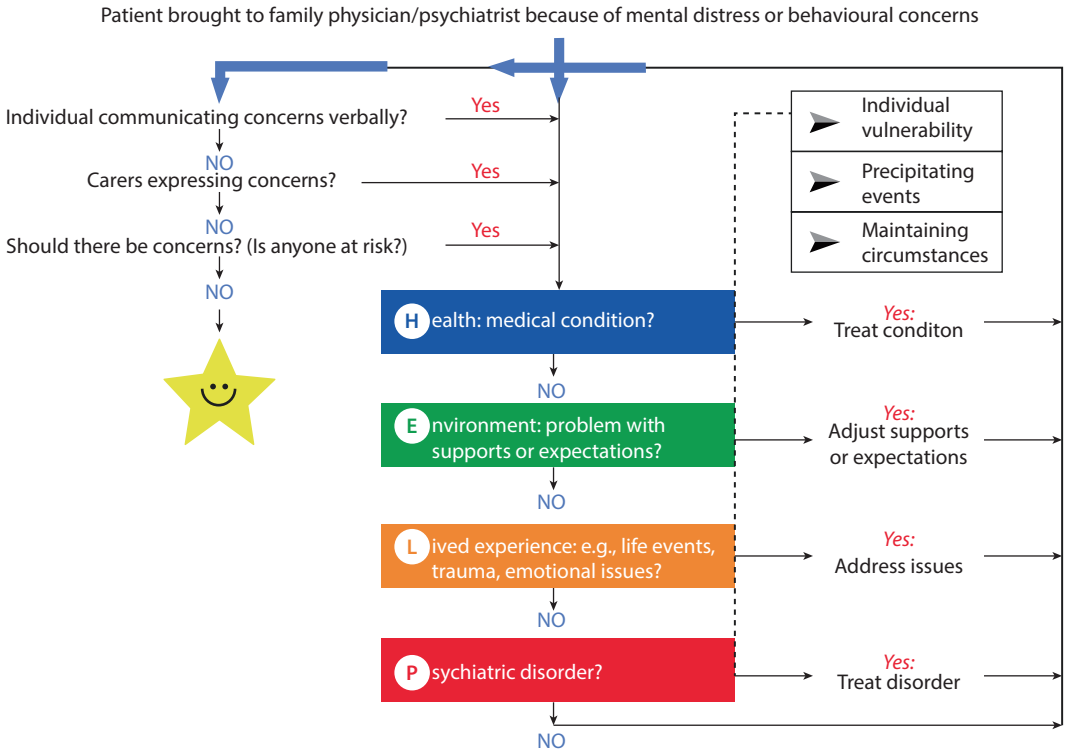

Fig. 7.3 H.E.L.P. diagram

interventions. Functional assessment can include interviews, naturalistic contingency observations and rating scales. In Table 7.4, we present some commonly used functional assessment rating scales. Various methods for functional and contextual assessments have been proposed with associated benefits and drawbacks [125]. Experimental manipulation of variables allows for the inference of causality; however, this method is time consuming, requires a high level of expertise and artificially producing AB using potentially aversive stimuli is unethical [125, 126]. Direct observation is also potentially an intensive resource, especially if behaviours are not exhibited frequently [125, 126]. In response, the use of interviews and checklists with informants are the most preferred and widely used methods for functional assessment [127] to provide a simple, quick, accessible assessment and requiring less expertise on the part of the clinician [125, 126].

7.12 Conclusion

PB, in general, and AB, in particular, is a complex phenomenon that involves a multitude of factors and is the outcome of an interaction between the person and her/his environment. It is difficult to define and classify PB using any standard psychiatric classification system such as the DSM-5 (disruptive, impulse control and conduct disorder) [21]. One reason is that by definition these disorders assume that the person is behaving with intent, which is difficult to determine in most cases of PB in ID. The other difficulty is to determine the criterion that these behaviours are inappropriate for their age as in ID the chronological age does not correspond to the developmental age. Also, a diagnosis of the intermittent explosive disorder cannot be made before 6 years of age or equivalent developmental level according to DSM-5 criteria, which makes this diagnosis impossible for individuals with severe and pro-

Table 7.5 Some rating scales used for the assessment of problem behaviour in people with intellectual disability

Instrument title Author(s) and year of publication	Description and target population	Items and subscales
Combined scales for assessment of behavioural and psychiatric problems		
Developmental Behaviour Checklist (DBC) (Einfeld and Tonge, 1994) [133]	Parent or teacher rated; children and adolescents Standardised and normed assessment of emotional and behavioural problems	96 items (parent version), 93 items (teacher version); 5 subscales: disruptive/antisocial, self-absorbed, communication disturbance, anxiety and social relating
Adaptive Behavior Scale (ABS) and ABS Residential and Community: 2nd Edition (ABS-RC:2) (Nihira et al., 1975) [134]	Informant rated; ABS-RC:2: all ages Measures personal independence and social skills divided into two parts: first measures individual responsibility and functioning or social adaptation; second part measures social maladaptation and can be used to measure problem behaviour Three types of score are available: domain, factor and comparison Adequate psychometric properties, however, has low inter-rater reliability for part 2	356 items, divided into two parts Second part has seven subscales: social behaviour, conformity, trustworthiness, stereotyped and hyperactive behaviour, self-abusive behaviour and social engagement
Diagnostic Assessment of the Severely Handicapped-II (DASH-II) (Matson, 1995) [135]	Informant rated; all ages; severe and profound ID only Developed to reflect DSM categories that are most commonly diagnosed in people with ID Six behavioural factors: emotional lability, aggressive behaviour/conduct disorder, language disorder/verbal aggressive behaviour, social withdrawal/stereotyped behaviour, eating disorder and sleep disorder Measures severity, frequency and duration of behaviours Good psychometric properties have been reported	84 items; Eight subscales: impulse control, organic syndromes, anxiety, mood/depression, mania, pervasive developmental disorder/autism, schizophrenia and stereotypies/tics
Matson Evaluation of Social Skills for Individuals with Severe Retardation (MESSIER) (Matson et al., 1998) [136]	Informant rated; all ages; severe and profound ID only Assesses social strengths and weaknesses including providing a profile of social skills both positive and negative and maladaptive behaviours. Good reliability data have been published by the authors of the instrument	85 items Six subscales: positive verbal, positive non-verbal, general positive, negative verbal, negative non-verbal and general negative

Table 7.5 (continued)

Instrument title Author(s) and year of publication	Description and target population	Items and subscales
Nisonger Child Behavior Rating Form (NCBRF) (Aman et al., 1996) [137]	Parent or teacher rated; children Social competence and problem behaviour subsections Adequate psychometric properties are reported The NCBRF has age- and gender-based norms on a restricted sample of children and adolescents	66 items in the problem behaviours subsection
Reiss Screen for Maladaptive Behavior (RSMB) (Reiss, 1997) [138]	Informant rated, all ages Psychopathology screening instrument (not a diagnostic tool) Clinically and factorially derived subscales Factor structure and good concurrent validity replicated	36 items; Eight subscales: aggressive behaviour, autism, psychosis, paranoia, depression (behavioural signs), depression (physical signs), dependent personality disorder and avoidant personality disorder
Scales of Independent Behavior-Revised (SIB-R) (Bruininks et al., 1996) [139]	Informant rated, all ages with developmental disabilities and other populations with needs for special assistance Designed to measure adaptive and maladaptive behaviour and to determine the type and amount of special assistance required Normed scores are available	Four clusters of independent behaviour and three clusters of problem behaviour: internalised, externalised and social
Vineland Adaptive Behavior Scales, second Edition (VABS) (Sparrow et al., 2005) [140]	Self-, parent- and teacher rated, all ages Measures adaptive and maladaptive behaviours among people with ID, ASD, ADHD, post-traumatic brain injury, hearing impairment and dementia One of the most widely used standardised assessment instruments. Survey and expanded interview version available	49-item optional maladaptive behaviour section: 10 internalising behaviours 10 externalising behaviours 15 'other' behaviour problems 14 'critical' items

(continued)

Table 7.5 (continued)

Instrument title Author(s) and year of publication	Description and target population	Items and subscales
Specific behaviour problem scales		
Aberrant Behavior Checklist (ABC) (Aman et al., 1995) [69], later *ABC-Community (ABC-C)* and *Aberrant Behavior Checklist, 2nd Edition (ABC-2)* (Aman and Singh, 2017) [141]	Informant rated; all ages Assessment of a broad range of problem behaviour Scale empirically developed and subscales determined by factor analysis. ABC-C developed to assess behaviour among community-residing individuals with ID Favourable psychometric properties; has been widely used and evaluated	58 items Five subscales: irritability, social withdrawal, stereotypic behaviour, hyperactivity/noncompliance and inappropriate speech Subscale scores should only be used; total score is not a valid measure [141]
Adult Scale of Hostility and Aggression: Reactivel Proactive (A-SHARP) (Matlock and Aman, 2014) [142] also Children's-SHARP (C-SHARP) (Farmer and Aman, 2009) [143]	Informant rated, adult and children's version Empirically developed measure of severity and/or frequency of interpersonal aggression among people with aggressive behaviour and ID Captures reactive and proactive aggression Factors derived from factor analysis	52 items; Five factors: verbal aggression, physical aggression, hostile affect, covert aggression and bullying
Behavior Problems Inventory (BPI-01) (Rojahn et al., 2001) [144] and the short version (*BPI-S*) (Rojahn et al., 2012) [145]	Informant rated, all ages Measures maladaptive behaviour in terms of frequency and seriousness Regularly used for clinical and research purposes Empirically derived factor structure Fair-to-excellent psychometric properties have been reported by the authors of the scale	BPI-01: 52 items; and BPI-S: 30 items. Three subtypes of behaviour: SIB, stereotyped behaviour and aggressive/destructive behaviour
Challenging Behavior Interview (CBI) (Oliver et al., 2003) [146]	Informant rated, all ages Assesses the severity of challenging behaviour Adequate psychometric properties reported by the authors of the scale	Five behaviours first rated for presence: Self-injury, physical aggression, verbal aggression, disruption of the environment and inappropriate vocalisation 14 questions to establish the severity of each behaviour rated as present
Checklist of Challenging Behavior (CCB) (Harris et al., 1994) [147]	Informant rated, all ages Developed to determine prevalence of challenging behaviour among people with ID Little validation data are available	Two parts: 14 items relating to aggression/physical contact with others or self 18 items relating to other problem behaviours including stereotypical behaviour and property destruction

Table 7.5 (continued)

Instrument title Author(s) and year of publication	Description and target population	Items and subscales
Modified-Overt Aggression Scale (MOAS) (Ratey and Gutheil, 1991) [148]	Informant rated, all ages Provides scores for the past week. Was not developed for use with people with ID but has been successfully used with people with ID and has established reliability [149]	16 items; Four categories of behaviour: verbal aggression, physical aggression against objects, physical aggression against the self and physical aggression against others
Repetitive Behavior Scale-Revised (RBS-R) (Bodfish et al., 2000) [150]	Informant rated, all ages Assesses the severity of abnormal and repetitive behaviours Empirically derived scale with good reported psychometric properties	43 items; Six subscales: stereotyped behaviour, self-injurious behaviour, compulsive behaviour, ritualistic behaviour, sameness behaviour and restricted behaviour
Problem Behaviour Checklist (PBCL) (Tyrer et al., 2016) [151]	Informant rated, all ages Based on factor analysis of the original 37-item scale; has good inter-rater reliability and convergence with MAS (Tyrer et al. [152])	28 items; Seven subscales: personal and property violence, self-harm, sexually inappropriate behaviour, contrary behaviour, demanding and difficult behaviour and wandering
Functional assessment		
Functional Assessment for Multiple Causality (FACT) (Matson et al., 2003) [153]	Informant rated, developed using a sample of adults Developed to assess problem behaviour that may be maintained by multiple functions. Uses forced choice questions Empirically derived with factors derived from factor analysis Limited data on psychometric properties; initial evaluation indicates good validity and reliability [153]	35 items; Five subscales: tangible (target behaviour has a history of producing tangible items), physical (behaviour is exhibited during physical discomfort), attention (behaviour attracts attention from others), escape (behaviour is used to escape disagreeable situations) and non-social
Functional Analysis Screening Tool (FAST) (Iwata and DeLeon, 1995) [154]	Informant rated, all ages Should be used as part of a comprehensive functional assessment Moderate psychometric properties reported	16 items; Four subscales: social/attention/preferred items, social escape, automatic sensory stimulation and automatic pain attenuation
Motivation Assessment Scales (MAS) (Durand and Crimmins, 1988) [155], (Spreat and Connelly, 1986) [156]	Informant rated, all ages Clinically derived subscales Relatively weak psychometric properties reported	16 items; Four subscales: sensory, escape, attention and tangible

(continued)

Table 7.5 (continued)

Instrument title Author(s) and year of publication	Description and target population	Items and subscales
Questions About Behavior Function (QABF) (Singh et al., 2009) [157]	Informant rated, all ages Clinically derived subscales, acceptable good psychometric properties have been reported	25 items; Five subscales: attention, escape, non-social, physical and tangible These subscales are cross-referenced against nine behaviours: physical aggression, verbal aggression, self-injury, tantrums, disruptive behaviours, stereotypes, inappropriate verbal behaviour and non-compliance
Contextual Assessment Inventory for Problem Behaviour (CAIPB) (Carr et al., 2008) [125], (McAtee et al., 2004) [126]	Informant rated; developed using a sample of adults For the assessment of contextual factors. Has adequate convergent and predictive validity and test-retest reliability but questionable inter-rater reliability	93 items; Four categories, each with two or three subcategories: social/cultural (including negative interactions and disappointments), tasks/activity (including factors related to tasks and daily routines), physical environment (including uncomfortable environment and changes in the environment) and biological (including medication and illness)
Setting Event Checklist (SEC) (Gardner et al., 1986) [158]	Informant rated; all ages Identifies motivational conditions or setting events. Limited psychometric evaluation	Over 78 setting events; Organised into nine categories: physical setting, time of day, day of week, weather conditions, activities, presence of particular clients, presence of particular staff, social context and personal context
Setting Event Inventory (SEI) (Tustin et al., 1997) [159]	Informant rated; all ages Identifies setting events for problem behaviour The authors report good inter-rater reliability and internal consistency	155 items; 19 scales, 6 of which measure preceding behaviours of individuals and 13 relate to the environment (including carer attention, instruction and touch; criticism or correcting; peer agitation or encroachment; possessions; task difficulty and organised activity; noise; change and disappointment)

found ID and also for most with less severe ID. Another major problem in defining PB is that almost inevitably PB is reported by someone else rather than the person exhibiting the behaviour. Therefore, the same behaviour which appears problematic to some may not appear so to others. Having said that, there are some severe behaviours, for example, SIB related to Lesch-Nyhan syndrome will almost universally appear as a PB. As a result, a dimensional approach to define psychopathology associated with PB in people with ID is the best approach in dealing with this behaviour instead of trying to categorise them according to any standard psychiatric classification system.

The management of PB will require a thorough assessment of causes and effects of the individual's behaviour. Therefore, we recommend that a formulation based on the bio-psycho-social causes and the effects of PB must be established first before initiating any

intervention. In assessing PB, the types of PB should be described along with severity ratings (mild, moderate and severe) and a frequency rating (low, medium and high). As the definition of severity and frequency would vary depending on many factors such as the type of behaviour and perception of the person reporting the behaviour, etc., neither 'severity' nor 'frequency' is defined here and is left with the clinicians to define under each specific circumstance.

AB remains a major management challenge and causes immense distress to people showing the behaviour and others around them. AB may lead to unnecessary hospitalisation and use of medication, exclusion from many social opportunities and is a source of stress for family and friends. Both neurobiological and environmental factors play a large role in predisposing to, precipitating and maintaining AB, and must be taken into account during the assessment (*B.M.P.P.S.*) (see Table 7.3). In addition, co-morbidities of physical and psychiatric disorders can contribute. A suggested schema (*H.E.L.P.*) (see Fig. 7.3) can be used to do this assessment, including a bank of specific tests of cognitive and executive function, medical investigations to investigate organic causes and consideration of psychosocial and environmental factors. This will ensure a holistic and optimum approach to successful management of aggression. In essence, as aggression is multi-factorial, its management has to be multi-faceted with multi-professional input in an integrated fashion with the ultimate goal to improve the people with ID and their carers' quality of life.

For pharmacological and nonpharmacological management of PB and intervention refer to ▶ Chaps. 11 and 12, respectively.

Key Points

- Problem behaviour is more prevalent in people with intellectual disabilities than those who do not have intellectual disabilities.
- Problem behaviour is multi-factorial in aetiology including internal factors like genetic disorders and external factors such as the environment and other psychosocial issues.
- Therefore, a multi-professional, person-centred, bio-psycho-social approach is needed for a thorough assessment of problem behaviour.
- Both pharmacological and non-pharmacological interventions are implemented for its treatment but their role remains limited, making this a major management problem.

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Instrumental Assessment

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Contents

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Learning Objectives

The issues discussed in this chapter do no focus on a specific area of mental health, but they are pivotal to the whole field of mental health assessment, certainly in people with ID and LF-ASD, but probably also across the whole population. The basic questions we are asking is 'how should we be doing assessment, and who should be collecting the information?'. In many service contexts, it is assumed that psychiatrists and psychologists will work mainly on the basis of their clinical knowledge and expertise, while other staff are the ones likely to use structured instruments. For patients with ID and LF-ASD, the problems of language often lead to a great emphasis on the completion of structured assessments by informants. Very often, a patient will receive multiple assessments by a variety of health professionals, but then someone has the task of synthesizing this into a meaningful formulation. Many questions arise by this scenario. For example, should all the clinicians use structured methods? What type of assessments should be used? How should we bring all this information together? Should we actually be structuring the whole assessment process, including the final formulation? This chapter's main aim is to offer some practical and theoretical guidance on the incorporation of structured assessments into clinical services working with people who have ID and/or LF-ASD.

8.1 Introduction

The broad aims of this chapter are to offer some guidance on the use of structured assessment methods with people who have intellectual disability (ID) and/or low-functioning autism spectrum disorder (LF-ASD), and to give a review of the main tools available as well as some examples of their use.

In order to consider the potential contribution of using structured methods, it is necessary to look closely at the fundamental issues relating to the whole process of mental health assessment. There are many reasons why someone might wish to employ a structured method, and there are likely to be differing viewpoints of the potential users of such assessments. An informed decision about the use of, and choice of, structured assessment inevitably needs a close look at some difficult questions; the answers to which may not always be clear cut. If structured assessments are to be used in a clinical service, this may be to measure changes in response to treatment, or as part of the diagnostic process. In either case, it is desirable to examine the quality of information they generate, how this information will be used, whether it aids communication between different professionals and how much confidence clinicians have in the information. Potentially, the adoption of structured methods can significantly improve the overall quality of assessment and monitoring, but in order to do this there needs to be agreement among the clinical team about what methods are to be used and when to use them.

8.2 Why Use a Structured Assessment?

These are some of the numerous reasons why one may wish to use a structured assessment:

- To improve the quality of one's own clinical assessment
- To draw on the knowledge of other staff who may not have a formal training in psychiatry psychology, but who knows the patient better than yourself
- To free up expert clinical time by getting lesser-paid people to collect information
- To get the patient's own perspective by asking them to complete their own answers
- To enable a common assessment framework across professionals within the service
- To enable the use of a potentially 'free' resource, that is, the person themselves and their family

Many clinical services employ questionnaires and checklists to collect part of their information. However, the choice and selection of assessments are not always done with a coher-

ent assessment policy in mind. Broadly speaking, psychiatrists and psychologists are much less likely to use structured methods than other staff, as they rely too much and too often on clinical impression. Whether it is desirable for the senior clinicians also to include structure in their assessments is something we will also consider.

8.3 Structured Assessments and Developmental Level

The fact that many people with ID have very poor language, or no language at all, sometimes leads to the suggestion that structured assessments are more important with people who have ID than with the general population. Certainly, if the person has very poor language, we become highly reliant on third-party reports, and these reports are often aided by checklists and questionnaires. If the person is of a developmental level sufficient to have good language, and their disorder does not lead to a lack of insight, then assessment relies largely on what the patient says. This does, however, lead to a fundamental question: why do we often use structured methods to collect information from third parties, but rarely structure the clinical interview itself? Interviewing is a highly volatile process, open to influence by numerous factors that will be discussed shortly.

8.4 Fundamental Dimensions of Structured Assessments

There are a great many published assessments, making it sometimes difficult to decide which ones to employ. The following notes hopefully give some guidance on the main differences between them.

8.4.1 Different Models of Mental Disorder

This is probably the most fundamental dimension that determines how we assess, and what structured tools we might employ in the process. Within this dimension, one of the biggest problems that faces any designer of assessments is how to reconcile psychiatric and behavioural approaches.

8.4.1.1 The Psychiatric Approach

Over the hundred or so years that psychiatry has existed as a discipline, it has been observed that symptoms cluster together. For example, a person who is depressed is also highly likely to have lost interest in things, feel low in energy and perhaps feel hopeless about the future. Through these observations, the classic diagnostic constellations or syndromes have gradually been identified and clarified (see ► Chap. 5). One of the main focuses of psychiatric assessment is, thus, to collect the information necessary to decide on which of these clusters the person's symptoms belong.

8.4.1.2 The Behavioural Approach

A behavioural analysis does not seek to identify pathologies, but rather looks at the relationships between antecedent conditions and behavioural consequences. This is often done with the aim of changing antecedent conditions to reduce undesirable behaviours. Behavioural assessments may, therefore, simply collect information about various behaviours, and may additionally provide some form of scoring which is often based on factor analysis. Behavioural analysis and interventions are widely used when working with people who have ID.

8.4.1.3 Reconciling These Two Approaches

What is the difference between a 'symptom' and a 'behaviour'? In a sense, all symptoms manifest of course in observed behaviours. However, the behaviours identified in behavioural analysis are usually ones that are considered of major concern in their own right; for example, aggression or self-injury. In comparison, symptoms like depressed mood or anxiety are probably better described as *indicators* of a general condition that may be manifested in many other ways beyond the actual listed symptoms in ICD-10 or DSM-5.

A real problem of reconciling behavioural and psychiatric approaches arises with symptoms/behaviours that can be viewed in radically different ways, for example:

- Worry about being abandoned
- Chaotic relationships
- Impulsiveness
- Self-harm
- Splitting
- Anger

From a behavioural/ecological perspective, such symptoms may be regarded primarily as a response to long-term history, including early childhood development, bonding and attachment, and ecological factors. These are, however, also symptoms for *borderline personality disorder*. These two different views require two different assessment approaches. If one is seeking primarily to determine whether the person meets the criteria for this disorder, then the chosen assessment tool would need to focus closely on severity and duration of the symptoms. If one is wishing to conduct a behavioural analysis, then the relationship between the behaviours and the person's wider ecology would need to be assessed. Each of these approaches would require a different assessment tool.

For people with ID/LF-ASD, a particular issue arises in relation to *challenging behaviour*. Challenging behaviours are highly prevalent in people with ID (see ▶ Chaps. 5 and 7), and it is notable that the term 'challenging' has been introduced in preference to the term 'problem behaviours'. What is the difference between a 'challenging behaviour' and a 'problem behaviour'? Many people with ID, particularly those with most severe cognitive impairment, can exhibit aggressive or destructive behaviours, but probably do not fully understand their impact or social significance. The term 'challenging' implies that the person with ID is not really in control of these behaviours, and that we, as service providers, have a responsibility to help the person. Diagnoses such as conduct disorder and personality disorder, on the other hand, imply that the person *does* understand the consequences of his/her action but nevertheless does it (or even enjoys doing it in the knowledge that it will cause upset to others).

The more severe the level of ID, the more difficult it becomes to judge whether the person understands the consequences of the action. While these judgements are very difficult, structured approaches to assessment can be helpful if they present the assessor with a formal framework that includes points to consider and appropriate choices to make. For instance, the Child and Adolescent Psychiatric Assessment Schedule (ChA-PAS) [1] section on Conduct Disorder offers the following four rating categories: *Staying out against parental/carer wishes*

1. Not present/not rated
2. [P] Has frequently stayed out at night against parental/carer wishes, but at least one of the following applies:
   (i) Does not fully understand the effect it has on others
   (ii) Behaviour appears to be driven by frustration, or an emotional reaction to life circumstances
   (iii) Gets genuinely upset by the resulting distress to carers
3. [P] Has frequently stayed out, and is more or less indifferent to the distress it causes to carers, despite being aware of the effect it is having
4. [P] Stays out repeatedly against carers' wishes, fully understands the effect it has, but responds in at least one of the following ways:
   (i) Takes pleasure in the upset it causes
   (ii) Makes up glib excuses that are clearly lies
   (iii) Expresses glib feelings of guilt and remorse that are clearly not genuine

Only levels 2 and 3 contribute to a diagnosis of conduct disorder. Level 1 scores as zero because of the extenuating circumstances that indicate it is not a symptom of this disorder.

8.4.1.4 The Statistical Approach

Some assessments do not start with the ICD-10 [2] or DSM-5 [3] rules, but instead take a purely statistical approach to the identification

problems. For instance, the Developmental Behaviour Checklist [4] was developed by starting with a very large number of behaviours, which were then subsequently rendered down into a number of factors using factor analysis. The advantage of such an approach is that it achieves a very wide coverage of all the behaviours of concern. The shortcoming is that the resulting factors may not be easy to interpret in terms of clinical practice or interventions.

8.5 The Coverage of the Assessment

This to some extent relates to the discussion of models of mental illness in the section above. However, it is important to distinguish broad-spectrum assessments from those that are for a specific area, such as autism, depression, and ADHD. When choosing a specific area assessment, it is important to consider whether the use of this tool might bias the final outcome in favour of the area covered. For instance, consider a clinic that specializes in ADHD, and routinely uses an ADHD-specific tool to collect the data. Unless adequate safeguards are in place, there is a risk that other potential explanations for person's problems, for example, bipolar disorder, will receive less consideration than they merit.

> Potentially, the adoption of structured assessment methods can significantly improve the overall quality of clinical assessment and monitoring, especially with reference to patients with lower levels of communication or cognitive ability. The selection of structured assessments, both informant based and self-rated, needs careful matching to the characteristics of the patient. If the assessment has a specific focus, for example, ADHD, it is important to consider whether that choice might bias the outcome in favour of the area covered.

8.5.1 The Level of Structure, Detail, Precision, Guidance and Analysis

The relationships between these five characteristics mentioned in the title are probably the most central aspects that distinguish between the various assessments that are available. To think about the interaction between these aspects, it is important to start with the desired outcome of using structured tools. There are actually many ways in which their use can be of benefit [5], but here we will consider four of the main reasons:

1. To screen for potential cases who will subsequently receive an in-depth assessment
2. To predict what an expert clinician would probably conclude if a full assessment was undertaken
3. To collect information that will form part of a subsequent diagnosis or formulation
4. To make actual diagnoses or formulations

Reasons 1 and 2 above have in common that no expert clinician is involved in the process: they both imply that the person competing it may not have a background in psychopathology. Screening tools are often highly structured, with little precision needed in categorizing the answers, and little guidance given beyond the wordings of the items themselves. Analysis takes the form of a built-in scoring system that renders an answer to the question, 'should this person receive a more in-depth assessment?'. Screening tools do, to some extent, predict what an expert would conclude, but research studies wishing to make more accurate predictions will often use a more complex assessment, probably requiring training. In either case, the psychometric properties of the assessment are very important. In the case of screening, the most important psychometric aspect is probably that no genuine cases are missed. To make more accurate predictions of expert opinion, the assessment must be reliable and valid. This is discussed in more detail shortly.

A very different form of structure is present in ICD-10 [2] and DSM-5 [3]. They are themselves structured assessments, in that they provide rules to determine what symptoms are associated with the various diagnoses, along with information relating to time course, etc. They have a high degree of general clinical guidance and distil the knowledge of a great many experts about, for example, the relationships between physical and mental disorders. They are, however, *low* in precision because they give relatively little guidance on how to identify different manifestations of the same symptom. This is particularly important in relation to people with ID because they very often show symptoms in a different way, particularly if their level of ID is severe. ICD-10 [2] and DSM-5 [3] also have low precision in terms of guidance on symptom severity. How severe should a symptom be before one includes it as present; how would a mild example of the symptom and a severe example differ?

The clinical interview is often one of the principal avenue by which these diagnostic decisions are made. Clinical interviewing is frequently conducted without a formal structure. In this next section, we will consider whether structuring such interviews could be beneficial.

Screening for cases to undergo in-depth assessment, predicting clinician conclusion, collecting information of clinical utility and making actual diagnoses or formulations represent the four main ways in which the use of assessment tools can be of benefit. It is important to know from the beginning the desired result of using structured instruments during the assessment, considering that the clinical interview is often the principal avenue by which the diagnostic decisions are made.

8.6 Clinical Interviewing

Interviewing patients and informants is one of the primary methods for collecting information on mental status. Generally speaking, nothing can replace the validity of the patient's own report; for which reason, clinical interviewing is often used to make pivotal decisions. Surprisingly, training in the process of interviewing is often neglected, and the use of structured formats is rarely employed. To illustrate the advantages of a structured approach, consider one of the core symptoms of depression, low mood itself. If a patient is showing evidence of this symptom, how should we decide if the symptom is strong enough to say it is present?

The Present State Examination, the Schedules for Clinical Assessment in Neuropsychiatry (SCAN) [6] and the Psychiatric Assessment Schedule for Adults with Developmental Disability (PAS-ADD) [7] assessments all take the view that it should be possible to rate a symptom as present, but that it is *below the threshold for clinical significance*. This is a very important point. Many of us experience symptoms of low mood or anxiety from time to time, but mostly these would not be considered pathological. The provision of a subclinical category also flags up the presence of a symptom that might subsequently become more significant in the future.

In order for the assessment of symptom severity to be workable and reliable, some rules need to be applied to each symptom. The Present State Examination relied mainly on use of the terms: *not present, mild, moderate* and *severe* as the rating categories. The limitation of this is that the terms mean different things to different people. The PAS-ADD [7] system has developed and evolved over about 25 years at the time this was written. Much of that evolution was in terms of the refinement of severity definitions. The current definitions for low mood (in the Mini PAS-ADD [8] Version 3) are as follows:

Item 7: Depressed mood

1. Not present or not rated.
2. Episodes of depressed mood occur most days, but [P] is sometimes more cheerful or can very often be cheered up by the intervention of others.
3. Depressed mood is a significant problem on most days, but attempts to cheer [P] will sometimes be successful.
4. The depressed mood is persistent and unresponsive to attempts to cheer [P] up.

When scoring, rating level 1 actually produces a score of 0. In other words, depressed mood is there, but it is considered to be sub-clinical.

The other dimension of structured interviewing is the wording of questions. It has been observed by one of us (SM) that it is easy to get involved in a lengthy discussion of a symptom with the patient or informant, yet still not have the information to tie down the symptom severity. The evolution of the PAS-ADD system has also resulted in question forms that are designed to determine symptom presence and severity as efficiently as possible. The question for low mood in the Mini PAS-ADD Version 3 (interview for informants) is as follows:

- **Item 7**
  - How is [P]'s mood?
  - Is [P] happy?
  - Does [P] feel depressed/low?
  - How does s/he show it? What does s/he do?
- **If Evidence of Depression:**
  - Is there anything you can do to cheer him/ her up when s/he is like this?
  - During {rating period}, has [P] been depressed all the time?
- **If not all the time:**
  - Are there some days when s/he is happier? Or just periods during the day?

Note how the wordings enable each of the severity categories to be evaluated.

8.7 Interviewing People with ID

A sizeable proportion of people with ID *can* respond meaningfully to a clinical interview, but the questions need to be carefully formulated and sensitively asked. Using the PAS-ADD 10, Patel, Goldberg and Moss [9] showed the following ratings of interview adequacy by the interviewer (who was an experienced psychiatrist).

Rating of subject's account of symptoms	Adequacy (%)
Subject responds adequately	36
Account somewhat inadequate but interview can proceed	13
Account seriously inadequate but interview proceeds in an attempt to rate some subjective responses	12
Impossible to continue with interview	38

Thus, nearly half the adults who were interviewed were able to give a useful contribution to a clinical interview. Given the unique validity of the patient's own report, it is highly desirable that any way of improving the quality of patient interviewing in people with ID is worth the effort. Good structured assessments, designed specifically for people with ID, can help in this respect, partly because they can provide ways of asking questions that have been tried and tested over a period of time. Also, the structure provides a way of helping the interviewer probe other possible problem areas that may not be clearly visible at the beginning, and hence, may have been omitted.

Training in the process of interviewing is often inappropriately neglected, and the use of structured formats is rarely employed.

Specifically designed, structured assessment can improve the quality of patient interviewing in people with ID/LF-ASD for many reasons, including the provision of questions that have been tried and tested over time and the possibility to probe problem areas that could be omitted.

8.8 Psychometric Properties

It is not proposed to have a lengthy discussion of psychometric issues here; they are well described elsewhere [10]. However, it might be helpful to offer some thoughts regarding the interpretation of psychometric data.

8.8.1 Reliability

First of all, nobody doubts the desirability of making assessments reliably. Any assessment, structured or otherwise, that produces different results from the same case presentation is of questionable value. The problem is that reliability in structured assessments is a result of various factors:

- The number of items
- The number of rating categories
- The tightness with which the ratings are defined
- The amount of training the users receive
- Whether the user retain their training or drift away from it

In choosing a structured assessment on grounds of reliability, an appropriate balance must, therefore, be struck. Does it cover the information you need, at the level of detail you require? If training is required, are you able to ensure the users get adequately trained? Using an assessment designed for trained raters which is being used by *non-trained* raters runs the risk of producing results that look highly informative but may simply be wrong.

8.8.2 Validity

The issue of validity is far more complex than that of reliability, and there is one main reason why this is the case. Consider the process of diagnosis in physical medicine. Over hundreds of years, the relationships between symptoms and their causes have become more and more precisely clarified. The validity of a physical diagnosis can be measured by the extent to which it correctly identifies the pathology. Nowadays, thousands of different procedures are used to identify pathologies, and the diagnosis is made in the light of that evidence. In comparison, we are making diagnoses of mental health problems, usually *in the absence* of clear evidence of physical causes. This lack of relationship between cause and effect makes the process of diagnosis fundamentally different. Diagnoses of mental health are, generally speaking, clinical judgements. This does not mean they have no utility, but it is vital to recognize this fact when considering issues of validity.

How, then, do we estimate the validity of a mental health assessment? Bearing in mind that ICD-10 and DSM-5 are themselves structured assessments, how could we estimate their validity? One answer would be that they are of course valid because they represent the distilled knowledge of clinical experts, collected over a very long period of time. As such, they can be considered a *gold standard*. However, the fact that they are not precise means that the validity of an assessment as measured by its conformity to ICD-10 or DSM-5 is not a very useful measure. A more common gold standard is that of clinical expertise. How far does the assessment predict what a clinical expert would say about the case? This approach has merit, but the limitation is that people completing the assessments are often in a very different situation from the clinical experts whose opinion is sought. The clinical expert may indeed have greater expertise to sift and judge clinical material, but may not be in such a good position to actually know the client.

Linking the knowledge and expertise of the various stakeholders in the case, from families through to all the health and social service professionals who may be involved, is something that structured assessments can potentially enhance.

8.8.3 Factor Structure

From a psychometric perspective, structured assessments are often considered to be better if their underlying factor structure shows a meaningful relationship to the concepts being

measured. For instance, items purporting to assess obsessive compulsive disorder should be closely correlated together, and less correlated with items in another section, such as depression. To some extent this is undoubtedly true. The person who is low in mood is often someone who has lost energy, feels hopeless for the future, etc. However, there is another consideration to be borne in mind. Both DSM-5 and ICD-10 take a *polydiagnostic* approach to diagnosis, that is, two different people can have widely differing symptoms yet still receive the same diagnosis. Also, it is well recognized that some symptoms overlap different diagnostic constellations, for instance, there are clear overlaps between ADHD and bipolar disorder, and between obsessive compulsive disorder and autism.

These considerations mean that an assessment that conforms very closely to the rules of ICD-10 and DSM-5, and is, hence, clinically very useful, may actually perform more poorly in terms of factor structure than an assessment that was designed primarily using a statistical approach.

> In choosing a structured assessment, an appropriate balance must be struck between psychometric properties (reliability, validity and factor structure) and contextual clinical utility. When necessary, users have to be adequately trained.

8.9 Some Examples of Structured Assessments

This chapter does not attempt a comprehensive review of structured assessments. Rather, we offer a sample of these, showing how they differ in terms of the various fundamental dimensions:

- Area covered
- Sophistication of rating and questioning
- The method of collecting information
- The primary aim of the assessment
- Who are the likely users

8.9.1 Fully Diagnostic Interview

8.9.1.1 The PAS-ADD Clinical Interview

The PAS-ADD Clinical Interview [7] is the most complex of the PAS-ADD assessments. It generates ICD-10 and DSM-5 diagnoses on a criterion-by-criterion basis, allowing the user to see clearly the extent to which individual criteria have been fulfilled. It covers all the principal axis I psychiatric disorders in detail, and additionally has an informant interview for ADHD and a screen for autism. There is a strong emphasis on final interpretation and diagnosis by the users themselves. There are no restrictions placed on who may use the interview, although it is clear that a background in psychiatry psychology is highly desirable in terms of interpreting results.

The PAS-ADD Clinical Interview was derived originally from the Present State Examination [11]. The original version, the PAS-ADD 10, was subsequently revised more than 20 years later in the light of information derived from training users over that period. During that time, many changes in question wording and definitions of rating severity were made.

The PAS-ADD clinical interview can be used with anyone who has sufficient language and cognitive development to be able to participate in a clinical interview. This includes children as well as adults, both with and without intellectual disability. The interview can also be used with an informant only.

Information is gathered through patient and/or informant interviewing, for whom there are separate sets of questions. Symptoms are rated on a 4-point scale of severity, the use of which enables a lot of precision to be built into the fulfilment of the diagnostic criteria. As with all the PAS-ADD assessments, the PAS-ADD clinical interview is equally valid for the general population, in which case questioning of the patient can use the informant questions. The PAS-ADD assessments are now also available in digital versions,

available via a website which can also be used off-line for conducting interviews where no Internet is available.

The refinement of the scoring system is illustrated by its approach to psychosis. Psychotic symptoms are notoriously difficult to identify reliably. In the PAS-ADD clinical interview, scores can only get the top severity rating if they are based on the patient's own account of symptoms rather than on an informant report. As such, diagnoses based on informant interviewing alone usually result in 'query' diagnoses for psychosis. The relationship between mood and psychotic symptoms undergoes a complex analysis to determine the final diagnosis.

Field trials investigated the validity of PAS-ADD in relation to the clinical opinion of referring psychiatrists [12, 13]. Inter-rater reliability of the PAS-ADD 10 [2] gave a mean Kappa of 0.65 for individual item codes of, and Kappa 0.7 for agreement on index of definition (clinical significance of the symptoms) [14]. The relationships between respondent (patient) and informant reports of symptoms, and the implications of deriving diagnoses solely from informant interviews, are discussed in Moss and colleagues [15]. The issues of using care staff as informants are discussed in Moss and Patel [16].

8.9.2 Other Structured Interviews

There are various structured assessments using an interview format that produce what would be better referred to as diagnostic 'indications'. These are not screening tools because they potentially provide enough information to actually make a diagnosis. They do not, however, provide a detailed criterion-by-criterion analysis that directly maps onto ICD-10 or DSM-5 criteria.

8.9.2.1 The PIMRA

The Psychopathology Instrument for Mentally Retarded Adults (PIMRA) [17–19] was designed for the use with adults who have mild-to-moderate levels of intellectual disability, and was originally derived from DSM III [20]. It is available in an informant interview version, and also for self-report (meaning self-completion by the informant). Questions have to be answered by yes or no. The PIMRA is designed to assess psychiatric disorder and psychopathology. It has eight sub-scales, scores from which measure seven diagnostic categories: schizophrenia, depression, psychosexual disorders, adaptation disorders, anxiety, somatoform disorders and personality disorders.

Many research studies have been conducted on the PIMRA, producing mixed conclusions about its psychometric properties. Thus, while the authors showed good internal consistency and acceptable test-retest reliability [18], others have shown lower reliability [21–25]. The construct validity was examined for the schizophrenia sub-scale first by Linaker and Helle [26] and then by Swiezy and collaborators [27], for the psychosexual sub-scales by Matson and Russell [28]. The construct validity of the remaining five sub-scales has not yet been rigorously evaluated. The factorial analysis of PIMRA was carried out first by Balboni and collaborators [29] and then by Sturmey and Ley [21]. The investigations did not produce clear and consistent results: only four main factors were identified and not all the eight sub-scales of the instrument were referred to independent constructs. The results of other researches indicate that the PIMRA is useful above all for research purposes, and therapeutic planning and outcome evaluation [27].

8.9.2.2 The DASH

The Diagnostic Assessment for the Severely Handicapped (DASH) [30, 31] assesses the presence of psychiatric disorders in people with severe and very severe ID. It consists of 84 items and organized in the following sub-scales based on the diagnostic criteria of DSM-III-R [32]: control of impulses, organic disorders, anxiety disorders, mood disorders, mania, pervasive developmental disorders, autism, schizophrenia, stereotyped behaviour, self-injurious behaviour, elimination disorders, nutrition disorders, sleep disorders and sexual disorders. The assessment is an informant interview, where each item is scored on a 3-point Likert scale for frequency, duration

and severity. The psychometric characteristics of the instrument have been well established, especially for the sub-scales of depression, mania and autism [33–37]. Overall, these confirm the reliability of the instrument, although further research is needed to prove the construct validity of many sub-scales.

The two-factor analysis conducted across time [31, 38] identified six and five factors, respectively, of which only the following three were in common: emotional lability, language disorders and sleep disorders.

DASH-II has been used in numerous clinical and epidemiological studies, including those referred to adolescence or early adulthood [39] and senescence [40].

8.9.2.3 Mini PAS-ADD and ChA-PAS

The Mini PAS-ADD [8] and the Child and Adolescent Psychiatric Assessment Schedule (ChA-PAS) are for completion by informant interview, and are widely used in Europe and Australia. They adopt the same four-point rating structure as in the PAS-ADD Clinical Interview, and similarly cover the principal axis I psychiatric disorders. Both include the autism screen, and the ChA-PAS [1] additionally covers ADHD and conduct disorder. In most cases, the scoring conforms exactly to the rating criteria laid down in ICD-10 [2] or DSM-5 [3]. However, there are some minor departures to enable a single score to be generated for each of the diagnostic constellations. For instance, the requirement for specific core symptoms of depression (low mood, loss of interest and loss of energy) is not specifically required.

The Mini PAS-ADD was conceived as an assessment that could provide an in-depth investigation of symptoms, but could be completed by people who did not necessarily have a background in psychology or psychiatry. The ChA-PAS follows this format, and was produced in response to requests for an assessment whose symptom definitions and questions were more appropriately directed towards younger people. These two assessments include a lot of guidance on how to recognize the various symptoms, and provide a semi-structured interview structure to help guide the way in which questions are formulated.

Psychometric properties of the Mini PAS-ADD can be found in Prosser et al. [41]. Results of a major study on the Dutch version of the Mini PAS-ADD have also been reported [42].

8.9.2.4 The CIS

The Clinical Interview Schedule (CIS) – mental handicap [43] – was created originally for the general population and adapted subsequently for the use of persons with ID. The version for the general population [44] was designed to be used by experienced psychiatrists, following a specific training, as a support in the formulation of ICD diagnosis. It is divided into four parts: one with the pathological anamnesis, both remote and near; one with a structured interview including ten groups of symptoms; one with additional information on the family and personal history and the last with a detail of anomalies communicated during the interview. This last part is the one that has undergone multiple changes in the adaptation to use with the ID, with seven additional items. It is also the one that, in the adapted version, presents the lowest inter-rater reliability. In general, the few data available on the psychometric characteristics of this scale are not encouraging and the terminology maintained in the various items seems to be less usable by most people with ID [45].

8.9.3 Full-Spectrum Questionnaires

These are wide-spectrum assessment, but are primarily checklists rather than interviews.

8.9.3.1 The P-AID

The Psychopathology Checklists for Adults with Intellectual Disability (P-AID) [46] is a battery of checklists to be used with informants, and is able to identify 10 different psychiatric disorders and 8 types of problem behaviours according to the Diagnostic Criteria for psychiatric disorders for use with adults with Learning Disabilities/mental retardation (DC-LD) [47]. It shows internal consistency, reliability and acceptable inter-rater, 8 units orthogonal to factor analysis,

while sensitivity and specificity are still to be explored [46].

8.9.3.2 The DBC

The Developmental Behaviour Checklist (DBC) [4] is an excellent example of an assessment whose development started from a primarily statistical standpoint. A large number of different kinds of behaviour in children were investigated in a large population of children with emotional or behavioural difficulties. From the scores, factor analysis derived a number of factors, and subsequently a smaller, refined set of items. The parent/carer version has 96 items in five different sub-scales. The items are scored on a 4-point Likert scale. The six factors identified by factor analysis are as follows: disruptive behaviour, self-absorbed behaviour, communication disturbance, anxiety, autistic-relating behaviour and antisocial behaviour.

The version for adults (DBC-A) [48, 49] includes 107 items, which are completed by family members, paid carers or someone else who know the person with ID well, reporting problems over a 6-month period. Each descriptive item of behavioural and emotional disturbance is scored on a 3-point Likert scale, from 'not true' to 'very true'. The DBC-A can be scored at three levels: (1) the overall measure, total behaviour problem score or alternatively the mean item score (MIS), the proportion of items checked (PIC) and the intensity index (II); (2) sub-scale scores measure disturbance in six dimensions, which may also be scored as total scores or as MIS with PIC and II and (3) scores on individual items.

Because it was derived from a statistical viewpoint, its psychometric properties are very good [50–54]. As such, it is very useful for epidemiological studies, but the factor scores may relate less to routine psychiatric practice.

8.9.3.3 The BSI

The Brief Symptom Inventory (BSI) [55] is an instrument of self-evaluation of psychological distress, developed for the general adult population, which essentially represents the brief form of the Symptom Checklist-90-R (SCL-90-R) [56]. It evaluates a wide range of symptoms through 53 items organized in the following nine sub-scales: somatization, obsession compulsion, interpersonal sensitivity, depression, anxiety, hostility, phobic anxiety, paranoid ideation and psychoticism. It also allows to obtain three global indices of psychopathology: general gravity, symptom distress and general symptomatological positivity. The BSI has been judged to be a valid tool for the self-assessment of psychological symptoms in people with mild ID or borderline intellectual functioning, although internal consistency and reliability were found to be rather low for all sub-scales [57–59]. The validity of the construct was verified by comparing sub-scales' average scores of three different groups of persons: persons living in the community attending a specialized clinical unit for assessment of ID severity (community group), persons under evaluation for co-occurrent psychiatric disorders (clinical group) and persons convicted of a crime (forensic group). The three groups presented significant differences in eight of the nine sub-scales and in two of the three global indices. The community group showed the lowest number of symptoms, followed by the forensic group and the clinician [58].

8.9.3.4 The PPS-LD

The Present Psychiatric State – Learning Disabilities (PPS-LD) [60] is an adaptation of the Present State Examination [61] and supports the psychiatric diagnosis according to the Diagnostic Criteria for Research-10 [62]. It contains 116 items and complements the information gathered by the evaluator and his closest assistants.

8.9.4 Screening Checklists

Screening assessments do not claim to provide comprehensive assessment on which a formulation or diagnosis can be made. Rather, they aim to make a statistical prediction of what an in-depth assessment would conclude, were to be undertaken, in the same way that a cancer screening gives an estimate of the probably presence of the disease. Very often, this simply takes the form of a yes/no conclusion, that is, 'yes', the person probably has a problem that

needs further assessment, or 'no', they probably do not have a mental health problem. Of course, the scores generated by screening assessments can also be valuable in their own right, giving an indication of the kinds of areas where problems might exist.

Two of the most important statistical characteristics of screening assessments are *sensitivity* and *specificity*. An assessment that is sensitive will be good at identifying all potential cases. An assessment that is specific will be good at distinguishing between non-cases and cases. Of course, one cannot get something for nothing. Checklists, when used by non-trained users, are very unreliable, and hence, not very specific. In order to increase sensitivity, it is inevitable that one has to lower the threshold score for identifying 'caseness', and this in turn increases the number of false-positive results (i.e. people who appear to have a problem when they actually do not).

In choosing a screening assessment, these points need to be considered carefully. If one wishes to definitely identify all possible cases, and then follow them up, a very sensitive assessment would be the choice. The disadvantage is that there will be many false positives, that is, people who triggered the screen but do not actually have a disorder. If resources for full assessment are limited, it may be appropriate to choose an assessment with higher specificity, on the assumption that all the severe cases would be identified. One would have to accept that some cases would be missed, but they would hopefully be the more mild ones.

8.9.4.1 The PAS-ADD Checklist

The PAS-ADD Checklist is a 25-item questionnaire, written in everyday language, designed for use primarily by care staff and families, the people who have the most immediate perception of changes in the behaviour of the people for whom they care. The Checklist aims to help staff and carers decide whether further assessment of an individual's mental health may be helpful. It can be used to screen whole groups of individuals, or as part of a regular monitoring of people who are considered to be at risk of mental illness. It is designed to record the presence of a range of problems, all of which may be part of a psychiatric condition. The scoring system includes threshold scores which, if exceeded, indicate the presence of a potential psychiatric problem, which may then be more fully assessed.

The PAS-ADD Checklist produces three scores, relating to:

1. Affective or neurotic disorder
2. Possible organic condition (including dementia)
3. Psychotic disorder

It also has a checklist for life events.

Factor analysis of the checklist completed on a community sample of 201 individuals yielded eight factors, of which seven were readily interpretable in diagnostic terms. Internal consistency of the scales was generally acceptable. Inter-rater reliability in terms of case identification, the main purpose of the checklist was quite good, 83% of the decisions being in agreement with expert clinical opinion. Validity in relation to clinical opinion was also satisfactory; case detection rising appropriately with the clinically judged severity of disorder [63]. Subsequent independent studies have further investigated the Checklist's psychometric properties [64], and established norms for an adult sample [65].

8.9.4.2 The ADD

The Assessment of Dual Diagnosis (ADD) [66] was developed to evaluate the full range of psychiatric disorders in adults with mild or moderate ID. To overcome any verbal capacity limits of the proband, the instrument can be administered to someone who knows the person to be evaluated well, such as a caregiver or family members. The scale consists of 79 items and 13 sub-scales organized according to the DSM-IV diagnoses: mania, depression, anxiety, post-traumatic stress disorder, substance abuse, somatoform disorders, dementia, conduct disorder, developmental pervasive disorder, schizophrenia, personality disorders, eating disorders and sexual disorders. Authors found good psychometric characteristics [66]. One study examined the correlations between the ADD sub-scales and a short version of the MMPI-168 (168-item Minnesota Multiphasic Personality

Inventory), adapted for use with people with ID [67]. The sub-scales of the two instruments, which measured similar psychological constructs, resulted not to correlate with each other, suggesting that ADD may have problems of construct and convergent validity.

8.9.4.3 The RSMB

The Reiss Screen for Maladaptive Behavior (RSMB) [68, 69] is one of the older and well-established scales evaluating psychopathology in individuals with ID through an interview to their caregiver or family member. It includes 38 items accompanied by a definition and some examples. The tool provides scores for the following eight dimensions: aggressive behaviour, autism, psychosis, paranoia, behavioural signs of depression, physical signs of depression, dependent personality disorder and avoidant disorder. The score assigned to each item is chosen on a 3-point scale based on symptom severity, that is, the impact of symptoms on the person's functioning during the last 2 months. A high score at one or more of the tools dimensions indicates a need for referral for more detailed evaluation.

The RSMB showed good psychometric properties across the range of ID, although some uncertainties on its factorial structure were pointed out as well as a higher sensitivity to depression than to other areas of psychopathology [68, 70–75].

8.9.4.4 The ABCL

The Adult Behavior Checklist (ABCL) [76] was not specifically designed for ID or LF-ASD. In fact it is an adaptation of the Young Adult Behavior checklist (YABCL) [77], created to support the clinician in the assessment of psychopathological symptoms in young adults of the general population. The ABCL includes 118 items to be self-rated by a proxy of the person with ID through a 3-point Likert scale ('not true', 'a little bit true' or 'sometimes true' and 'quite true') and in reference to the last 3 months. When used with persons with ID, the ABCL confirmed the good psychometric characteristics showed with use with the general population, including high concordance with clinical diagnoses based on DSM-IV criteria [78, 79]. The factor analysis identified eight syndromic subgroups: anxiety/depression, social withdrawal, somatic complaints, altered thinking, problems of attention, aggressive behaviour, transgressive behaviour and intrusiveness. Furthermore, two broad groups were also identified and defined as 'internalizing' and 'externalizing' disorders. The former showed the strongest correlations with the sub-scales of social withdrawal, somatic complaints and anxiety/depression, the latter with the sub-scales of transgressive and aggressive behaviour. In addition, the ABCL showed good predictive abilities of individual functioning, as measured by the Global Assessment of Functioning (GAF) [79], the Social Functioning Scale for the Mentally Retarded (SRZ-P) [80] and the Best Status Index (Best) [78, 81]. An equivalent of the ABCL for the developmental age, the Child Behavior Checklist (CBCL) [82], has also been repeatedly studied in children and adolescents with mild ID, with as many results of validity [83–85].

8.9.4.5 The SPAIDD-G

The Systematic Psychopathological Assessment for persons with Intellectual and Developmental Disabilities – General screening (SPAIDD-G) [86, 87] is part of a wide tools system to support professionals working with persons with ID and LF-ASD in the identification of psychopathological symptoms and syndromes. It includes 56 items, which represent descriptions of the most frequent observable and behavioural aspects of all the symptoms that appear in the various DSM-5 diagnostic categories. These items were developed to be rated by a mental health professional through the information gathered by interviewing a family member of the person with ID/LF-ASD or any other proxy who have a good perception of changes in the behaviour of the people for whom they care.

Raters do not have to attribute or rate the severity of any score, they only have to indicate the presence or absence of an item by ticking the appropriate box.

The SPAIDD-G evaluates the following syndromic groupings, consistent with those included in the DSM-5: nutrition/feeding disorders, psychotic disorders, mood disorder –

depression, mood disorder – mania, anxiety disorders, side effects of drugs, delirium, dementia, substance-related disorders, odd personality disorder, dramatic personality disorder, anxiety personality disorder, impulse control disorder, autism spectrum disorder, dissociative identity disorder, somatic symptom disorder, sexuality disorder and obsessive compulsive disorder.

The SPAIDD-G showed very good psychometric characteristics. To date it is available only in Italian, although validations of the English, German and French translations are already underway.

8.9.5 Assessments Focusing on a Specific Area

Most of the assessment tools for specific psychopathological areas are adaptations of tools originally created for the general population. Their validity for use with persons with ID/LF-ASD is uncertain, especially with reference to those with greater difficulties of communication and psychological insight.

The vast majority of tools for specific psychopathology concern mood and anxiety disorders. In recent years, there has been an increasing focus of research on both autism spectrum disorder (in persons with ID) and dementia. This interest is likely to result in the development of further assessments in the future.

8.9.5.1 Mood Disorders

In the area of mood disorders, the literature includes significant reports for all the following tools: Affective Rating Scale [88], Hamilton Depression Scale – Mental Handicap Version [89], Beck Depression Inventory (BDI) [90], Mental Retardation Depression Scale (MRDS) [91, 92], Self-Report Depression Questionnaire (SRDQ) [93], Zung Self-Rating Depression Inventory: Mental Handicap Version [94–96], Intellectual Disability Mood Scale [97], the Anxiety, Depression and Mood Scale (ADAMS) [98], Mood, Interest and Pleasure Questionnaire (MIPQ) [99], Glasgow Depression Scale for people with a Learning Disability (GDS-LD) [100], Mood and Anxiety Semi-Structured Interview (MASS) [101] and interRAI Intellectual Disability Assessment System (InterRAI ID) [102].

8.9.5.2 The SRDQ

The Self-Report Depression Questionnaire (SRDQ) [93] is not, as the name suggests, for completion by the patients themselves. Rather, it is a series of questions posed *to* the individual themselves. There is a pre-test to determine whether the person is able to respond meaningfully to the questions. The items are rated on a 3-point scale.

The SRDQ has good psychometric properties [103, 104] and is probably helpful in clinical work around depression in this population [105–107].

8.9.5.3 The IDMS

The Intellectual Disability Mood Scale (IDMS) also showed good psychometric characteristics [97]. However, it has been studied almost exclusively with persons with mild ID. It is articulated in six dimensions: anger, confusion, depression, fatigue, tension and vigour. Given the possible difficulties of employing self-report measures with people who have intellectual disabilities, numerous extra procedures were used, including pictures of buckets filled to various levels and marked to represent the five points of the response scales.

8.9.5.4 The ADAMS

The Anxiety, Depression and Mood Scale (ADAMS) [98] is an informant interview and can be used for all levels of ID severity. It is composed of 28 items organized into five sub-scales: manic/hyperactive behaviour, depressed mood, social avoidance, generalized anxiety and compulsive behaviour.

8.9.5.5 The MIPQ

The Mood, Interest and Pleasure Questionnaire (MIPQ) [99] includes 25 items grouped into two sub-scales (mood and interest/pleasure). It is an informant interview with Likert scale scores and for use with reference to every person with ID or LF-ASD, regardless of communication and insight impairments. It showed good psychometric properties, including high concurrent validity with the Aberrant Behavior Checklist (ABC) [108].

8.9.5.6 The GDS-LD

The Glasgow Depression Scale for people with a Learning Disability (GDS-LD) [100] integrates the information collected by interviewing the person with ID (mild-to-moderate degree) with that provided by their usual carer. For this purpose the instrument is divided into two parts containing 20 and 16 items.

8.9.5.7 The MASS

The Mood and Anxiety Semi-Structured Interview (MASS) [101] is a standardized diagnostic interview specific for anxiety and mood disorders, as indicated by its name. It has been designed to be used with the usual caregivers of the person with ID and is composed of 35 items corresponding to as many symptoms of DSM, whose presence or absence has to be indicated by the rater with respect to the month before the compilation. MASS showed good sensitivity and specificity, with high concordance rates both with clinical diagnoses and with the scores of the Hamilton Depression Rating Scale (HDRS) [109]. As mentioned above (see ▶ Sect. 6.1), about 10 years ago a diagnostic interview was also published for mood and anxiety disorders, called Mood and Anxiety Semi-Structured Interview (MASS) [101].

8.9.5.8 The InterRAI ID

The interRAI Intellectual Disability Assessment System (InterRAI ID) is a comprehensive, standardized instrument for evaluating the needs, strengths and preferences of adults with intellectual or developmental disabilities. It includes two sub-scales to assess depression and aggression. The InterRAI ID is in turn part of a wide tool battery developed in 2007 to promote social and community engagement by identifying persons who may benefit from additional supports or services in specific areas [102].

The concurrent validity of the InterRAI ID sub-scales for depression and aggression was evaluated through comparison with the RSMB and showed statistical significance [102].

Anxiety Disorders

In the area of anxiety disorders and obsessive compulsive disorder, the following tools have all been valued in the scientific literature across time: Zung Anxiety Rating Scale: Adults Mental Handicap Version [110, 111], Glasgow Anxiety Scale for people with an Intellectual Disability (GAS-ID) [112], Fear Survey for Adults with Mental Retardation (FSAMR) [113] and Yale-Brown Obsessive Compulsive Scale (Y-BOCS) [114, 115].

The GAS-ID consists of 27 items referring to physical symptoms and single phobic aspects, while the FSAMR is much more complex, including 85 items. Both instruments are designed to be used with people with mild-to-moderate ID or borderline intellectual functioning, but the GAS-ID shows the advantage of requiring a lower attention span.

Autism Spectrum Disorders

Tools to screen for ASD in adults with ID have to be chosen on the basis of their suitability with different levels of conceptual and communication impairments. The 10-item Autism Spectrum Quotient (AQ-10) [116] is one of the most common for use with borderline intellectual functioning and mild ID, while the Diagnostic Behavioral Assessment for Autism Spectrum Disorder – Revised (DiBAS-R) [117] and the Pervasive Developmental Disorder in Mental Retardation Scale (PDD-MRS) [118] seem to be more appropriate for moderate ID. In most severe cases, the Systematic Psychopathological Assessment for persons with Intellectual and Developmental Disabilities – Autism Spectrum Disorder (SPAIDD-ASD) [119] or the Autism Spectrum Disorder – Diagnosis Scale for Intellectually Disabled Adults (ASD-DA) [120] should be preferred.

To support clinicians in the refinement of the diagnosis, the Autism Diagnostic Observation Schedule (ADOS; ADOS-II) [121, 122] and the Autism Diagnostic Interview – Revised (ADI-R) [123] are widely employed, although they have shown to present some problems of sensitivity and validity

when used with adults with ID, especially in highest degrees of severity and in the presence of other psychiatric disorders [124]. The Gilliam Autism Rating Scale – Second Edition (GARS-2) [125] seems to show greater discriminative capacity, even if research evidence is limited.

The Diagnostic Interview for Social and Communication Disorders (DISCO) [126] also offers a comprehensive evaluation of the ASD across the range of ID, allowing to subtype on the basis of overall functioning [127] and supporting the differential diagnosis between ASD, ID and schizophrenia spectrum disorders [128].

Recently, the Social Communication Questionnaire (SCQ) [129] has been adapted for use with adults with ID of various severity (SCQ-AID) [130] and can be used as a screening tool. However, it still has major specificity problems: the high scores often do not derive from the presence of many DSA nuclear symptoms but from that of CP, co-occurring psychiatric disorders or undesirable effects of psychoactive drugs.

More information on diagnostic instrumental assessment of ASD in persons with ID is provided in ► Chap. 16.

Dementia

In the area of dementia and other major neurocognitive disorders, three well-known and widely studied tools are the Dementia Scale for Down Syndrome (DSDS) [131], the Dementia Questionnaire for Mentally Retarded persons (DMR) [132] and the Dementia Screening Questionnaire for Individuals with Intellectual Disabilities (DSQIID) [133]. For persons with mild ID and good introspective and communicative skills, some adaptations of instruments widely used in the general population are also available, such as the Shultz Mini-Mental State Exam [134] and the Cambridge Examination for Mental Disorders of Older People with Down Syndrome and Others with Intellectual Disabilities (CAMDEX-DS) [135].

The DSDS includes 60 items organized into three categories, one for each of the stages in which the progression of the disorder is normally divided. The DMR instead has two categories, in which it orders 50 items and 8 sub-scales: the first category concerns cognitive functions, such as memory and orientation, while the second refers to social-emotional skills, such as mood, interests, speech, practical skills and problem behaviour. Although DMR has been designed to be used with persons with ID of any degree of severity, its sensitivity has been shown to decrease in those with highest cognitive impairment [132].

Another instrument of interest is the Multidimensional Observation Scale for Elderly Subjects (MOSES) [136], which is also centred on the detection of significant changes in behaviour through the reports of reliable informants. Its structure is based on three behavioural factors: adaptive, maladaptive externalizing and maladaptive internalizing [137].

The Dementia Screening Questionnaire for Individuals with Intellectual Disabilities (DSQIID) [133] was designed with the aim of overcoming the main limits of previous instruments. It focuses only on recent behavioural changes and shows very good psychometric characteristics. For these reasons it was included, with some adaptations, in the evaluation package of the National Task Group on Intellectual Disabilities and Dementia Practices (NTG), called NTG-Early Detection Screen for Dementia (NTG-EDSD) [138], and recently translated and validated into various languages.

A comprehensive review of measures used in the screening, assessment and diagnosis of dementia in persons with ID has been recently published by McKenzie and collaborators [139].

Personality Disorders

In the area of personality disorders, two relevant tools are the Standardized Assessment of Personality (SAP) [140, 141] and the Minnesota Multiphasic Personality Inventory 168(L) [MMPI-168(L)] [142].

Although initial studies using the SAP were limited to people with mild and moderate ID [141, 143], subsequently it has been used across the whole range of severity, showing good psychometric properties [144–147]. The SAP is a semi-structured diagnostic

interview which relies on having an informant who knows the patient well from at least 5 years.

The MMPI-168(L) [142] represents a modified version of the MMPI-168 [148], which is in turn a reduced version of the well-known Minnesota Multiphasic Personality Inventory. The MMPI-168(L) [142] was produced for use with persons with mild ID and includes simplified questions that can be answered yes or no, rather than true or false. A group of L (Lie) items was also added to increase validity. It provides scores for three validity scales and ten personality scales. The MMPI-168(L) has been shown in empirical studies to possess good psychometric characteristics, particularly towards odd cluster personality disorders [67, 149–151].

8.9.6 Structuring the Whole Case Formulation

Little attention has been given to the idea of actually providing structure to the way the whole case is formulated, but such an approach deserves consideration. It is very clear that most people's mental health problems are a complex mixture of fundamental factors: biological, developmental, psychodynamic and ecological. As such, it is rare that a single person provides all the information necessary to understand fully why the person is experiencing the problems that they have. Multidisciplinary assessment is normally used, but with it comes the problem that different professionals have different ways of thinking, and are often in very differing relationships with the patient. Family members know the person best, but rarely have professional knowledge of psychopathology. Senior clinicians have the best ability to synthesize clinical information, but are often not in the position to collect it themselves. In a large organization, different professional groups sometimes work so independently that it is difficult to bring together all the relevant information pertaining to the specific case.

It is often true in case formulation that a given set of observations and symptoms can be accounted for by a number of different hypotheses. Given all the information, how do we decide which hypothesis is true?

8.9.6.1 The PAS-ADD Clinical Interview: Formulation Section

The score form for the PAS-ADD clinical interview provides an opportunity to bring together all these disparate pieces of information in one place; essentially, to make a dossier of the information necessary to make diagnostic decisions. The last part of the score form provides a way of identifying the key elements of the case in relation to various fundamental dimensions: psychiatric assessment, birth history and developmental factors, problem behaviours, physical health status, intellectual adaptive and language functioning, current ecology and 'contingencies'. This last item refers to the pivotal evidence that can arise when something suddenly changes in the person's behaviour that relates to something that has happened in their life. Such contingencies can often give unique insight into what is actually happening.

The key to completing this section of the score form is that the various items are rated according to their judged relevance to *understanding the case*, rather than to their severity. For instance, someone may have a severe self-injurious behaviour, but it always happens in the same way and at the same level. This, in itself, does not tell us much about what is going on. In comparison, someone's self-injurious behaviour that fluctuates markedly in response to what is happening around them, or started after a major life event, can tell us a lot more.

This part of the form has not been tested in any study; indeed, it would be very difficult to mount such a study. However, clinicians have reported that the completion of this section by a small group of professionals over a short period of time can often highlight hypotheses for why the person is suffering the problems, and can also identify further key pieces of information that would be desirable to collect. This is of course the same process that goes on in many case discussions. As with

other structured assessments, however, the provision of a structure can sometimes help the team consider all the pieces of evidence in a more impartial way.

8.9.6.2 The SPAIDD: An Integrated System of Tools

As mentioned above, the Systematic Psychopathological Assessment for persons with Intellectual and Developmental Disabilities (SPAIDD) [87] is an example of a comprehensive set of tools for all the different practical needs related to psychopathological assessment and monitoring, to be used by mental health professional and the whole multidisciplinary team working with people with ID and LF-ASD. In fact, it includes tools for every phase of the clinical intervention, such as general psychopathological screening, diagnostic categorical specification, dimensional diagnosis and symptoms monitoring.

The items of the SPAIDD system have been kept unchanged across all the battery tools since they represent observable and behavioural aspects of cross-categorical symptoms.

In designing and implementing this system, authors tried to overcome also the other main limits of previous tools, such as the impossibility of being used for all the degrees of intellectual and communication impairment, the misalignment with the DSM or the ICD, the lack of some main symptoms or syndromes, the lack of chronological criteria, the long times of administration and the scarce interdisciplinary usability [152].

To date, the SPAIDD system includes tools for general screening, follow-up, autism spectrum disorder, mood disorders, anxiety disorders and psychotic disorders, which are available only in Italian, although validations of the English, German and French translations are underway.

8.9.7 Other Structured Assessments

The above sections covered only a small proportion of the numerous structured assessments that are available for use with people who have ID/LF-ASD. Hopefully, the ones we have highlighted give some insight into the dimensions that need to be considered when selecting appropriate ones. Useful sources of further information are represented by the reviews by Flynn and collaborators [153], Hermans [154] and Tyrer and collaborators [155].

Assessment tools can be distinguished on the basis of their fundamental dimensions, we can count: (a) fully diagnostic interviews such as PAS-ADD; (b) other structured diagnostic interviews like PIMRA, DASH, mini PAS-ADD, ChA-PAS and CIS; (c) checklists such as P-AID, DBC, BSI and PPS-LD; (d) screening checklists like PAS-ADD Checklist, ADD, RSMB, ABCL and SPAIDD-G; assessment focusing on a specific area, especially mood and anxiety disorder and dementia. New comprehensive sets of tools for all the different practical needs related to psychopathological assessment and monitoring are being developed for interdisciplinary use by mental health professional and the whole multidisciplinary team.

8.10 Choosing a Structured Assessment

The appropriate choice of assessment(s) for a research project may be very different from those needed for a clinical service. In either case, however, the choice should be guided by a consideration of the desired outcome of the assessment.

8.10.1 Measuring Change

One of the most straightforward uses of structured assessments is probably for the measurement of change, either in response to treatment or over a period of time. For this purpose, issues of validity become less significant. *Reliability* is the central requirement. One needs to have faith in the ability of the assessment to reproduce the same results in

the same set of circumstances. A change in these results, therefore, indicates a change in the person's mental status. Although most structured assessments give estimates of reliability, it is important to bear in mind that this often depends on the training of users. In a research study, it is relatively easy to obtain good reliability through rigorous training of the users. Some structured assessments suggest, or require, some form of training. If the particular assessment is to be used, therefore, it must be accepted that resources are allocated for that training. Even after training, however, it is possible that users will drift away from the correct procedure. Indeed, Havercamp and Reiss [24] indicated that problems of reproducibility of PIMRA results by different evaluators seemed to arise because professionals in different areas recorded differently, or had not received specific training in the use of the tool. In a real-life clinical setting, it would certainly be desirable to monitor and update the training of users from time to time.

8.10.2 Diagnosis and Formulation

Clinicians such as psychiatrists and psychologists often do not use structured methods. One possible reason is that they feel it would be more time consuming. Another is that they may feel their clinical training is sufficient that a structured approach is not necessary. The numerous studies of the reliability of clinical interviewing indicate that the latter reason is questionable. Diagnostic reliability tends to be low [156–158], but this is certainly not due to a lack of clinical skills. Rather, it is an indication of the multiple sources of information that tend to influence outcome, for example, reason for referral, or bias in the people who know the individual.

It would not be sensible to suggest that everybody always should use a formal structured assessment in every case. Rather, structured tools should be like any other tools, that is, to be used where appropriate. More broadly, it is very useful to learn how to use some of these methods so that even routine clinical interviewing can be conducted with a more rigorous structure in mind. This can help overcome sources of influence and bias and can also give the opportunity for other possible diagnoses and formulations to be given consideration.

Ultimately, the various assessments are probably best judged by their *usefulness*; do they help to achieve the desired outcome? In clinical work, structured assessments can contribute to the quality of the patient's clinical information, and any issues of reliability and validity can be handled at the level of case discussion. For instance, a structured interview may statistically be reliable and valid, but the clinical team knows that the informants are not giving a true picture, either because they have their own agenda or they are unable to be objective. In an extreme example, someone with psychopathy may *deliberately* mislead the assessors. Such things can only be judged in the context of the case, and lie outside the confines of the assessment's statistical properties.

8.11 Structured Assessments as Part of a Protocol

Finally, it is important to mention that the use of structured assessments in clinical work is much more beneficial if it is part of an overall policy or protocol on assessment in the service. Unfortunately, this often does not happen. It is sometimes perceived, for instance, that one professional group should receive training in a particular assessment, but the rest of the health professionals are unaware of this. As a result, the information from these assessments may be considered by other clinicians to be of questionable validity.

It is only by getting health professionals together to learn about and discuss the assessments that they can become part of the flow of information, leading hopefully to better case formulations in the future.

One of the most straightforward uses of structured assessment is probably the measurement of change, either in response to treatment or over a period of time. To this purpose, reliability is a central

requirement and consequently specific and updated training of users. More broadly, it is very useful to learn how to use some assessment tools so that even routine clinical interviewing can be conducted with a more rigorous structure in mind. This can help overcome sources of influence and bias and give the opportunity for other possible diagnoses and formulations to be given consideration.

Tip

The use of structured assessments in clinical work is much more beneficial if it is part of an overall policy or protocol on interdisciplinary assessment in the service.

The instrumental assessment of psychiatric disorders in persons with lower communication and cognitive skills needs substantial improvement and it is probable that this will be brought in the next years. Thus, constant update is suggested.

Key Points

- Interviewing is a highly volatile process, open to influence by numerous factors that can be very difficult for people with ID associated with a very poor language, or even no language at all. This sometimes leads to the suggestion that structured assessments can be more important with people who have ID than with the general population.
- When choosing to use a structured approach, however, it is important to distinguish broad-spectrum assessments from those involving a specific area with the aim of not influencing the final diagnostic outcome and determining the presence and severity of symptoms as efficiently as possible.
- An assessment tool should have good psychometric properties, in particular, reliability and validity associated with a factorial structure in significant relationship with the measured concepts. Reliability seems to be the central requirement so that a change in the results indicates a change in the person's mental state and is important to bear in mind that this often depends on the training of users.
- The ideal is a tool that allows both to synthesize all the information obtained from the patient himself, from third parties close to the patient or from other colleagues who have previously examined him or, at the same time, in the case of a multidisciplinary work group, and to evaluate the various symptoms significant for diagnosis. This is even more true in the case of mental health professionals who work with people with ID and LF-ASD.

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Epidemiology of Psychiatric Disorders in Persons with Intellectual Disabilities

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Contents

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Learning Objectives

This chapter is devoted to the epidemiology of psychiatric disorders in people with intellectual disabilities (ID). It starts by considering the types of psychiatric disorders that people with ID experience, within the context of diagnostic overshadowing and the multi-morbidity that typically occurs in people with ID, and the implications when conducting assessments. It then considers the reported prevalence of psychiatric disorders across the life-course and compares this with the prevalence in the general population, and also for people with co-occurring ID and autism spectrum disorder. Relevant aspects of aetiology are then considered, followed by information on the course of psychiatric disorders in this population.

1. To understand how commonly psychiatric disorders are experienced by children, young people, and adults with ID
2. To understand variation in the prevalence of psychiatric disorders in people with ID compared with the general population
3. To understand the influence of co-occurring autism spectrum disorder with ID on the prevalence of psychiatric disorders
4. To understand the course of psychiatric disorders in people with ID

9.1 Introduction

People with ID can experience all types of psychiatric disorders that occur in the general population. Some disorders occur more frequently in people with ID than in the general population, and some common disorders, in particular problem behaviours, are not often seen in the general population. Psychiatric disorders are common in people with ID, which is perhaps not surprising, given the complex mix of biological factors, psychological and social disadvantages, and additional developmental factors that people with ID have and are exposed to.

Psychiatric disorders are commonly overlooked in people with ID for a number of reasons including diagnostic overshadowing, complexity and multi-morbidity, reliance on carer reports, and differing presentations (see ▶ Chap. 5). A previously undiagnosed psychiatric disorder can be found in around 30% of adults, especially in those with more severe ID [1]. The term 'diagnostic overshadowing', which is commonly used, describes how the additional problems experienced by a person with ID tend to be misattributed as due to their ID rather than correctly attributed to co-occurring disorders [2]. Failure to identify additional disorders results in failure to offer interventions and care and so is a very important problem.

Psychiatric assessments are complex in people with ID, as psychiatric disorders tend not to occur in isolation. Psychiatric disorders can occur on top of existing long standing neurodevelopmental disorders such as autism spectrum disorder (ASD) or attention-deficit-hyperactivity disorder (ADHD) and co-occur together with physical disorders, impairments, and disabilities. Indeed, multi-morbidity is the norm for people with ID; assessments reveal that almost all adults with ID have multi-morbidity (two or more disorders as well as their ID) [3]. Other studies have found that even when the conditions were restricted to a limited number of physical and psychiatric disorders receiving input from general practitioners (i.e. already identified), the majority with ID had multi-morbidity, and the extent of multi-morbidity in young 20 years old adults with ID was similar to that in the 50- to 54-year-old general population [4, 5]. Additionally, a range of neurodevelopmental conditions tend to co-occur, for which the term ESSENCE has been coined (Early Symptomatic Syndromes Eliciting Neurodevelopmental Clinical Examination) [6]. ESSENCE includes major problems in: motor skills, general development, speech and language, social interaction and communication, behaviour, hyperactivity or impulsivity, hypoactivity, inattention, sleep or feeding difficulties [6]; genetic data also increasingly supports the ESSENCE clustering of neuro-developmental (including epilepsy) and psychiatric disorders [7]. The identification of co-occurring psychiatric disorders in persons with ID or ID

and co-occurring ASD requires specific clinical skills and specific tools, particularly to differentiate between psychopathological symptoms and behavioural changes due to other factors, such as physical pain, response to trauma, or overwhelmed neuro-autonomic, flight, or freeze reactions to environmental stressors (see ▶ Chap. 5).

Psychiatric disorders, physical disorders, and drug side-effects can mimic each other, so given the high levels of multi-morbidity, psychiatric disorders should not be considered in isolation. Some physical disorders relate to the persons underlying cause of ID, but lifestyle and environmental factors, sub-optimal support, and healthcare are also important contributors [8]. Multi-morbidity also results in polypharmacy and potential problems with disease-disease, disease-drug, and drug-drug interactions. People with ID may not be able to self-report drug side effects and are reliant on others observing these, so pharmacovigilance is essential. Anticholinergic burden can also be an issue due to polypharmacy and the strong anticholinergic effects of many psychotropic drugs. This has potential negative side effects such as further impairments of cognition [9, 10].

As well as multi-morbidity, assessments are complicated further by limitations in reciprocal communication and understanding, and decision-making capacity, visual and hearing impairments, and reliance on carer observations and reports, sometimes several carers/teams of carers. Together, this means that differential diagnosis is not only crucial but also complex, in order to establish the best treatment and care plan for the individual.

People with ID experience the full range and high rates of psychiatric disorders. These are often overlooked or misattributed to the persons' ID, causing unnecessary suffering. Psychiatric assessments are complex as, in addition to having ID, multi-morbidity is typical. Interventions need to be tailored to each individual's needs, taking account of physical health as well as the psychiatric disorders.

9.2 Prevalence of Psychiatric Disorders

The prevalence of problem behaviours will not be further considered within this chapter, as it is considered in depth in ▶ Chap. 7.

Studies differ in the prevalence of psychiatric disorders they report, due to the methods they use. In particular, differences in reported rates relate to the disorders included as psychiatric disorders, for example, whether or not problem behaviours and ASD are included along-side other psychiatric disorders, and also how representative the studied population with ID is. Given that the boundary between mild ID and the general population merges, particularly in adulthood, some studies report rates separately for people with mild ID from people with moderate-profound ID. Rates vary also by the age ranges included in the studies. Study results also vary for other reasons, such as the types of assessments completed and diagnostic criteria used, or whether information has been taken from existing records such as general practitioner health records, which are likely to underreport the extent of psychiatric disorders. Studies also vary in the size of the populations studied, therefore limiting conclusions that can be drawn, particularly for less common conditions. Some studies report point prevalence of psychiatric disorders, some report period prevalence, or lifetime prevalence, and some do not provide precise distinction between these, which also impacts on reported rates, contributes to variation in rates, and limits the validity of comparing rates between studies. Some authors have attempted to synthesise information on studies of prevalence of psychiatric disorders in children and young people [11] and in adults [12, 13], but these syntheses have inherent limitations in view of all the reasons given above, as the authors acknowledge. As one would expect, recent meta-analysis found that pooled prevalence is lower in population-based studies than in non-population-based studies, and in low overall risk-of-bias studies as compared to the moderate overall risk-of-bias studies [13].

Reported point prevalence rates of psychiatric disorders in children and young people with ID range from 30% [14, 15] to 50% [16], though it should be noted that these rates include problem behaviours. A study examining robust data from UK private household surveys reported a prevalence of 36% in 641 children and young people (aged 5–16 years) with ID [15]. This study was only of children/youth living in private households, and in view of the general population sampling frame, the individuals with ID had predominantly mild ID. A representative sample of US adolescents found that 65.1% has a lifetime prevalence for a psychiatric disorder [17].

For adults with ID, reported prevalence ranges from 14.5% (excluding problem behaviours, ADHD, ASD, dementia, and personality disorder, people aged 65 and over, and people with severe ID) [16] to 43.8% (adults with moderate to profound ID only) [18], depending on the conditions included and the quality assessment applied [16–21]. The largest adult population-based prevalence study in which each person (aged 16 years and over) was individually assessed included 1023 adults with ID [19]. Using robust methods, it reported a point prevalence of psychiatric disorders of 28.3% (or 40.9% if problem behaviours are also included). It reported rates separately for adults with mild ID at 25.4% and for adults with moderate to profound ID at 30.2% [19].

The effect of severity of ID on prevalence of psychiatric disorders is controversial, with some findings indicating higher rates in persons with severe/profound ID [22–24] and some others indicating the opposite [13, 25]. Some differences are accounted for by whether or not problem behaviours and/or ASD were included within the definition of psychiatric disorders, as they occur more commonly in people with more severe ID.

The effect of sex on prevalence of psychiatric disorders of any type is also currently not clearly understood. Most studies found no differences [13, 17, 26, 27], but some have identified a significantly higher prevalence among males than females [28]. Some of these discrepancies may be explained by whether or not the definition of psychiatric disorders included problem behaviours and/or ASD.

To date there is a lack of prevalence estimates of co-occurring psychiatric disorders identified using the new Diagnostic and Statistical Manual of Mental Disorders, fifth edition (DSM-5) [29] (and proposed International and Statistical Classification of Diseases and Related Health Problems 11th revision: ICD-11) definition of ID/disorders of intellectual development (see new ICD-11 term) [30], and differentiated by severity levels of adaptive functioning and specific (not general IQ) cognitive functioning [31]. Platt and collaborators found low adaptive behaviour to be associated with a higher rate of psychiatric disorders than low IQ, although results of their study have to be interpreted in light of several limitations [17].

Reported point prevalence rates of psychiatric disorders in persons with ID range from 30% to 50% in children and adolescents and from 14.5% to 43.8% in adults. Several factors account for the different rates reported, especially the specific types of conditions included in the definition of psychiatric disorder (e.g. whether ASD and problem behaviours are included), the type of sample, and the assessments used.

9.3 Prevalence of Psychiatric Disorders Compared with General Population

Presentation of psychiatric disorders can differ from that seen in the general population, and so the categories of psychiatric disorders as described in the standard psychiatric diagnostic manuals, the International and Statistical Classification of Diseases and Related Health Problems tenth revision (ICD-10) [32], and the DSM-5 [29] are not always easy for generalists to apply. For this reason, the manuals have been interpreted for use with people with ID, with the publication of DC-LD (Diagnostic Criteria for Psychiatric Disorders for Use with Adults with Learning Disabilities/Mental Retardation) [33], com-

plementary to ICD-10, and The Diagnostic Manual-Intellectual Disability 2 (DM-ID-2) [34], complementary to DSM-5. DC-LD is currently being revised into a second edition, in keeping with the publication of ICD-11. Whilst these differences pose challenges when comparing prevalence of types of psychiatric disorders between the population with ID and the general population (i.e. comparing like with like), work does suggest that similar broad categories of conditions do occur, with the addition of problem behaviours. For example, a study using exploratory and confirmatory factor analyses on two large datasets of psychopathology in adults with mild-profound ID ($n = 457$; $n = 274$) extracted a model of psychopathology with five factors: depressive, anxiety, cognitive decline, psychosis, and affect dysregulation-problem behaviour [35, 36]. The affect dysregulation-problem behaviour factor had good discriminate validity, face validity, and predictive validity over a 5-year follow-up period. This study provides a conceptualisation of problem behaviours within the broad over-arching family of psychiatric disorders, whilst being distinct from the other categories of depression, anxiety, cognitive decline, and psychosis, all of which resonate with the broad categories seen within the general population.

The study of 641 children and young people described above [15] reported a prevalence of psychiatric disorders including problem behaviours/conduct disorder of 36% in 641 children and young people aged 5–16 years with ID, compared with 8% of 17,774 children without ID [15]. Hence in this study, the children and young people with ID accounted for 14% of all children with psychiatric disorders, and their prevalence of psychiatric disorders was higher than for other children and young people for 27 out of 28 ICD-10 categories [15].

A recent study has drawn direct comparison between children/young people with ID and the child/youth general population, using the same methods for both, and appears to be unique in including a whole country sample, so is large, includes people of all ability levels, and is representative [37]. The study used data from Scotland's Census, 2011, which had an estimated 94% coverage of the whole country, and systematically enquired for everyone whether they had ID (and distinguished this from specific learning disabilities and from autism spectrum disorder) and whether they had a mental health condition. It included 5234 aged 0–15 years with ID and 911,097 without ID. Of the children/young people with ID, 12.8% had mental health conditions compared with only 0.3% of the children/youth without ID. Mental health conditions were significantly more prevalent at all age groups, children, youth, and adults; indeed ID (all ages) had an odds ratio of 7.1 in predicted mental health conditions. The disparity between the people with ID and those without was apparent at all age groups, but more so for the children and young people than for adults. Mental health conditions were also related to poorer general health status [37].

The study using Scotland's Census, 2011 described above, also reported on adults, drawing direct comparison between adults with ID and the adult general population, using the same methods for both [37]. It included 18,660 adults aged 16–64 years with ID compared with 3,470,078 without ID and 2455 adults aged 65 years and over with ID compared with 887,879 without ID. Of the adults with ID, 23.4% of the adults and 27.2% of the older adults had mental health conditions compared with 5.3% and 4.5% of the general population, respectively. Mental health conditions were significantly and substantially more prevalent at all age groups. Mental health conditions were also related to poorer general health status. Contrary to the general population, males rather than females were more likely to have mental health conditions [37].

A Swedish prevalence study specifically concerning older people had a large cohort of people with ID ($n = 7936$), aged 55 years or more and an age, and sex-matched cohort from the general population [25]. Information regarding psychiatric diagnoses was collected from the National Patient Register during 2002–2012. Seventeen per cent of persons with ID had at least one psychiatric diagnosis versus 10% of the general population cohort (OR 1.84). Most common diagnoses in the ID

cohort were 'other' psychiatric diagnoses (i.e. not included in any of the diagnostic groups) (10%) and affective disorders (7%), whilst the general population cohort was most often diagnosed with affective disorders (4%) and alcohol/substance-abuse-related disorders (4%). The highest odds for ID were found for psychotic disorder (10.4), ADHD (3.81), dementia (2.71), personality disorder (as a unique group) (2.67), affective disorder (1.74), and anxiety disorder (1.36) [25].

Despite the problems in comparing like for like between the population with ID and the general population, there is considerable evidence to show that most psychiatric disorders are more common in adults with ID than in the general population [23]. This includes schizophrenia [38, 39], bipolar disorder and mania [17, 40, 41], dementia (particularly in adults with Down syndrome [42, 43], but also in people with ID not due to Down syndrome [44, 45]), ASD [15, 46, 47], and ADHD [15].

Unipolar depression and anxiety are common in people with ID. Some research suggests they are more common than in the general population [17], whilst other studies do not [40, 48, 49]. It is perhaps surprising that unipolar depression and anxiety are common, given the high levels of mood stabilising drugs prescribed for people with ID: About 25% of people with ID have epilepsy [3] and so are prescribed anti-epileptic drugs, and whilst mood-stabilisation is not the purpose for their prescription in this situation (epilepsy), most do also have mood-stabilising properties. Epilepsy is much less common in the general population, at about 1% of the population, so the use of these drugs is much less prevalent. Longitudinal studies suggest that depression and anxiety may be more enduring conditions in people with ID than in the general population, although there are small numbers of people with ID within these longitudinal studies, and of those, almost all have mild ID, so are not representative of all people with ID [50–52].

There is considerable evidence to show that some psychiatric disorders are more common in children and adults with ID than in the general population, including autism spectrum disorder, ADHD, bipolar disorder, dementia, and schizophrenia. Depression and anxiety are also common; it is disputed whether or not they occur more commonly than in the general population, but given the high rate of usage of mood stabilising drugs (for epilepsy), it is notable that they have not been reported to be less common than in the general population.

9.4 Prevalence of Psychiatric Disorders in Persons with Co-occurring Intellectual Disabilities and Autism Spectrum Disorder

Persons with both ID and ASD may present several symptoms and deficits which are not seen in those with ID or ASD alone, and a different frequency of co-occurrent disorders [53], some of which may be accounted for by differing population characteristics such as a higher proportion of males and higher proportion with more severe ID.

As above mentioned, in persons with ID and ASD, one of the main issues in diagnosing co-occurrent psychiatric disorders is represented by the difficulty to distinguish between behavioural changes associated to psychopathological symptoms and those that can be attributed to physical, psychological, or environmental factors. In other words, behavioural responses to pain (including pain associated with sensory hypersensitivities), traumatic life events and life stressors (such as change and loss, both of which are frequent in the lives of people with ID and ASD), overlap with behaviours and symptoms that constitute criteria for psychiatric diagnoses. An essential part of diagnostic practice therefore working with people with ID and ASD is to include assessment of the support environment (e.g. for sensory triggers in ASD, whether expectations are appropriate or overwhelming), as well as examining the lived experience of the individual for past or present trauma circumstances that may be triggering intense emotional responses. In the absence of recog-

nising such life events, other psychiatric disorders (e.g. mood and anxiety disorders) may be diagnosed in error. Epidemiological studies of psychiatric disorders in ID and ASD have not routinely included trauma and adjustment-related diagnoses; the high prevalence of anxiety and mood disorders in this population may relate in part to these diagnostic errors [54].

Investigations on which types of instruments used to support direct clinical assessments are the most valid have been carried out, in particular with regard to screening tools and semi structured interviews (see ▶ Chap. 8). This point should be considered also whilst reviewing evidence produced so far. Instruments that do not consider the context of the individuals lived experience (e.g. screening instruments considering only behaviours and symptoms in the absence of considering what may have triggered these) will not be able to differentiate between psychopathology associated with unrecognised medical conditions, trauma, and adjustment-related conditions from other major psychiatric disorders.

Research shows an inverse relationship between IQ and severity of ASD, with rate of problem behaviours in ASD being lower at higher IQ [55–58]. Conversely, severity of ASD symptoms – and not that of ID – is directly associated with the rates of stereotypies [59, 60].

Although still limited and controversial, literature on the prevalence of psychiatric disorders in persons with both ID and ASD may indicate a higher rate of the full range of psychiatric disorders in comparison with those with ID or ASD alone [61–63]. Conversely, a study of adults with ASD compared rates with controls with ID matched for age, gender, ability-level, and Down syndrome [64]. They found that although the adults with ASD had a higher point prevalence of problem behaviours compared with the whole adult population with ID, there was no difference in prevalence, or incidence of either problem behaviours or other psychiatric disorders when compared with their matched controls [64]. They concluded that the differences in rates of psychiatric disorders were accounted for by other factors, including ability level and Down syndrome. The adults with ASD who had problem behaviours were, however, less likely to recover over a 2-year period than were their matched controls [64]. Greater agreement has been identified on higher rates of problem behaviours, stereotypies and rituals, communication difficulties, and social impairment in comparison to ID alone [57, 58, 65–67]. Nevertheless, specific investigation on causes and triggers is lacking and current prevalence might present some overlap issues with behavioural changes of non-psychopathological origin.

Furthermore, the co-occurrence of ASD with ID has been associated with a higher probability to be hospitalised and to receive a psychopharmacological treatment [68–70]. Lunsky and collaborators found that more than half of adults with ID and ASD have a co-occurring psychiatric disorder (26.1% mood disorder, 26.1% psychotic disorder, and 4.3% personality disorder), although they did not include trauma nor adjustment disorder. However, when compared with those with only ID, they did not show statistically significant differences except for the probability of the diagnosis of a psychotic disorder being lower in the co-occurring ID and ASD group [71]. In addition, individuals with problem behaviours and ASD were less likely to receive a diagnosis of schizophrenia, whereas the latter increased if problem behaviours were absent [66]. In all cases, the presence of problem behaviours had a negative impact on overall functioning and quality of life [61, 66, 69, 72–77].

The presence of ID in persons with ASD seems to negatively impact on the possibility of having an adjunctive diagnosis of psychiatric disorder. In a 10-year longitudinal study, Selten and collaborators found that the co-occurrence of ID in persons with ASD reversed the trend of a higher rate of diagnoses of bipolar and psychotic disorders than the general population [78]. Individuals with ASD who have greater cognitive abilities have been reported to have higher rates of depression than those with co-occurrent ID, which has simplistically been explained with a greater awareness of their difficulties [79–82].

Specific diagnostic procedure and tools seem to represent a significant determinant for the reliability of prevalence findings in persons with ID and ASD. These often include too many subjective symptoms which are not easy to identify in this group of patients and should be substituted by behavioural and observable equivalents (see ▶ Chap. 5). Different studies have found an increasing prevalence of psychiatric disorders as long as the specificity of their assessment tools rose. By using the Psychiatric Assessment Schedules for Adults with Developmental Disabilities (PAS-ADD), Thalen found an overall rate of 69.6% vs 8.6% in persons with ID alone [83], and through the Diagnostic Assessment for the Severely Handicapped-II (DASH-II), Matson and Cervantes found statistically significant differences in 8 out of the 12 subscales of the tools, on anxiety, mania, schizophrenia, stereotypies/tics, self-injurious behaviour, eating disorders, sexual disorders, and impulse control [63]. Nevertheless, the sensitivity for psychopathological features of some tools, such as the DASH, has not definitively been ascertained and high score reports may express mental distress due to other causes, including adaptive response to overwhelming stress.

Among specific psychiatric disorders, mood disorders [62, 83], anxiety disorders [26, 73, 83], somatic symptoms disorder, and impulse control disorders [61, 84–86] have been reported as the most frequent. Nevertheless, most of these study findings could be hampered by issues of multimorbidity and differential diagnosis.

Higher rates of inattention, hyperactivity, and impulsive behaviours have been observed in teens with both ASD and ID in comparison with those without ASD [87]. In addition, people with both conditions resulted to be more vulnerable to sleep problems, organic syndrome, stereotypies, and tics, although the psychopathological nature of this difference has not been confirmed [61].

The co-occurrence of psychiatric disorders has been found to be higher in males, in youth, and in persons with mild-to-moderate ID [66, 69, 71]. Thalen found psychoses and anxiety disorder to trend inversely to the severity of ID more than other disorders [83].

Some high rates of psychiatric disorders in persons with ID and ASD, such as that of bipolar disorder, seem to be related to behavioural dysregulation, as expressed by restlessness, agitation, or distraction, and may reflect the symptom overlap between psychopathological features, autism, and ID rather than a true co-occurrence of psychiatric disorders [26]. A subgroup of persons with ID and ASD, which shows adjunctive symptoms that cross diagnostic boundaries and include psychotic symptoms, excessive anxiety, mood lability, and information-processing deficits, seems to identify a complex neurodevelopmental disorder deserving a new diagnostic category [88]. Multiple complex developmental disorder (MCDD), multiplex developmental disorder (MDD) [89], and multidimensionally impaired disorder [90] are the terms and underlying models proposed for this category.

For psychotic disorders, most prevalence studies did not include adequate methods of assessment that distinguished from autistic features, psychotic-like symptoms, and symptoms of other psychiatric disorders, especially psychotic features of a post-traumatic stress disorder [91]. Psychotic-like behaviour such as talking to one's self, regression, and particular postures can be a common response of person with ID (with or without ASD) to stressful and other negative events [10], whilst bizarre expression or fantastic thinking can represent normal ways of experiencing the world of many persons with ASD. The link between Adverse Childhood Experiences and later psychosis increasingly identified in the general population during the last 20 years [92, 93] is being shown to occur also, or even more, in persons with borderline intellectual functioning, ID, and ASD [94–97].

Compared with the general population, people with co-occurring ID and ASD are markedly more likely to report a mental health condition. A whole population study included 5709 children and adults with co-occurring ID and ASD compared with 5,289,694 other children and adults in the general population [98]. After adjusting for age and sex, the odds ratio for a mental health condition was 25.6 or 130.8 when the interaction between age

co-occurring ID and ASD was also adjusted for [98]. Further study with the same data set reported on the relative independence of ID and ASD in contributing to the high odds ratio for a mental health condition compared with the general population [99]. It reported that 9396 of the 1,548,819 children had ID, 25,063 of the 1,548,819 children had autism, 16,953 of the 3,746,584 adults had ID, and 6649 of the 3,746,584 adults had autism. In the children, both ID (odds ratio of 7.0) and autism (odds ratio of 23.1) independently predicted mental health conditions. Also in adults, ID (odds ratio of 3.5) and autism (odds ratio of 5.3) both independently predicted mental health conditions [99].

Although still limited and controversial, the literature on the prevalence of psychiatric disorders in persons with both ID and ASD indicates a higher rate in comparison with those with ID or ASD alone.

9.5 Aetiology of Psychiatric Disorders

The aetiology of psychiatric disorders in people with ID is typically multi-factorial. Some specific causes of ID are associated with specific types of psychiatric disorders ('behavioural phenotypes'), for example, Down syndrome and dementia; other factors can also influence presentations of behavioural phenotypes. Multi-factorial aetiology can be considered on biological, psychological, social, and developmental dimensions and in terms of predisposing, precipitating, and maintaining factors. Aetiology has been represented graphically across the life-course, in detail, by Accessible Cause-Outcome Representation and Notation System (ACORNS) [100]. This shows that some of the multiple, interacting aetiologies are transactional; and linear cause and effect directions cannot usually be inferred. Considering aetiology is helpful in finding ways to prevent or reduce psychiatric disorders, or to aid recovery.

There are both similarities and additional complexity in aetiology of psychiatric disorders in people with ID compared with the general population. Similarities include that some psychiatric disorders have a degree of heritability so the person may have acquired the disorder even without having ID. Neurological disorders such as epilepsy are considered to increase the risk of psychiatric disorders in the general population, and whilst the ID literature is a bit mixed on this point, epilepsy is about 25 times more common in people with ID compared with the general population [101]. The addition of the developmental dimension adds complexity, including parental bonding and development of attachments at early age, impairments in attentional control, and communication limitations [102–104]. There are transactional effects of the person with ID and carers; psychiatric disorder in the person with ID affecting the carer-person with ID interaction, and carer stress and health, in turn further affecting the mental health of the person with ID; a spiralling vicious circle can be established [105]. Being dependent upon others in daily tasks can be restrictive upon what one can achieve and aspire to, which may also impact upon health.

Further complexity stems from people with ID having greater burden in all of the dimensions, such as the behavioural phenotypes of some causes of ID; additional physical disorders and disabilities; psychological factors associated with ID; and multiple social disadvantages. Abuse, neglect, or exploitation have been shown to predict psychiatric disorders in adults with ID [106]. Life events are more common for people with ID, and when they do occur, they tend to be multiple, for example, the death of a parent-carer leading to multiple changes in the person's life and care, move of home, and dislocation from former environment, family friends, and occupations [15, 19, 48, 107–109]. Lone parent family, poor family functioning, lack of parental educational qualifications, income poverty, and households with no paid employment have been shown to be associated with psychiatric disorders in children and young people with ID [15, 110]. Social disadvantages experienced by people with ID also include lack of paid employment, poverty of environment and recreational opportunities, social exclusion, and

experience of bullying, harassment, stigma, and hate crimes [111–114]. Aetiology of specific psychiatric disorders is considered in greater detail in the relevant chapters.

The aetiology of psychiatric disorders in people with ID is typically multi-factorial, with biological, psychological, social, and developmental dimensions and in terms of predisposing, precipitating, and maintaining factors. Some specific causes of ID are associated with specific types of psychiatric disorders.

9.6 Incidence and Course of Psychiatric Disorders

There has been less research conducted on the incidence and course of psychiatric disorders in people with ID, compared with prevalence studies. ► Chapter 7 considers the course of problem behaviours. The few studies on other psychiatric disorders suggest a more enduring pattern for psychiatric disorders than one might expect from studies with the general population, albeit with episodes of remission along the way. It is not clear if this pattern is due to greater severity of psychiatric disorders in people with ID, or poorer interventions/access to interventions, compared with the general population.

Regarding children and young people, a study followed a cohort of Australian children and young people with ID, aged 4–19.5 years, over 14 years. Hyperactivity was more prominent at younger ages and persisted for longer in the children and young people with more severe ID. Emotional disorders emerged later in childhood [115–117]. Similar findings have been reported from longitudinal studies with children in the Netherlands [118, 119].

Regarding adults, the incidence of mental disorders, excluding problem behaviours, has been reported to be 12.6% over a 2-year period, 8.3% for affective disorders, 1.7% for anxiety disorders, and 1.4% for psychotic disorders [39, 106]. Despite high mood stabiliser use (22.4%), 2-year incident mania has been reported to be 1.1% which is much higher than the general population, with a standardised incident ratio of 41.5 for first episode (or 52.7 excluding Down syndrome) [41]. Full remission of psychosis after 2 years was reported to be only 14.3% [39], poorer than it is for aggression or self-injury [120, 121]. At age 65 years or older, the standardised incidence ratio for dementia is reported to be 4.98 [122], showing it is much higher in adults with ID – it is even higher in adults with Down syndrome. These findings show that incidence is higher than that found in the general population for psychosis, bipolar disorder, and dementia, whilst much of the current high prevalence of common psychiatric disorders is due to enduring disorders, rather than new episodes. The evidence-base is, however, limited in quantity.

The few available studies on the course of psychiatric disorders in persons with ID suggest a more enduring pattern than in the general population.

9.7 Conclusion

In conclusion, psychiatric disorders are common in people with ID across the whole life-course. Point prevalence (excluding problem behaviours) is about 30% and may be higher in people who have co-occurring ID and ASD. Psychiatric disorders appear to be more enduring than in the general population, but there have been few longitudinal studies investigating the course of psychiatric disorders. Many factors contribute to the high rates. Given the extensive multi-morbidity that people with ID experience, and the added developmental dimension, assessments are complex, but important to get right, so that the most appropriate interventions can be offered to improve the person's quality of life.

Key Points

- Psychiatric disorders are common across the whole life-course, and much more so than in the general population.
- The point prevalence of psychiatric disorders (excluding problem behaviours) is about 30%; having ID increases the odds of psychiatric disorders by about 7 times compared with the general population.
- Schizophrenia, bipolar disorder, dementia, autism, and ADHD all occur more commonly than in the general population.
- The co-occurrence of ID and autism may be associated with a higher rate of the full range of psychiatric disorders in comparison with those with ID or autism alone.
- Aetiology is multi-factorial, due to a range of biological, psychological, social, and developmental factors.
- Psychiatric disorders may be more enduring than in the general population, but there are few studies on the course of psychiatric disorders.
- Psychiatric disorders typically coexist with other physical disorders and impairments, and assessments are therefore complex.

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Aetiology and Pathogenesis

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Contents

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Learning Objectives

- To understand the biological, psychological and social risk factors for the development of mental illness in individuals with an intellectual disability
- To understand how the diagnosis of mental illness in people with intellectual disability can be overshadowed, especially when there are comorbid conditions such as autism, ADHD, epilepsy and cerebral palsy

10.1 Introduction

Intellectual disability (ID) is defined as a developmental condition that affects both intellectual and adaptive functioning in conceptual, practical and social domains [1]. ID affects approximately 1% of the population [2]. ID is often classified into mild, moderate, severe and profound based largely on the results of IQ testing, with corresponding differences in life skills and the need for support. The majority of people with an ID have a mild ID [1]. There are multi-faceted explanations for ID and autism, from medical to cultural and spiritual. The cause of mild ID is most often unknown. Conversely, there is often a demonstrable cause for moderate to severe ID [3], for example, a chromosomal abnormality or perinatal complication [4]. It is expected that with increased uptake of genetic testing and increased resolution of genetic testing, previously unknown causes will be shown to have a genetic basis.

Language of the disorder has changed over the last 50 years, from mental retardation to learning disability and now increasingly intellectual disability, reflecting a change in wider societal attitudes towards, and understanding of, ID. In the relatively recent past, people labelled with 'mental retardation' were housed in institutions and were not believed to have the capacity to develop mental illness. Our knowledge and conceptualisation of ID and psychiatric illness is ever expanding. People with ID are not a homogenous group and an individual's risk of mental illness will vary based on constitutional and environmental factors. The reported prevalence of mental illness in people with ID differs owing to different methods of sampling and case ascertainment. The recognition and diagnosis of mental illness in people with ID can be complicated by atypical presentations and communication limitations. It is now understood that people with ID across the lifespan are at least as likely to experience mental health conditions as the general population [5–7], with certain groups being at particular risk [6, 7]. People with more severe ID or with lower ability levels have been found to have an even higher prevalence of mental ill health [6]. In addition, challenging behaviours that are unrelated to mental illness are present in a significant proportion of individuals with ID [8]. The impact of these behaviours can be significant, for example, increasing the need for costly support packages, potential exclusion from social activities and substantial caregiver burden.

The aim of this chapter is to discuss the factors that contribute to the development and maintenance of mental illness in ID and their interplay. We have followed a conventional bio-psycho-social approach.

10.2 Biological Factors

There are a plethora of biological risk factors for the development of a mental illness in an individual with ID, ranging from genetic predisposition to complications that occur in utero or around the time of birth. Significant perinatal complications which have led to an admission to a special baby care unit or neonatal intensive care have been implicated in the aetiology of ID [9]. Pregnancy and birth complications, as well as sometimes being a cause of ID, have also been associated with later development of mental illness, particularly schizophrenia, in people with ID [10].

As with the general population, certain mental illnesses can occur in multiple members of one family due to shared genetic and environmental risk factors [11]. A number of genetic syndromes are associated with ID, physical and mental illness, for example, Down syndrome. Chronic physical illness in itself can predispose an individual to certain

mental health conditions such as depression or anxiety [12]. For example, a meta-analysis performed by Anderson et al. [13] showed that the presence of diabetes doubled the odds of comorbid depression. In addition, certain physical health conditions can be mistaken for mental illness. For example, individuals with Down syndrome are at greater risk of developing thyroid disease [14] which can mimic affective disorder. This underscores the importance of accurate history taking and physical examination with appropriate investigations in people with Down syndrome.

Advances in genetic research have spawned interest in behavioural phenotypes, which are a particular cluster of cognitive, linguistic and behavioural profiles associated with specific genetic syndromes [15]. In addition, several genetic conditions are associated with specific mental illnesses. This information can allow us to predict and closely monitor for the development of particular psychiatric illnesses [15], although the nature of the link is not always clear (i.e. the mechanisms from genotype to phenotype have not been well defined). Examples of well-known associations are as follows:

1. *Prader-Willi syndrome (PWS) and psychotic disorder:* The most striking feature of PWS is insatiable appetite and food-seeking behaviour; individuals with the disorder may also have increased propensity to develop psychotic disorder. The additional risk seems to be confined to those with maternal uniparental disomy (accounting for approximately 25% [16] of cases) and the onset of the psychotic symptoms is in early adult life [17].
2. *Velo-cardio-facial (DiGeorge) syndrome and schizophrenia:* Associated features of velo-cardio-facial syndrome include palatal anomalies, cardiac anomalies and dysmorphic facial features. Psychotic disorders (most commonly schizophrenia and schizoaffective disorders) are far more common in children, adolescents and young adults than in populations without velo-cardio-facial syndrome that have been matched for IQ [18]. Schizophreniform disorders develop in up to 41% [19] of people with the syndrome.
3. *Down syndrome (DS)/trisomy 21 and Alzheimer's dementia (AD):* In addition to characteristic facial features, people with DS have associated physical health conditions such as cardiac defects, increased risk for leukaemia, autoimmune disorders, and by 50 years old, up to 55% [20] of those with DS have clinical features of dementia. Evidence suggests common pathogenic pathways between DS and AD, including the over-expression of the Amyloid Precursor Protein (APP) gene coded for on chromosome 21 which is important in the neurodevelopment of AD [21].
4. *Williams syndrome (WS) and anxiety:* Individuals with WS are known to appear as being hyper-social, outgoing and friendly. Despite this, they experience greater levels of anxiety than both individuals without ID and those with ID due to another cause [22] (■ Table 10.1).

In addition to our current understanding of genetic syndromes and their specific behavioural phenotypes, developments in molecular genetics have made it possible to study rare chromosomal copy-number variations (CNVs). CNVs have been implicated in the development of a broad range of complex conditions including ID, severe mental illness and autism [23]. As the resolution of genetic testing improves, it should one day be possible to test for CNVs in routine clinical practice leading to further discoveries. Wolfe et al. [23] found that up to 11% of new genetic diagnosis could be uncovered if chromosome microarray analysis was performed routinely. This is still a new and expanding field but with further research, geneticists should be able to uncover new genetic diagnoses with characteristic psychiatric phenotypes with potential implications for management. Overlap in the genetic risk markers (CNVs) for classically neurodevelopmental disorders (including Attention-Deficit/Hyperactivity Disorder (ADHD), Autism Spectrum Disorder (ASD) and ID) and those that had previously been considered neurodegenerative disorders (namely adult-onset schizophrenia and bipolar affective disorder) challenge existing psychiatric nosology and suggest shared pathways

Table 10.1 Examples of different syndromes with associated symptoms/behavioural phenotypes and associated psychiatric illness

Genetic syndrome	Psychiatric symptoms and behavioural phenotypes associated with syndrome	Psychiatric illness associated with syndrome
Prader-Willi syndrome – Chromosome 15 abnormality (paternal deletion, maternal uniparental disomy or imprinting defect)	Hyperphagia, skin picking, temper tantrums, aggression, hoarding, obsessional traits Sleep disturbance Stubbornness	Affective disorders often with psychosis Obsessive-Compulsive Disorder
Velo-cardio-facial (DiGeorge) syndrome – Deletion of genetic material from chromosome 22	Shy, withdrawn Problems in social relationships Concrete thinking Mood fluctuations	Schizophrenia spectrum disorders ADHD
Down syndrome – trisomy 21	Self-talk Particular deficits in working memory Sociable nature Age-related cognitive decline Aggression Stubbornness	Alzheimer's type dementia, atypical psychotic features, Obsessive-Compulsive Disorder, anxiety Depression
Williams syndrome – deletion of genetic material from chromosome 7	Autistic traits Overly friendly/gregarious Loquacious/'cocktail party speech' Hyperactivity	Generalised and social anxiety disorder Phobias/fears Obsessive-Compulsive Disorder

and the development of what had previously been considered discrete disorders [24]. How and why the same genetic variation is expressed differently remains undetermined but is an exciting area of research [11, 24, 25].

The development of mental illness in a person with ID is multi-factorial. Despite DNA sequencing technology being available and evidence of a strong inherited component to many psychiatric diagnosis, researchers have not been able to find clear genetic causes for most of these conditions [26]. Research into the way in which our environment can influence the development of mental illness and modulate the expression of genetic predisposition to disorders is helping us to gain new insights into the aetiology of mental illness in those with and without ID. Epigenetics describes the modification of gene expression (epigenome) that arises without altering the genetic code due to non-genetic influences, such as physical or emotional events [27]. Epigenetic mechanisms of gene regulation are composed of a complex, interconnected and plastic network comprising DNA methylation, post-translational modification of histones and non-coding RNAs [28]. Recent research on transcriptional activity at the molecular level has identified higher-order layers of regulation, including three-dimensional chromosome organisation, chromatin accessibility and RNA epigenetic modifications (epitranscriptome) [29–31]. Depending on the immediacy between an individual's life and the occurrence of changes in his/her epigenome, two main types of epigenetics are distinguished, *direct* and *indirect*. *Direct* epigenetics refers to changes that occur in the lifespan of an individual, due to direct experiences with his environment, while *indirect* epigenetics concerns changes that occur inside of the womb, due to events during gestation, or changes that affect the individual's predecessors, due to events that occur even long before conception and that are transmitted across generations [32–34]. Intrauterine epigenetic changes, called *foetal programming* (or *re-programming* in reference to the very

first weeks after conception), are hypothesised to constitute the most numerous and most important epigenetic changes for an individual's life and to represent a form of adaptive response to environmental stimuli, as to prepare an organism to tolerate negative external factors that may be encountered after birth [32, 35–38]. Intrauterine epigenetic changes can mark germinal cells and be transmitted to the offspring (imprinted genes) [35]. If, however, offspring's environmental conditions are markedly different than predecessors' ones, the offspring would be epigenetically mismatched and have a phenotype not appropriate for that environment. This could explain why highly stressful life circumstances or certain compounds such as alcohol or environmental toxicants, which profoundly alter the epigenetic make-up, could exert undesirable transgenerational effects and determine vulnerability to neurodevelopmental and psychiatric disorders [32, 39]. Likewise, the alteration of partial (alleles from both parents are differently expressed) and complete (complete suppression of one parent's allele) genomic imprinting of certain genes seem to influence the physiology of neural circuits and affect mental functioning and behavioural phenotypes [40].

Also foetal programming has been shown to link maternal pregnancy stress, exposure to toxic substances and viral infections with brain development and emotional reactivity of the offspring [35, 37, 41–43].

However, the epigenetic load accumulated over the course of an individual's lifetime might also bring risk factors for disease, as some direct epigenetic changes have been associated with predisposition to psychiatric disorders [26, 44–47]. The epigenetic relationship between stressful early life adversity and anxiety and depressive disorders has been extensively studied, with particular regard to the brain-derived neurotrophic factor (BDNF), hypothalamic–pituitary–adrenal (HPA) axis, serotonin transporter and FKBP5 (a critical regulator of the HPA cortisol response) genes [48]. Interestingly, variants in genes encoding for epigenetic modifiers have also been reported on [49].

In the last decades, the availability of high-throughput technologies has facilitated the generation of vast amounts of public epigenome-wide datasets that enable less biased and more integrative views on how the epigenetic regulation works in health and disease. The hope is that once we understand more about the interaction between genetics and psycho-social risk factors in influencing the susceptibility of mental illness, we will be able to think about new options for prevention and treatment of mental illness [50].

Several genetic syndromes show particular behavioural, cognitive and linguistic profiles related to specific mental illnesses. Epigenetics modification has also been associated with predisposition to psychiatric disorders, in both direct (during the person's life span) and indirect (during gestation or before the conception and transmitted across generations) ways. Epigenetic foetal programming links maternal pregnancy stress, exposure to toxic substances and viral infections with brain development and emotional reactivity of the offspring. Perinatal complications, chronic physical illnesses, sensory impairments, comorbidities and related medication can also contribute to mental health problems.

10.3 Psychological Factors

Vulnerability to the development of mental illness could be related to an individual's early life experiences and relationships. Attachment theory describes the notion that human beings are motivated to seek proximity to 'attachment figures', typically the primary caregiver [51]. An inconsistent and insecure relationship with such a figure could lead to the development of emotional problems and in some cases mental disorders due to reduced resilience in coping with stressful life events [52]. This theory has been influential in shaping our understanding regarding the psychological factors leading to the possible development of a mental illness, but research into

how the attachment theory could affect the mental health of people with ID has been relatively limited. The available research suggests that although attachment behaviours are generally similar to those without ID, they may be delayed or blunted in people with ID [53]. Another hypothesis is that the ID in itself could be experienced as a trauma, for both the parents at diagnosis and the child when they become cognitively aware of their diagnosis, and therefore affects the bond between a child and their attachment figure [54]. Reactive attachment disorder symptoms (unusual social behaviours including being withdrawn, disinhibited or overfriendly) has been found to be present in individuals with ID that suffered early childhood adversity, although symptoms seem to diminish with age [55].

The diathesis-stress model proposed by Abramson and colleagues [56] outlines the concept that psychological distress or the development of mental illness arises from the interaction between pre-existing (genetic) vulnerability to mental illness and stressful life events. Studies have supported the theory that stress is associated with the development of depressed mood in both the general and ID populations [57].

Individuals with ID are more likely to experience adverse life events such as stigma and discrimination, social exclusion, reduced employment and vocational opportunities and poverty [58]. Individuals with ID are also more likely to experience all types of abuse due to their vulnerability. A review of the limited available evidence into adverse life events in people with ID, performed by Martorell and colleagues [59], highlighted the importance of understanding and identifying the role of life events and traumatic experiences as predictors of psychopathology in people with ID. Adverse life events and psychosocial disadvantage have been linked to the development of psychiatric morbidity in young people and adults with ID [60]. Ali and colleagues [61] showed that higher rates of self-reported stigma were associated with higher psychological distress and service use. People that have experienced adverse life events may later go onto develop difficulties with processing their emotions which can manifest as interpersonal difficulties, presenting in some cases as emotionally unstable personality disorder or depression [62, 63]. Certain experiences which may not seem particularly traumatic to the general population could induce a post-traumatic reaction in a person with ID. For example, moving home may be experienced as a traumatic event for a person with ID as they may have no control over any aspect of the move and no choice about where they move to or who they will be living with. These experiences may be expressed in individuals with ID by an apparent increase in 'challenging behaviour' which in a person without an ID would be interpreted as anger or avoidance associated with Post-Traumatic Stress Disorder (PTSD) [59].

People with ID, specifically those with moderate ID [64], often feel that they have been discriminated against due to their ID. Ali and colleagues [65] described how this can result in barriers to accessing health care for several reasons, including poor communication. For example, a person can feel discriminated against if their health practitioner does not modify their communication in accordance with their needs, making it impossible for them to understand the salient points of the consultation [66]. Such discrimination can lead to feeling stigmatised, which impacts negatively on an individual with ID psychological wellbeing and can cause psychological distress [66].

The persistent exposure to uncontrollable, stressful situations can contribute to 'learned helplessness', that is, that a person 'learns' that he/she is helpless in many situations and does not attempt to try and change them, even when change is possible [67]. Learned helplessness can increase a person's risk of depression [68]. Although this can be a universal condition, those with ID are much more vulnerable [69, 70] since the tendency to overestimate their performance due to a lack of ability to recognise their deficits, difficulty in connecting their performance to internal causes (external locus of control[1]) and effectively judging their ability, and difficulty in

1 A belief that life is controlled by outside factors which the person cannot influence.

identifying the cognitive exhaustion state symptoms [69, 71].

Another vulnerability factor, conceptually associated and partially overlapped to external locus of control and learned helplessness, is represented by low environmental mastery. Environmental mastery refers to the ability to autonomously access and modify the surrounding context as well as being able to control events [72]. People with ID often receive excessive long-term support for all activities of daily living, which strongly limits the opportunities of successful experiences and the development of a sense of self-efficacy.

Psychological factors associated with psychopathological co-occurrences in persons with ID are also described within ▶ Chap. 3.

Adverse life events, including some experiences that may not seem overly traumatic to the general population, psychosocial disadvantage, poor coping skills, communication problems and issues with understanding and expressing emotions, have been linked to the development of psychiatric comorbidity in young people and adults with ID. Other psychological factors commonly associated with psychopathological causality are represented by the perception of being different and inferior to others or dependent on the support of others, poor self-esteem, negative self-image, low environmental mastery and learned helplessness.

10.4 Social Factors

Involvement in meaningful activities and living in a supportive environment is important for everyone, particularly those that have an ID, yet they may have more difficulty than those in the general population in achieving this [73]. Individuals with ID may have poor coping skills related to difficulties with problem solving and therefore require good support networks. Despite the increased levels of support that individuals with ID require, the subjective feeling of loneliness is a widespread problem in people with ID with reported prevalence rates of up to 45% [74]. A person with ID will often be socially isolated (only 6% of people with ID are in paid employment) [75], meaning they often only have a small social network mainly consisting of other service users, paid carers and family members [76]. There is a probable bi-directional relationship between loneliness in an individual with ID and physical and mental health problems [74]. Currently there is only limited evidence to suggest that loneliness in itself directly leads to the development of mental illness in the ID population, although there is some evidence to suggest that loneliness can contribute to a person with ID's existing predisposition toward mental illness [77]. Research in the general population indicates that loneliness, along with a psychological sense of belonging, can affect the experience of depression (biological, affective, behavioural and cognitive symptoms) more than the actual social support available [78].

Social factors associated with psychopathological co-occurrences in persons with ID are also described within ▶ Chap. 3 (■ Table 10.2).

People with ID are often socially isolated and discriminated or have a small social network which does not protect them from feelings of loneliness. Experiences of financial disadvantage are also frequent.

They may not be aware of their rights and be exposed to all kinds of abuse, for which they have to rely on the support of others. Their access to healthcare is often difficult and limited, and they have no control over any aspect of their living environment and no choice as to where they move or who they live with. This affects the possibility of self-determination and making decisions for one's life and health.

Table 10.2 Factors contributing to the development and maintenance of mental illness in persons with ID/ASD

Biological	Psychological	Social
Pregnancy and birth complications Pregnancy and birth complications, as well as sometimes being a cause of ID, have also been associated with brain damage and/or later development of mental illness, particularly schizophrenia	*Attachment factors* Inconsistent and insecure relationships with parental figures	*Social isolation* Social isolation, small social network (mainly consisting of other service users, paid carers and family members) and feeling of loneliness
Genetic factors Several genetic conditions show well-known behavioural phenotypes, associated with specific mental illnesses (e.g. Prader-Willi syndrome, Williams syndrome, Down syndrome, DiGeorge syndrome)	*Adverse life events* Adverse life events have been linked to the development of psychiatric morbidity in young people and adults with ID/ASD	*Economic disadvantage* Unemployment and economic disadvantage are common among people with ID/ASD, with all its consequences on mental health vulnerability
Epigenetic factors Direct (during the person's life span) and indirect (during gestation or before the conception and transmitted across generations) epigenetic changes have been associated with mental health issues	*Traumatic experiences* Proneness to be traumatised by life experiences is higher in persons with ID/ASD with higher impact on mental health. Life experiences that may not seem overly traumatic to the general population could induce a post-traumatic reaction in persons with ID/ASD	*Discrimination and stigmatisation* Discrimination by society can lead to stigmatisation in people with ID and affect their self-esteem and self-image
Physical illness Some physical illnesses, especially chronic physical illness, can predispose to certain mental health conditions such as depression or anxiety, or can determine various symptoms of mental disorders	*Difficulty to express emotions* People with ID/ASD often have difficulty to process and express their emotions, which cause in turn further psychological and relational issues	*Abuse* Individuals with ID/ASD are likely to experience all types of abuse They may not be aware of their rights and have to rely on the support of others to be advocates for their needs
Sensory impairment Sensory impairment can increase psychopathological vulnerability through a negative impact on many psychological and relational aspects	*Poor coping mechanisms* Individuals with ID/ASD may have poor coping skills related to difficulties with problem solving, managing frustration, anger and the consequences of their own behaviours	*Access to physical and mental health care* Persons with ID/ASD may face issues in accessing health care services for several reasons, including communication failure, setting inadequacy and lack of specific knowledge of the medical staff. This may negatively impact on self-determination and making decisions for their own health. This can also hinder early diagnosis and early intervention

Table 10.2 (continued)

Biological	Psychological	Social
Medication Side effects of psychotropic and non-psychotropic drugs can negatively impact on mental functioning, especially in case of polypharmacy and/or long-lasting treatments	*Self-worth* The aspects of life that society values (e.g. high social status, independence, employment, relationships and family) are those in which people with ID/ASD may have greatest difficulties, which can affect their self-esteem	*Living in inappropriate environment* Living in a pleasant and supportive environment is important for everyone, but persons with ID/ASD may have more difficulty than those in the general population in achieving this. They often have no control over any aspect of the living environment and no choice about where they move to or who they live with
	Self-image ID and ASD themselves may be experienced as a trauma, both by the person with the condition and their family. Persons with ID/ASD may feel different, inferior to others or dependent on the support of others and develop a poor or negative self-image that can contribute to mental health problems	
	Learned helplessness The persistent exposure to uncontrollable stressful and negative situations, which characterises the life of many persons with ID/ASD, can significantly reduce the individual's belief in their innate ability to achieve goals	
	Low environmental mastery Persons with ID/ASD often need and receive considerable and long-lasting support for many activities, including daily living activities. This limits the development of a sense of self-efficacy and internal locus of control	

10.5 Comorbidities

Diagnostic overshadowing occurs when the symptoms or signs of a physical or mental illness are incorrectly attributed to an individual's diagnosis of ID and hence potentially treatable causes for the presentation are neglected [79]. Overshadowing is even more likely when a person suffers from an ID and a comorbid condition, and this can further complicate the clinical picture. For this reason, it is important to be aware of common comorbidities and their association with mental illness to avoid delayed diagnosis or treatment. Psychopathological co-occurrences are the main object of the whole textbook the present chapter is included in. Heredown some short paragraphs are reported in reference to main co-occurring developmental disorders and neurological conditions.

10.5.1 Autism

Autism spectrum disorder (ASD) are a group of conditions that are characterised by a combination of social communication deficits, restricted interests and repetitive behaviours [1] (see ► Chap. 16). The prevalence of autism in those with ID is reportedly as high as 35% (in comparison to 1% in the general population) and is found to be higher in those with a lower verbal IQ or more severe disability [80]. Conversely, an IQ below 70 is found in at least

half of all people with autism [81]. There is therefore significant aetiological and diagnostic overlap between ID and ASD, leading to challenges in clearly distinguishing between the two conditions [82]. Owen et al. [24] proposed that ID, ASD and schizophrenia are connected and are part of the same neurodevelopmental disorder. He goes on to suggest that researchers need to reconsider the way in which they use diagnostic categories and focus on the developmental context of pathogenesis of certain syndromes, which could result in changes in the way in which psychiatric services are delivered.

Comprehension deficits experienced by an individual with ID may exacerbate the deficits in communication that an individual with ASD has, leading to even more challenges in diagnosing mental illness in this group. Buck et al. [83] describe how the presence of an ID makes identifying subjective symptoms of psychiatric disorders in people with ASD even more challenging due to limited expressive language resulting in those with comorbid ID and ASD being less likely to receive a diagnosis of anxiety or depression than those with ASD alone. An increased prevalence of physical aggression has been found to be associated with individuals with comorbid ID and autism [84], specifically self-injurious behaviour [85].

There is a large variation in mental illness prevalence rates in people with ASD, which may be due to difficulties in distinguishing between psychiatric illness and ASD clinically due to the conceptual overlap between the two conditions [86]. Depressive symptoms can also be misattributed to autism as symptoms such as flattened affect and social withdrawal occur in both conditions [87]. It can be difficult to distinguish the restricted and repetitive behaviours associated with ASD with the same symptoms present in obsessive compulsive disorder [88], again leading to diagnostic confusion and uncertainty. Similarly, the lack of conformity to social convention in people with ASD can be misinterpreted as mental illness. Other factors that contribute to difficulties in diagnosis of psychiatric illness in individuals with ASD include atypical psychiatric presentation in those with ASD, lack of validated instruments and lack of experienced clinical staff [89]. This emphasises the importance of taking a thorough developmental history and gathering collateral history.

Despite the variation in prevalence rates, psychiatric disorders have been found to occur frequently in adults with ASD. Mouridsen et al. [90] found rates of comorbid schizophrenia spectrum disorders to be as high as 35% in individuals with atypical autism. Conversely autistic-like traits and rates of ASD are higher in people with a diagnosis of psychosis than in the general population [91]. Recent studies suggest there are shared risk pathways between ASD and psychosis including a common genetic mechanism [92]. Anxiety is known to occur frequently in individuals with ASD [93]. Hyper- or hyporeactivity to sensory information is a known feature of ASD occurring in up to 96% of children with ASD [94]. This sensory over responsivity has been found to predict later development of anxiety disorders [95].

10.5.2 ADHD

Attention-deficit/hyperactivity-disorder (ADHD) is characterised by symptoms of inattention, hyperactivity and sometimes impulsive behaviours which interfere with functioning [1] (see ▶ Chap. 17). Pooled prevalence rates of ADHD indicate rates of 2.5% in adults without intellectual disability but higher rates in those with ID [96, 97].

Higher rates of a range of psychiatric illnesses have been found in people with ADHD [98]. The most frequent comorbid conditions are mood disorders, anxiety disorders, substance misuse disorders and personality disorder [99]. There are specific challenges in diagnosis of these comorbidities, for example, the emotional dysregulation present in ADHD could lead to misdiagnosis of a mood disorder or be labelled as challenging behaviour [100]. Equally an anxiety or mood disorder could be misattributed to ADHD symptoms. For this reason, rates of comorbid conditions vary widely in the available literature and this is an area which requires further research.

10.5.3 Epilepsy

Population-based studies of adult ID populations have found a prevalence of comorbid epilepsy to be in the region of 25% [101]. The prevalence increases with the severity of ID, occurring in 45% of those with severe ID in comparison to 15% with mild ID [102] (see ▶ Chap. 29). Up to 50% of people with comorbid ID and epilepsy have psychiatric or behavioural problems [103]. There is a complex relationship between the presence of behaviour that challenges and epilepsy in a person with ID. Although a link may exist, Blickwedel and colleagues [104] postulated that it is unlikely that epilepsy alone is the cause of challenging behaviour.

Epilepsy is associated with a range of psychiatric conditions in the general population including depression, anxiety, psychosis and personality disorders [105]. These illnesses are associated with the severity and chronicity of the epilepsy as well as type of epilepsy (increased in temporal lobe or refractory epilepsy) and adverse medication side effects.

10.5.4 Cerebral Palsy

More than 40% of children with cerebral palsy have a co-existing ID [106]. It is estimated that half of children with hemiplegia have a problem with behaviour, emotions or relationships including irritability, hyperactivity and anxiety, which has been associated with the underlying brain damage [107].

The risks of diagnostic overshadowing and misinterpretation of signs and symptoms increase along with the number and complexity of physical or mental illness that co-occur with ID/ASD. Mental health professionals must be aware of common comorbidities (i.e. ADHD, epilepsy and cerebral palsy) and their association with mental illness to limit the risk of diagnostic overshadowing and consequent delays in diagnosis or treatments.

Summary

In this chapter, we have discussed in detail the causes of mental illness in ID and their interaction. A conventional biopsychosocial framework was used to outline the causes and consequences of mental illness in people with intellectual disability. We have also discussed the challenges of diagnosing mental illness in individuals with ID and common comorbidities such as autism, ADHD and epilepsy.

Tip

Neurodevelopmental disorders frequently co-occur with other mental health problems to the point that some of them have been proposed to be part of the same neurodevelopmental disorder. Future research and clinical attention needs to reconsider the way in which diagnostic categories are used and focus on developmental context in the pathogenesis of mental illness.

Key Points

- Aetiology and pathogenesis of mental illness in ID is multi-factorial.
- There are biological, psychological and social risk factors for development of mental illness which combine to predispose an individual with ID to mental illness.
- There are several comorbid conditions which are associated with mental illness. One must be aware of overshadowing and consider assessment for mental illness.

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Psychopharmacology

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Contents

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Learning Objectives

- Rate of psychotropic use among adults with intellectual disabilities
- RCT-based evidence for individual psychotropic medications
- Polypharmacy
- High dose psychotropic use
- Adverse events of psychotropics
- Withdrawal studies of anti-psychotics

- A high proportion of people with intellectual disabilities receive medication for both physical and mental health problems.
- A high proportion receive off-label psychotropic medication for the management of problem behaviour in the absence of any psychiatric disorder.
- The evidence for the effectiveness of psychotropic medication to treat problem behaviour per se in people with intellectual disabilities is poor.
- On the basis of current evidence, the use of psychotropic medication could neither be supported nor refuted in this context.
- Non-pharmacological interventions should always be considered first or along with medication.
- A thorough assessment (including appropriate examination and investigations) of the causes and effects of problem behaviour followed by an appropriate formulation is a pre-requisite for using medication.
- People with intellectual disabilities and their carers should be involved in the decision-making process all along.
- Where appropriate other relevant professionals should be involved.
- Start with a low dose and increase the dose slowly modulating one medication at a time and avoid polypharmacy.
- Monitor effectiveness and adverse events associated with the intervention, and quality of life of the person with intellectual disabilities.
- At all subsequent follow-up assessments, consider withdrawal of medication and alternative non-medication-based management options.

Tip

Psychotropic medications are used commonly to treat problem behaviour in people with intellectual disabilities.

The evidence in support of their effectiveness is poor.

Non-pharmacological interventions should always be considered first for the management of problem behaviour in people with intellectual disabilities.

11.1 Introduction

11.1.1 Frequent Use, Polypharmacy and High Dose

Psychotropic medications such as anti-psychotics, anti-depressants, mood stabilisers including anti-epileptic medications and lithium, anti-anxiety medications including benzodiazepines, psychostimulants, beta-adrenergic blockers, opioid antagonists are used widely among people with intellectual disabilities (ID) [1]. The reported rate of use of these medications among people with ID varies between 32% and 85%; some of these studies have reported the rate of anti-psychotic use alone [2–11]. These medications are used in addition to existing high use of medication for physical problems (see ► Case Study 1) [12]. These medications are used for both mental illness and also problem behaviour (PB) for which they are not licenced [13]. A recent analysis of pooled data from the UK primary care (General Practice) database showed that of the 33,016 adults with ID, 49% (n = 16,242) received psychotropic medication between 1999 and 2013, and only 21% (n = 7065) had a diagnosis of mental illness but 25% (n = 8300) also had a diagnosis of PB [14]. In a previous study, Clarke and colleagues (1990) found that 36% of those who received psychotropic medication did not have a psychiatric diagnosis [15]. However, it is worth remembering that diagnosing both mental illness [16] and PB could be difficult in many people with ID [17].

Case Study 1: Frequent Use of Psychotropic Medication in People with Intellectual Disabilities

- Risperidone 2.5 mgs
- Carbamazepine 2000 mgs
- Sodium Valproate 2700 mgs
- Lamotrigine 400 mgs
- Lithium Carbonate 1200 mgs
- Methylphenidate 5 mgs
- Procyclidine 15 mgs
- Ferrous Fumarate + Vitamins + Lactulose + cod liver oil + various skin ointments
- PRN (as required medication): Clobazam + Lorazepam
- Rectal diazepam + buccal midazolam as rescue medication for status epilepticus.

The above is a list of a daily dose of medications for an adult with intellectual disabilities. This is not atypical for many people with ID. Notice the polypharmacy of psychotropic medication here (anti-psychotic + anti-epileptic + lithium + stimulant) and also the combination of three anti-epileptic medications at a high dose. Both sodium valproate and carbamazepine are used at a higher than recommended dose. If used simultaneously with sodium valproate, the dose of lamotrigine should be lower. If risperidone and anti-epileptics are already used, why is lithium needed to be added to control behaviour? Also, why a psychostimulant is used if already a number of psychotropics are used to control behaviour. If this is used for ADHD, why are the other psychotropics used for behaviour control? It seems that a number of psychotropic medications have been combined here in desperation without a proper formulation/rationale for this drug regime. Also, note that regular use of procyclidine with anti-psychotics can precipitate tardive dyskinesia in a person with an underlying brain damage. Also, note the prescription of a large number of medications here for medical conditions over and above the psychotropic use.

The rate of use of different psychotropic medications may vary from country to country. For example, in a recent community psychiatric clinic-based study of psychotropic medication use for management of PB in 100 adults with ID in the UK, 48% received new generation of anti-psychotics, 41% anti-epileptic medications (although in a high proportion of these participants, they were used for epilepsy treatment rather than any mental health problem and epilepsy can affect up to 25% of adults with ID [18]), 29% new generation anti-depressants, 27% old generation anti-psychotics, 9% anti-anxiety medications, 8% beta-blockers, 5% older generation anti-depressants, 4% psychostimulants and 3% lithium [19]. However, among Dutch population of 2373 adults with ID, de Kuijper and colleagues (2010) [9] found 32% received anti-psychotics (of these 65% received old generation and 23% new generation anti-psychotics), 58% of them received these medications for PB, 22.5% for psychoses (11.7% of them were diagnosed using DSM-IV criteria) [20], and in 18.5% of cases no reason was described in the patient records. In the same population, 17% also received anti-depressants and 20% benzodiazepines. Therefore, it appears that compared with the UK population, a higher proportion of participants received older generation of anti-psychotics and anti-anxiety medications such as benzodiazepine, but a lower proportion received anti-depressants in the Dutch population. However, the profile of psychotropic medication prescription found in these studies [9, 19, 21, 22] is similar to what was reported in a consensus survey in which the UK psychiatrists were asked to rank order their preference of use of psychotropic medication for the management of PB in people with ID, although most psychiatrists preferred to use a non-medication-based approach first [23]. Aman and colleagues' (2000) [24] international consensus study found a similar trend which preferred valproic acid and carbamazepine among the mood stabilisers/anti-epileptics. In the UK, it seems that the use of lithium as a mood stabiliser medication as well as for the management of PB is falling out of favour.

Reilly and colleagues (2014) [25] reported among individuals with ID in the UK and in Ireland that of four neurogenetic syndromes, psychostimulants were used among 21% of individuals with Fragile X syndrome (FXS), 3% with Prader-Willi syndrome (PWS), 4% with Williams syndrome (WS) and none with Velo-cardio-facial syndrome (VCFS). Risperidone was used among 7% of FRX, 6% of PWS and none of WS or VCFS individuals. SSRIs were used for 1% of FRX, 4% of PWS and none of WS or VCFS individuals. On the other hand, melatonin was used among 9% of FRX, 3% of PWS, 15% of WS and 8% of VCFS individuals.

The expectation that the use of psychotropic medication will decrease considerably once people with ID move out of long-stay institutions into the community has never materialised. All studies on this have found that the rate of psychotropic use has remained the same after discharge from institutions, some showing a decrease in anti-psychotic use at the expense of increased anti-depressant use, although overall there is an increase in the use of new generation than old generation anti-psychotics [26–29].

11.1.1.1 Polypharmacy

Despite the widespread condemnation of and lack of evidence for the use of polypharmacy (simultaneous use of more than one) of anti-psychotic medications, this practice remains widespread [30]. On the other hand, the merits and demerits of combining an anti-psychotic with another group of psychotropic medication specifically for the management of PB in adults with ID are currently unknown [31]. In Deb and colleagues' (2014) [19] study, however, the rate of polypharmacy of anti-psychotics alone is relatively low (10%), although the use of polypharmacy of all types of psychotropics remains relatively high (45%). Branford (1996) [32] showed that with an active programme, polypharmacy of anti-psychotics alone could be reduced, and in some cases, anti-psychotics could be withdrawn altogether or their dose reduced without any adverse consequences. However, most places lack such active programme for anti-psychotic withdrawal. However, it is worth keeping in mind that in a number of individuals where two anti-psychotics were prescribed simultaneously, the clinicians may be in the process of switching over from one to another.

11.1.1.2 High-Dose Anti-psychotics

Deb and colleagues' (2014) [19] study has also highlighted a trend towards the use of high-dose anti-psychotic medications in a number of adults with ID. According to the British National Formulary (BNF) (► bnf.org.uk), the usual maintenance dose of chlorpromazine for the treatment of psychoses is up to 300 mg a day, although a maximum dose of 1000 mgs has been recommended for exceptional cases. In Deb and colleagues' (2014) [19] study 23% of adults with ID received daily chlorpromazine-equivalent anti-psychotics dose of more than 300 mgs. Interestingly, these proportions are very similar to what was described in Branford's (1996) [3] study 13 years prior to Deb and colleague's (2014) [19], which reported the rate of anti-psychotic medication use in a population-based sample of adults with ID in the UK. In Branford's (1996) [3] study of the 198 patients who received anti-psychotic medication, 56 (28%) received at a daily dose over 300 mgs.

In Deb and colleagues' (2014) [19] study, the dose used for both risperidone and olanzapine was very low. However, the recommended daily dose suggested in the BNF is for the licenced indication for the treatment of psychosis. Therefore, it is anticipated that dosage will perhaps be lower when used for the management of PB. It is possible that in the case of anti-psychotics, clinicians feel that a lower dose acts as an anti-anxiety/anti-arousal medication rather than an anti-psychotic, with the primary intention of the behavioural management being to decrease the underlying anxiety/arousal.

11.1.2 Quality of Evidence

La Malfa and colleagues' (2006) [33] review on the use of anti-psychotic medications in people with ID identified 195 studies of which 127 were of type V, 44 of type IV, 9 of type III, 21 of type II and 4 of type I evidence.

One hundred and seventeen studies included old generation anti-psychotics and 78 new generation. Randomised controlled trials (RCTs) are seen as the gold standard for evidence in medicine and there are major challenges in involving people with ID in RCTs [34]. Mulhall and colleagues (2018) [35] in their recent systematic review summarised the main challenges as (a) difficulties recruiting, (b) obtaining consent, (c) resistance to the use of control groups (placebo), (d) engaging with carers, staff and stakeholders, (e) the need to adapt interventions and resources to be disability-accessible and (f) staff turnover. The authors felt that with reasonable adjustments, most of these barriers could be overcome. Another problem is with defining the clinical phenomenology of PB in order to be specific about the therapeutic objectives. The lack of RCT means that people with ID are deprived of receiving evidence-based intervention and as a result, clinicians' bias is rife in the day to day practice. Only 11% of the population in 100 RCTs of anti-psychotics for the treatment of PB included people with ID [36] as often disability in general and ID, in particular, is an exclusion criterion in pharmacological studies.

Therefore, the evidence presented in this chapter on the effectiveness of psychotropic medications has to be interpreted with caution. Most studies in this field are case reports on a small number of participants. It is known that studies with positive findings are more likely to be published than studies with negative findings. This is likely to create a reporting bias for the published case reports. There are only a few RCTs, but they often used a small cohort size, resulting in insufficient statistical power to draw firm conclusions. The outcome measures used are often not appropriate or validated. The method of selection of the control group and the experimental group is not always clear or appropriate, and outcome data are often not presented in an appropriate manner. For example, most studies neither quote the 'number needed to treat' (NNT) nor use analysis based on the 'intention to treat' (ITT). Most studies do not distinguish symptoms of psychiatric illness from those of PBs, and often researchers do not take into account the effect of co-morbid ASD and ADHD on PB symptoms. Also in many studies, participants with co-morbid psychiatric disorders were not excluded. It, therefore, remains unclear whether the psychotropic medications used in these studies treated the underlying psychiatric condition or the PB per se.

PBs are usually long standing; therefore, short follow-up periods used in most studies meant that it is not possible to know whether patients would derive any benefit in the long term. Only long-term follow-up will determine the effect of many confounding factors such as environmental changes, etc., that are concomitant with the use of psychotropic medications. Most studies do not take into account the confounding effect of concomitant non-medication-based management of behaviour, which may have a profound effect on the behaviour (although an adequately powered RCT should balance out these confounders in both groups). Similarly, in most studies, the anti-psychotics were used as add-on therapy, which made it difficult to tease apart the confounding effect of concomitant medications. For example, the use of anti-epileptic medications is common among adults with ID [37] and these medications may have an effect on the behaviour [38, 39]. However, an RCT design should take care of some of these confounding factors.

Another problem of interpreting the case report-based data is that many patients who showed improvement on a particular medication may have had an unsuccessful trial of other medications that have been shown to be effective in other case studies. Therefore, the individualised response to specific medication is always going to be difficult to determine. An individual's genetic make-up is likely to influence her/his response to a particular medication. There may be many causes of PBs among individuals with ID and many factors including medical, psychological and social may influence behaviour (see ▶ Chap. 7 on problem behaviour) (also see https://spectrom.wixsite.com/project). It is, therefore, imperative to carry out a detailed assessment of the causes and consequences of PBs before an intervention is implemented. However, none of the studies provides any detail of behaviour analysis. This sort of issue could be addressed by

including an overall quality of life (QoL) outcome measure. However, up until Unwin and Deb's (2014) [40] publication of Caregiver's Concern-Quality of Life Scale (CC-QoLS), no health-related QoL (HRQoL) existed for individuals with ID who manifest aggressive behaviour. Future studies should also assess the effect of interventions on family carers' burden and cost effectiveness [41, 42].

11.1.3 Indication

Although these cannot be seen as indications, psychiatrists usually consider psychotropic medications for the management of PB in individuals with ID under following circumstances: (a) failure of non-medication-based interventions, (b) risk/evidence of harm to others, property and self, (c) high frequency and severity of problem behaviours, (d) to treat an underlying psychiatric disorder or anxiety, (e) to help with the implementation of non-medication-based interventions such as positive behavioural support (with concurrent use of medication), (f) risk of breakdown of the individual's community placement, (g) lack of adequate or available non-medication-based interventions (although this should not be used as a rationale for using medication), (h) good response to medication in the past and (i) patient/ caregiver choice.

There is public concern regarding the use of psychotropic medication in individuals with ID for the management of PB in the absence of a diagnosed psychiatric disorder (see ► Case Study 2). Some of the reasons for this concern are: (a) perceived excessive use of medication (polypharmacy, use of higher than recommended dose of anti-psychotics) [43], (b) long-term use without reviews [13], (c) worry about adverse effects which could be difficult to assess, and inappropriate concomitant use of drugs to counteract adverse effects [19], (d) overall lack of evidence to support the effectiveness of psychotropic medications to manage PB [44–51], (e) out of licence use of psychotropic medications [52], (g) use of medication without explicit patient consent which often occurs in ID, (f) difficulty in assessing adverse effects in many individuals with ID, (g) difficulty in obtaining necessary investigations such as serum lithium level or blood tests for other adverse effects [53] and (h) difficulty in securing explicit informed consent in many cases [13, 53].

Case Study 2: Inappropriate Use of Psychotropic Medication.

A proprietor of a group home brought a 63-year-old gentleman with moderate ID to a psychiatric clinic asking for psychotropic medication to control his non-co-operative and anxious behaviour. Further examination revealed that the gentleman did not want to go to the day centre and became disturbed when he was forced to do so. When asked the proprietor of the group home told the clinician that they do not have a contract with social services department for payment of this gentleman's day care within the group home, therefore, they do not have a choice but to send him to the day centre. The clinician refused to prescribe psychotropic medication and asked the proprietor to sort out the contract issue with the social services department.

The general principles for using psychotropic medications for different psychiatric disorders among individuals with ID are similar to those used for the general population who do not have ID. As no specific evidence base exists to recommend specific medications for psychiatric disorders among adults with ID, the readers are directed to some of the generic guidelines on pharmacological management of common psychiatric disorders such as schizophrenia, depressive disorders, etc., such as the NICE guidelines (► www.nice.org.uk) and the Maudsley guide in the UK [54], the Frith Prescribing Guidelines in the UK [55] and others for specific application to individuals with ID [1, 56, 57]. However, the primary aim of this chapter is to discuss issues related to the use of psychotropic medications for the management of PB among people with ID.

11.1.4 Off-Label Use

When psychotropic medications are used for PB in the absence of a psychiatric disorder, they become the off-label or unlicenced use of a licenced medication. It may, therefore, be necessary to use a lower dose than what it is licenced for as the drugs may produce excessive adverse effects. For example, European legislation requires a Paediatric Investigational Plan (PIP) for all drugs that are marketed with indications for adults or the elderly. In Italy and in many other countries, off-label use is regulated by laws. The Italian Drug Agency (AIFA) specifies that 'if carried out in an appropriate manner, off-label use adds important therapeutic tools, especially for what concerns the therapy of rare diseases or involving population groups not normally included in common clinical trials'. When using medication out of its licenced indication, it is important to consider the following: (a) assess the risk of the medication in the ID population, (b) ensure that the medication has been shown to be safe for use in the ID population, (c) ensure that the evidence of efficacy is established in other population or in the ID population for other indications, (d) if necessary ask for a second opinion from a colleague who has experience of using the medication in the ID population, (e) involve as much as possible the person with ID, her/his family and other carers and professionals involved in the care, (f) make it explicit to relevant people that the medication is used outside its licenced indication and explain the rationale for its use and any potential risk involved and (g) make sure that the procedure for appropriate monitoring and follow-up is in place and adhered to.

11.1.5 Safety and Tolerability

The issue of safety and tolerability is of paramount importance in case of people with ID as they may have a different metabolism than the general population that may create different pharmacokinetics and pharmacodynamics. As a result, they may be vulnerable to develop more adverse effects than usual and also communication difficulties may make detection of adverse effects difficult. There is very little evidence available to guide clinicians on the safety and tolerability of psychotropic medication in the ID population. Therefore, the therapeutic drug monitoring remains an effective way to minimise the risk of adverse effects or toxicity. Some of the common and important adverse effects related to psychotropic prescribing in the ID population are listed below.

(a) Hyperactivity, restlessness, irritability and aggression are often observed with treatments with selective serotonin reuptake inhibitors (SSRIs) and sometimes benzodiazepines.
(b) Serotonin syndrome is a rare but serious adverse effect usually associated with the use of SSRI (particularly more than one SSRI). Symptoms include tachycardia, sweating, raised blood pressure and body temperature, dilated pupils, myoclonus, leading to shock. Treatment is symptomatic and immediate withdrawal of SSRI and if necessary, use of serotonin antagonist such as cyproheptadine.
(c) SSRI withdrawal may cause withdrawal symptoms in the form of increased restlessness and agitation. Therefore, it is necessary to start with a low dose and increase dose gradually. If necessary, restart slowly or use a different SSRI.
(d) New generation anti-psychotics can lead to obesity, metabolic syndrome and diabetes. These adverse effects, if not promptly treated, may bring on a significant increase in the risk of premature death.
(e) Disorders of the extrapyramidal system such as akathisia (often confused with agitation and improperly treated as such), oculogyric crises and Parkinsonian motor symptoms appear primarily during treatment with old generation anti-psychotics but may also occur from risperidone and other new generation anti-psychotics. These

symptoms may be misinterpreted as part of ID phenotype.

(f) Neuroleptic malignant syndrome (NMS) a rare but life-threatening adverse effect is associated with anti-psychotic treatment. Symptoms include raised body temperature, fluctuating blood pressure, muscle stiffness, sweating and other evidence of autonomic dysregulation. Muscle CPK is raised and treatment is symptomatic and immediate withdrawal of anti-psychotics.

(g) Constipation, if neglected, can cause severe distress (e.g. headache, depression, abdominal pain) which, in people with ID, may be expressed as sleep disorders, loss of appetite, agitation and aggression. This is mainly associated with treatments with tricyclic anti-depressants and some anti-psychotics.

(h) Anti-cholinergic syndrome, associated with the use of tricyclic anti-depressants and some anti-psychotics, may lead to agitation, motor restlessness, dysarthria, disorientation, hallucinations and convulsions. More severe symptoms include severe constipation, urinary retention, dry mouth, fever and tachycardia.

(i) Sedation, drowsiness and lethargy are associated with many psychotropic medications use.

11.1.6 Types and Rate of Anti-psychotic Adverse Effects

Mahan and colleagues (2010) [58] showed that among 80 individuals with ID in the USA, polypharmacy of psychotropic medication from different classes increased the likelihood of developing adverse effects. A number of measures have been used either in their original form or in adapted versions in order to assess adverse effects of psychotropic medications in individuals with ID. Some of these are Udvalg for Kliniske Undersøgelser (UKU) scale [59], Abnormal Involuntary Movement Scale (AIMS) [60], the Dyskinesia Identification System Condensed User Scale (DISCUS) [61], and Barnes Akathisia Rating Scale-Revised [62]. Some scales were specifically designed for use among individuals with ID such as the Matson Evaluation of the Drug Side Effects (MEDS) [63], CLAMPS [64], Monitoring of Side Effects Scale (MOSES) [65].

One recent Dutch study found that 84.4% of 103 adults with ID and PB had at least one adverse event and 45.6% had over three adverse events. Presence of adverse events had a significantly negative influence on the person's quality of life. Respectively 13% of the patients who did not receive psychotropic drugs (23% of all participants) and 61% of the patients treated with more than two psychotropic drugs had more than three adverse events [66]. Matson and Mahan (2010) [67] reviewed the studies on adverse effects due to psychotropic medication use among individuals with ID, and Matson and Hess (2011) [68] did the same for individuals with ASD. As expected, most studies are on adverse effects of anti-psychotic medications as they are the most widely used psychotropic medication for individuals with ID. Most studies reported adverse effects associated with old generation anti-psychotic medications such as haloperidol, chlorpromazine, etc. For the old generation anti-psychotic medications, extrapyramidal adverse effects such as Parkinsonism, akathisia and tardive dyskinesia are reported in most studies. One recent Dutch study of 99 individuals with ID who were treated with anti-psychotics found extrapyramidal symptoms in 53%, overweight or obesity in 46% and the metabolic syndrome in 11% of participants. In addition, hyperprolactinaemia was present in 17% and the evidence of abnormal bone metabolism in 25% of participants. Increasing age and more severe ID were associated with dyskinesia and a higher dosage of the anti-psychotic medication was associated with Parkinsonism. Less severe ID was related to higher Body Mass Index and the use of atypical anti-psychotics was also associated with a higher diastolic blood pressure [69].

Anti-psychotic-treated people with ID reported to have a significantly higher incidence of movement adverse effects particu-

larly Parkinsonism and akathisia (and possibly the rare NMS) when compared with the anti-psychotic-treated non-ID general population [70]. It is not always clear whether some of these abnormal movements are caused by the underlying brain damage or the medication or the combination of the two as studies show that individuals with ID are more prone to develop these symptoms [58, 66, 71]. However, within the ID population, the rate of these symptoms is higher among those who are treated with anti-psychotic medications than those who are not. For example, in Mahan and colleagues' study of 80 in-patients with ID in the USA, abnormal involuntary movements were recorded among 30% of those who did not take any psychotropic medication compared with 86.7% of those who took those medications [58]. In Ganesh and colleagues' (1989) [72] study among 66 in-patient adults with ID in the UK who were treated with anti-psychotic medication, five (7.6%) developed akathisia and 18 (27%) developed tardive dyskinesia. However, it is also reported that severity of ID is a risk factor for tardive dyskinesia [71], perhaps indirectly implying the role of underlying brain damage in the causation of these symptoms. A similar argument exists as to whether schizophrenia itself rather than the anti-psychotic medications per se is a risk factor for some of these adverse effects, particularly glucose intolerance.

There is also debate as to whether the new generation of anti-psychotic medications are less prone to cause adverse effects in general but extrapyramidal symptoms in particular when compared with the old generation anti-psychotic medications. In general, it has been shown that the new generation anti-psychotic medications produce less adverse effects particularly extrapyramidal symptoms than the old generation medications, although these adverse effects have also been reported among individuals with ID who have been treated with risperidone and also olanzapine. There are also anecdotal reports of risperidone causing serious adverse effects such as neuroleptic malignant syndrome (NMS) in individuals with ID. However, for the new generation anti-psychotic medications, the metabolic syndrome including weight gain, diabetes mellitus, change in lipid profile and hyperprolactinaemia are the most commonly described adverse effects. Only a handful of studies have assessed these symptoms in individuals with ID (see next section). However, interestingly a number of studies have reported dyskinesia and other extrapyramidal symptoms upon withdrawal of anti-psychotic medication in a high proportion of individuals with ID [73, 74] (60% in one study) [67, 75] (see ► Case Study 3). This phenomenon may suggest that these symptoms may sometimes have been suppressed by the use of anti-psychotic medications.

Case Study 3: Appearance of Dyskinesia After Withdrawal of Anti-psychotic Medication.

A 54-year-old lady with mild ID who has a long history of paranoid ideas and crying behaviour became confused and developed oro-facial dyskinesia after withdrawal of long-standing risperidone. These symptoms improved after re-instating risperidone.

There is also concern whether anti-psychotic medications have an effect on cognition, particularly among children. Aman and colleagues (2009) [76] have reviewed this subject. At least one RCT [77] did not find any effect of risperidone on cognition of children with ID when compared with the effect of placebo. On the other hand, Stratta and colleagues' (2013) [78] review has shown that in the non-ID general population, administering anti-psychotic medication in an individually optimised manner seems to have the potential of improving the cognitive aspect of schizophrenia, regardless of the kind of anti-psychotic medication used. Similar findings were reported for children with ID who were treated with risperidone for PB [76].

The main concern of using the new generation anti-psychotic medications is the adverse effect of metabolic syndrome. Zuddas and colleagues (2011) [79] reviewed the adverse effects of newer anti-psychotic medication including metabolic indices among children

and adolescents including those with ASD but not necessary with ID. Based on the review of RCTs, the authors showed that adverse effect of weight gain is worst with treatment with olanzapine and best with aripiprazole with intermediate effect from risperidone and quetiapine. Indeed, Deb and colleagues' (2014) [51] systematic review on aripiprazole has shown that adverse effects such as hyperprolactinaemia had improved in a number of individuals with ID when switched from other anti-psychotics such as risperidone to aripiprazole. A number of studies assessed the adverse effects of metabolic syndrome/disorders associated with newer anti-psychotic medications in individuals with ID [80–86]. Frighi and colleagues (2011) [86] observed the rate of metabolic syndrome among 138 anti-psychotic-treated and 64 anti-psychotic naïve individuals with ID and compared that with a non-ID general population that was treated with anti-psychotic medication. Among individuals with ID, 48% received risperidone, 18% olanzapine, 10% thioxanthenes, and 24% other anti-psychotic medication. Ninety-five percent of individuals with ID received monotherapy of average duration of 8 years and of an average chlorpromazine equivalent daily dose of 108 mgs (range 16–667 mgs). Metabolic indices were the same or more favourable in individuals with ID compared with the non-ID general population. However, overweight/obesity and type 2 diabetes were more prevalent among women with ID than the non-ID control group. There were no statistical or clinical differences in the metabolic indices between the anti-psychotic-treated versus anti-psychotic naïve individuals with ID. However, there was a non-significant trend towards a higher rate of type 2 diabetes in the group that received anti-psychotic medication.

It is not clear whether individuals with ID are more prone to develop metabolic syndrome compared with the non-ID general population irrespective of anti-psychotic treatment. Among Taiwanese population, Chang and colleagues (2012) [87] reported that metabolic syndrome and obesity are common in adults with ID. de Winter and colleagues (2011) [88] reported that among 412 elderly Dutch population with ID over age 50 years, a weighted prevalence of metabolic syndrome was significantly higher (25.1%) than the non-ID population (15.7%) of the same age, with a significantly higher risk for individuals with mild ID. On the contrary, Hsu and colleagues (2012) [89] reported a lower rate of metabolic syndrome among 164 institutionalised (overall rate of 11.6, with 8% in males and 17.2% in females) individuals with ID compared with the non-ID general Taiwanese population. The authors defined metabolic syndrome on the basis of the presence of three or more of the following; (a) central obesity, (b) elevated blood pressure, (c) elevated fasting glucose level, (d) elevated triglycerides and (e) reduced high-density lipoprotein (National Institute of Health, USA, 2002) [90].

11.1.7 Withdrawal Studies

A recent systematic review showed that withdrawal particularly of anti-psychotic medication is possible in a proportion of people with ID (4–74%) (*see* ▶ Case Study 4) [91]. However, most of the studies included in this systematic review are from the USA that included patients from long-stay institutions [32, 92–94]. We have found that there were two studies in the UK [32, 95] and one recent one from the Netherlands [96] where a concerted structured effort was made to withdraw anti-psychotic medication. In Branford's (1996) [32] study, 25% (31/123) achieved a total withdrawal, and in 42% (52/123) of cases, an attempt to withdraw or reduce dose-precipitated problem behaviour that led to reinstatement of anti-psychotics. In Ahmed and colleagues' study, 33% (12/36) [95] achieved complete withdrawal, and another 19% (7/36) achieved at least a 50% reduction in dose. In de Kuijper and colleagues' (2014) [96] study, 44% (43/98) achieved a complete withdrawal, but in 16% (7/43) of cases anti-psychotics were reinstated at 12 weeks follow-up, so 37% (36/98) remained off anti-psychotics at 12 weeks.

There is another recent open label discontinuation study from the Netherlands [97]. In this study, of 129 participants, 61% had completely discontinued anti-psychotics at 16 weeks, 46% at 28 weeks and 40% at 40 weeks. In 49% of participants, behaviour deteriorated at 16 weeks follow-up leading to re-instatement of medication. In our recent study in Cornwall in England, we have managed to withdraw anti-psychotics totally among 46.5% (33/71) and reduced dosage by at least 50% in another 11.3% (8/71) of adults with ID. At 12 weeks follow-up, no one required hospital admission or change in placement [98].

Case Study 4: Successful Withdrawal of Anti-psychotic Medication

A 19-year-old young man with moderate ID and ASD became aggressive towards his parents who asked for psychotropic medication. He was prescribed risperidone at 0.25 mg daily oral dose which was increased to 0.25 mgs twice daily dose with good effect on the behaviour. However, further assessment revealed this young man's obsession with water and fear of noise. After a few months of behaviour modification, his behaviour improved when his risperidone was gradually withdrawn without any deterioration in behaviour.

11.1.7.1 Withdrawal Symptoms

Some studies showed either no change in behaviour or improvement after withdrawal of anti-psychotics in the majority of cases [91, 94–96] but others showed worsening of behaviour in a higher proportion of cases [32, 99]. A number of studies showed that withdrawal of anti-psychotics (particularly the old generation ones like chlorpromazine, haloperidol, thioridazine) may precipitate extrapyramidal symptoms (mostly demonstrated by increase in DISCUS score), particularly dyskinesia and akathisia [75, 91, 95, 100–102]. These symptoms might be misinterpreted as recurrence of the original problem behaviour [103]. Rebound akathisia might appear within the first few days, whereas rebound parkinsonism usually emerges after a week and rebound dyskinesia might only become apparent within a month [104, 105]. However, most studies show that DISCUS score tends to come back to baseline after a few weeks and months and dyskinesia improves at a follow-up [96, 106]. This is an indicator for the clinicians who are considering withdrawal of anti-psychotics that instead of re-instating anti-psychotics straight away because of the resurgence of PB after withdrawal of anti-psychotics, they should wait (if necessary with the help of as necessary PRN prescription) until the behaviour improves (Deb et al., 2009, WP) [107] (see ▶ Case Study 5 and 6) (see ◘ Fig. 11.1).

However, other possibilities also need to be considered. For example, behaviour may deteriorate for reasons that are not at all related to the withdrawal process, so one should not automatically relate deterioration of behaviour to the dose reduction or withdrawal. Instead a thorough assessment of behaviour should be carried out (see ▶ Chap. 7). Sometimes an underlying psychiatric disorder may be unmasked upon withdrawal of medication or dose reduction, particularly if the medication has been used for a long time. A careful assessment of mental state will be necessary in that case as making a diagnosis of a psychiatric disorder in a person with ID could be difficult (see ▶ Chap. 7). In some cases, previous behaviour may return upon dose reduction or withdrawal. In these circumstances, a full functional assessment of the behaviour will be necessary including the assessment of pre-disposing, precipitating and perpetuating factors (see ▶ Chap. 7).

It is worth keeping in mind the so-called 'placebo' or 'nocebo' effect in this context. For example, some people with ID or their carers may have severe anxieties about the withdrawal process as they are fearful that upon withdrawal of medication, the person's behaviour may deteriorate and they do not want to upset the *status quo*. Because of this effect, some carers may have an exaggerated perception of the severity of the emergent problem behaviour.

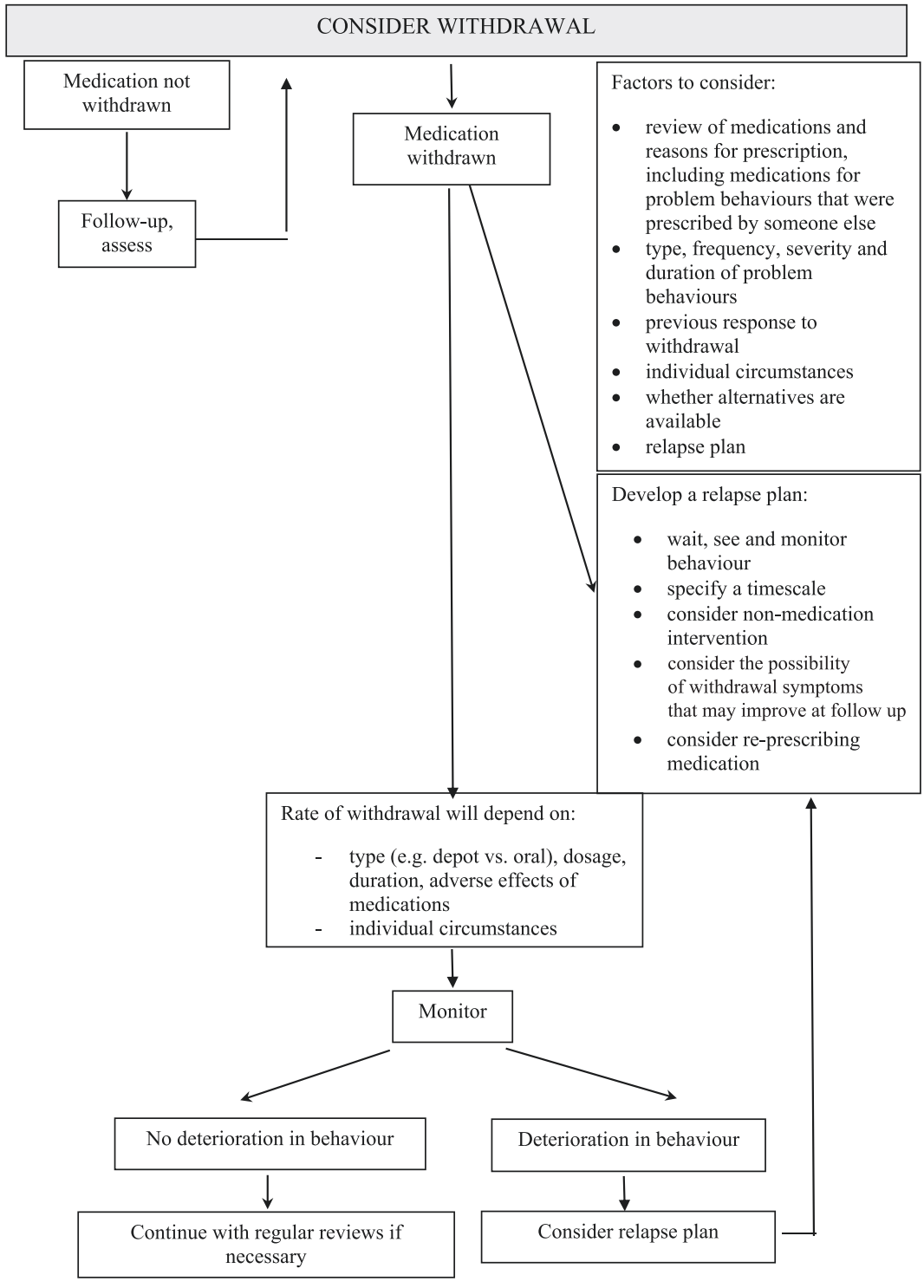

Fig. 11.1 Flow chart for considering withdrawal of medication. (Adapted from Deb et al., 2009; ▶ http://www.ld-medication.bham.ac.uk)

Case Study 5: Unsuccessful Withdrawal of Anti-psychotic Medication

A 68-year-old gentleman with mild ID who developed dementia was treated with risperidone for many years because of history of aggressive behaviour. After 3 months of gradual withdrawal of risperidone, the man became physically aggressive on one occasion in an evening club. The care staff panicked, and local police were called out at night, which led to re-instating of risperidone by an emergency doctor.

Case Study 6: Successful Withdrawal of Anti-psychotic Medication

A 26-year-old man with severe ID who had no speech became disturbed when his risperidone, which he received for many years, was gradually withdrawn. The care staff became very anxious and there was pressure on the clinician through his general practitioner for re-instating risperidone. However, the clinician explained to the care staff that sometimes after years of use, drug withdrawal may produce rebound PB, sometimes caused by withdrawal dyskinesia. The man was treated with PRN (as required) medication over the next 3 months and his behaviour gradually settled without requiring any regular medication.

11.1.7.2 Factors Affecting Withdrawal

A number of factors have shown to affect withdrawal. For example, Branford (1996) [32] found that a lower dose of anti-psychotics, minimal psychopathology, lack of aggression, stereotype and hyperactivity at baseline helped with the successful withdrawal. Ahmed and colleagues (2000) [95] on the other hand highlighted the environmental and organisational factors. For example, they suggested that experienced and full-time staff in regular employment, low staff turnover, staff training, courses for managing problem behaviour, less reliance on environmental restrictions are likely to facilitate the withdrawal process. de Kuijper and colleagues (2018) [97] found that female gender, a lower rate of baseline problem behaviours and lower baseline dosage are in favour, and the presence of severe behaviour and autonomic and extrapyramidal symptoms at baseline are factors against a successful withdrawal. de Kuipjer and colleagues (2018) [97] also found that the presence of co-morbid autism, a higher dose of anti-psychotic drug, higher behaviour rating and akathisia scores, and more-frequent worsening of health during discontinuation were associated with a lower incidence of complete discontinuation. These factors also affect re-instatement rate. For example, Janowsky and colleagues (2006) [99] found within a large population in an American residential institution, 66.3% (55/83) of individuals remained anti-psychotic-free almost 10 years after withdrawal. However, in a subsequent study, Janowsky and colleagues (2008) [100] demonstrated that unfortunately, it becomes difficult to withdraw anti-psychotic medications altogether in a very high proportion of those who showed worsening of behaviour after one or two attempts of withdrawal.

A number of authors have highlighted the influence of staff perception on withdrawal studies (see ► Case Study 7) [101, 102, 106, 108]. Greater restriction and lesser scope of adaptation within the living environment, poor staff training and organisational policies and both clinicians and care staff's lack of confidence in dealing with transient behaviour change upon withdrawal of medication are also some of the important factors [95]. Proper training and support for care staff is thus of paramount importance for a success with an withdrawal programme. Deb and colleagues have recently developed an online staff training programme called SPECTROM (Short term PsychoEducation for carers to Reduce Overmedication of people with intellectual disabilities) to help with the reduction of overmedication of adults with ID [109, 110].

Case Study 7: Carer Attitude Influencing Psychotropic Prescribing

A proprietor of a community group home brought a 32-year-old man with moderate ID, who does not speak, to the psychiatric clinic. The person has been on multiple psychotropic medications for several years including a number of anti-psychotics at a high dose along with a regular dose of procyclidine. Examination revealed that the man had severe tremor and dyskinetic body movements including movements in facial muscles. When the clinician proposed a reduction in the number of psychotropic medications, the proprietor became very anxious, suggesting that the man has a history of very difficult behaviour and he needs all his medication otherwise he would become very aggressive. It was also suggested that the movement disorders were nothing new, therefore, indirectly suggesting that they could not be harmful and may be part of his intellectual disabilities. However, the clinician eventually went ahead with the reduction in the medication and told the proprietor that medicines regulatory bodies will raise concern if medication reduction is not tried and the license for the group home may be at stake.

11.1.7.3 Need for a Structured Pathway

It is clear that in order to succeed with the withdrawal of anti-psychotics, it is imperative to follow a structured pathway, which includes a withdrawal plan presented in ◘ Fig. 11.1 and proper consideration of initiation of medicinal treatment as per the International Guideline (see ◘ Table 11.1) recommendations [107]. Furthermore, the clinicians need to consider complex factors involved in prescribing which will affect the withdrawal process such as variables related to patients, treatment modalities, type and dosage of medication, environment which may not be appropriate for the person, and other resources, including appropriate psychosocial support structure around the person as well as their caregivers. In the process of withdrawal, clinicians also need to consider factors mentioned earlier in the text that may influence successful withdrawal, including carer training and organisational support structure. Apart from our recent paper on anti-psychotic withdrawal [98], no other study so far described a structured pathway for withdrawal.

11.1.8 Outcome Measures

The outcome measure is part of a quality assessment system that also includes structural and process indicators. In addition, the comparative assessment of the efficiency of health interventions is a fundamental strategy to promote quality and equity of care. A myriad of factors influences the outcome and its measures in the treatment of mental health in people with ID. For example, the level of abilities, communication skills, level of support may all affect outcome measurement. Outcome measures may involve an assessment of rate and severity of target symptom/behaviour, adverse events, the burden of care, patients and their family's quality of life, patient-related outcome and process outcome. As the intervention must be person-centred so an assessment of the quality of life (QoL) is of paramount importance. For example, an intervention may improve symptom but producing harmful effect may reduce rather than improve the patient's quality of life. The term effectiveness describes the combined effect of the efficacy of an intervention with its safety/tolerability and adherence to the intervention. The mental health services for people with ID overlap among health care and social care, general and specialised health care, services for children and adults. Therefore, adoption of a holistic, multidimensional, bio-psycho-social approach to health care when it comes to treatment of mental health in people with ID is an ideal option. Among the person-centred outcome measures, the QoL measures are gathering momentum in the individualised planning of the interventions, in the implementation of

Table 11.1 Recommendations for good practice prescribing for PB for people with ID [170]

The prescriber needs to ensure that a thorough assessment using a bio-psycho-social approach has been conducted and recorded prior to initiating treatment	The prescriber should provide the person and/or their family or carers a written treatment plan at the time of prescribing
The prescriber should ensure that an appropriate formulation is carried out and a treatment plan drawn, prior to instigating any intervention	The method and timing of the assessment of treatment outcome should be set at the beginning of the treatment
The prescriber needs to ensure that appropriate physical examinations and investigations have been carried out	As far as possible, there should be an objective way to assess outcomes (the use of standardised scales is recommended)
The prescriber is responsible for assessing the person's capacity to consent to treatment	The prescriber should ensure that follow-up assessments have taken place
The prescriber should discuss the formulation and treatment plan with the person and/ or their family or carers	As far as possible, one medication should be prescribed at a time
The prescriber should allow the person and/or their family or carers to influence the decisions that are made and included in the treatment plan	Start with a low dose and increase the dose gradually until improvement or appearance of the adverse effect
The prescriber should clarify to the person and/or their family or carers if the medication is prescribed outside their licenced indication. If this is the case, they should be told about the type and quality of evidence that is available to demonstrate its effectiveness	As a general rule, the medication should be used within the recommended dose range
Where possible, and when necessary, the prescriber should discuss the formulation and treatment plan with other relevant professionals	Consideration for withdrawing medication and exploring non-medication management options should be on-going
The treatment plan should be part of a broader care plan that takes a person-centred approach	The prescriber should remember that medication might be used at the same time with non-medication management
The treatment plan must comply with the country's legal framework, including the relevant Mental Health and Capacity Act	The prescriber should document all appropriate information and share it with appropriate individuals when necessary
The formulation and treatment plan should be shared with all the relevant parties, including GPs, as soon as possible	The prescriber should discuss with the person and/ or their family, carer or key person common and serious adverse events related to the treatment (where possible, they should provide accessible information in writing)
The prescriber should identify a key person who will ensure that medication is administered appropriately and communicate all changes to the relevant parties	The prescriber should advise what action to take if a serious adverse event takes place
The consultation should take into account the communication needs of the person	When 'as required' medication is prescribed, the prescriber is responsible for providing as much information as possible about why and when the medication may be used and should monitor this regularly

clinical trials and in the organisation of services [40, 108, 111]. A list of commonly used outcome measures for PB in ID is provided in ▶ Chap. 7 on Problem Behaviour.

11.1.9 Ethical Issues

When treating people with ID, informed consent and compliance are two important aspects of both research and clinical practice. These involve issues such as personal freedom, confidentiality, and understanding on their part, particularly in the context of cognitive impairment. Very rarely people with ID are involved in the decision-making process. Similarly, taking consent through deception or misleading information or by use of physical force, of any type of coercion may not only be unethical but may be illegal. Similar ethical concern applies regarding the covert use of medication, particularly concealed in food. Compliance with psychological and behavioural therapies is also often low, especially for the use of concomitant psychotropic medication. One of the main reasons seems to be the prejudice and stigma associated with ID, and as a result, people with ID are either marginalised or patronised. This is reflected in the common practice whereby the professionals and clinicians ignore the person with ID in front of them and speak about them with their caregivers. One key problem is gathering informed consent from a person with a severe cognitive impairment. It is necessary to use accessible information or pictures in order to facilitate communication with many people with ID. Professionals involved in the care of a person with ID should always document the assessment of capacity and ability to provide informed consent to the proposed intervention. In the absence of this capacity, a consensus should be gathered as close as possible between the multidisciplinary team, the legal representative of the person with ID, the family and other significant figures of relationship. The decision should be made in the best interests of the person with ID.

One practical example of this is the use of lithium, which when started is very difficult to withdraw. Lithium also has a narrow therapeutic window leading to toxicity easily for which regular monitoring of blood tests is necessary, which is often not possible in many people with severe/profound ID thus raising an ethical issue. However, in the absence of capacity, it is a difficult decision to implement this intervention, particularly where other alternative medications such as anti-epileptic mood stabilisers are available.

11.2 Pharmacological Classes

11.2.1 Anti-psychotic Medications

11.2.1.1 Old-Generation Anti-psychotics

There are a number of old controlled trials involving old generation anti-psychotics [33, 112]. Some of them have been summarised here. Wardell and colleagues' (1958) [113] 3-month RCT on 41 adults with ID showed a marginally worse response on chlorpromazine (average dose 400–600 mgs/day) when compared with the placebo. Weir and colleagues' (1968) [114] 12 weeks study of randomisation of 45 in-patients with ID into pericyazine vs. chlorpromazine vs. placebo (doses are unknown) did not show any intergroup significant difference in PB outcome. Vaisanen and colleagues (1975) [115] used sulpiride (150–300 mgs/day) vs. chlorpromazine (25–50 mgs/day) vs. placebo in 60 in-patients with ID in a cross-over trial for 4 weeks. Behaviour was changed towards a more positive direction when sulpiride replaced placebo as per the global assessment scale only ($p < 0.05$). Sulpiride was found to be as effective as chlorpromazine ($p < 0.1$). Elie and colleagues' (1980) [116] study of 51 adults with ID showed placebo was superior to thioridazine in improving PB. Aman and colleagues (1989) [117] randomised 20 children and adults in institutions in a cross-over trial to receive either haloperidol high dose (daily 0.05 mg per kg body weight) or low dose (daily 0.025 mg per kg body weight) or placebo for 3 weeks. Haloperidol was found to be marginally better than placebo in high dose. Malt and colleagues (1995) [118] in a cross-over trial among 31 adults with ID showed

that zuclopenthixol (5.5 mgs/day) was better than haloperidol (1.5 mgs/day). Singh and Owino (1992) [119] showed efficacy of zuclopenthixol tablets in a double-blind study of a small number of in-patients with ID and PB.

11.2.1.2 New-Generation Anti-psychotics

Risperidone

There are three RCTs of risperidone among adults with ID [43, 120, 121]. Two of these studies showed that risperidone was significantly better than placebo in improving PB but Tyrer and colleagues' study [43] did not find a significant difference in outcome among groups treated with risperidone, haloperidol and placebo. There are six RCTs among children with ID with or without ASD [77, 122–126]. RUPP study (2002) [125] and Shea and colleagues (2004) [126] primarily included children with ASD, some of whom also had ID, whereas Aman and colleagues (2002) [124] and Snyder and colleagues (2002) [77] primarily included children with ID but excluded those who had ASD. Of these four studies, only the RUPP study (2002) [125] was not sponsored by a pharmaceutical company. McDougle and colleagues (1998) [127] in an RCT showed that some core symptoms of ASD improved significantly in the risperidone group compared with the placebo group. Many of these children also had ID and some showed PB such as aggression as well. All these studies showed a significant improvement in PB in the risperidone group compared with the placebo group.

There was a significant reduction in scores for the risperidone compared with the placebo group on the Clinical Global Impression-Improvement (CGI-I) scale [60], Aberrant Behaviour Checklist (ABC) [128, 129] and Visual Analogue Scale (VAS) to rate the most troublesome target behaviour ($p < 0.05$). A total of 109 (54.5%) participants were rated as 'much improved' or 'very much improved' at endpoint on the CGI-I in the risperidone treated groups ($n = 200$) and 24 (11%) in the placebo groups ($n = 218$). Our meta-analysis showed that the overall Number Needed to Treat (NNT) to achieve a rating of 'much' or 'very much improved' on the CGI over a period of 4–8 weeks of treatment with risperidone versus placebo is 3 with a range between 2 and 5 [47]. However, main concern about using risperidone is its adverse effects such as somnolence and weight gain (not much evidence is available from the RCTs on other adverse effects such as metabolic and cardiac).

Three of the RCTs involving children were continued for many weeks using open label designs [82, 83, 130]. These studies showed that the efficacy of risperidone had been maintained over 52 weeks and medication adverse effects were by and large tolerable. Croonenberghs and colleagues' (2005) [131] open-label one-year follow-up study of 504 risperidone-treated children with ID reported long-term efficacy and tolerability. Seventy-three percent of the children completed the study with a mean dose of 1.6 mgs/ day risperidone. The most common adverse events were somnolence (30%), rhinitis (27%), headache (22%) and weight increase (17.3%). Overall 91.7% had shown some adverse events. The incidence of movement disorders was low, and no clinically significant changes in mean laboratory values were noted, except for a transient increase in serum prolactin levels. Improvement in behaviour which was first observed within the first week of treatment initiation was maintained at the follow-up and significant improvements were also noted in positive social behaviour.

Aripiprazole

Deb and colleagues' (2014) [51] systematic review on the efficacy of aripiprazole in the management of PB among individuals with ID and with or without ASD reported two RCTs that included 75 and 218 children, respectively (age 6–17 years), with ASD and with and without ID in a parallel design RCT over 8 weeks period [132, 133]. Both these studies have been carried out by the pharmaceutical company that produces aripiprazole, and it is not clear whether there is any overlap among the participants in these two studies. In both studies, the diagnosis of ASD was confirmed by the Autism Interview-Revised (ADI-R) [134] assessment and both studies included children with the ABC-I score [129] of >=18 and CGI-S >=4 [60]. Both studies excluded children with a diagnosis of Fragile

X syndrome and any other major psychiatric disorder such as schizophrenia, depression and bipolar disorder. The first RCT gave flexible doses of aripiprazole with the average daily dose being 10 mgs [132]. There was a significant improvement in the ABC-I score (score change at follow-up: −12.9 in the risperidone group vs. -5 in the placebo group) and a significant mean improvement in CGI-I score (2.2 or much improved in the risperidone vs. 3.6 or no change in the placebo group). The second study used fixed doses of aripiprazole [133]. According to the ABC-I score, the aripiprazole groups showed significant improvement compared with the placebo group (score change at follow-up: −12.4 for 5 mg, −13.2 for 10 mg, −14.4 for 15 mg aripiprazole, respectively, compared with −8.4 for the placebo group). CGI-I showed statistically significant improvement for all the aripiprazole groups compared with the placebo group. Most studies reported adverse effects from aripiprazole in the form of weight gain, increased appetite, sedation, tiredness, drooling and tremor. However, aripiprazole improved serum prolactin level in some participants and overall did not show any adverse effect on QTc interval. There was an open label extension of one study [135] following the RCT that included 330 children, which showed that the improvement shown in the aripiprazole group during the RCT lasted for 52 weeks and the adverse effects were tolerable [136]. Deb and colleagues (2014) [51] have also found another 17 studies of either prospective or retrospective case reports that reported improvement in most people with ID treated with aripiprazole. However, there is a need for more carefully designed RCTs into the use of aripiprazole in the management of PB in individuals with ID and/or ASD conducted independent of pharmaceutical companies.

Other Newer Anti-psychotic Medications

Singh and colleagues (2010) [137] reviewed studies on clozapine for the management of PB in individuals with ID. They included 13 small studies, of which only three seem to be controlled studies with a very small number of participants of one, two and three, respectively. However, Boachie and McGinnity's (1997) [138] study was not included in this review, who examined case notes and medication charts of 17 individuals with ID who received clozapine 200–900 mgs/day with an average range of treatment for 32.1 months. According to Singh and colleagues' (2010) [137] review, the dose range of clozapine in different studies varied from 10 to 900/day, the average being 300–400 mgs. Most studies are retrospective case notes review that included a small number of participants. All studies reported improvement according to subjective measures such as CGI and about one third of those who used rating scales have shown improvement according to a rating scale although most studies did not use any rating scale. Adverse events reported included extrapyramidal symptoms, vomiting, drowsiness, weight gain, hypersalivation, tachycardia, hypotension as well as hypertension, urinary incontinence, leukopenia (reported in one study) and seizures (reported in one study that used 10–300 mgs daily dose).

Janowsky and colleagues (2003) [139] reported a retrospective study on 18- to 55-year-old individuals with ID (45% were male), the majority of whom had severe to profound ID. Olanzapine 2.5–22.5 mgs/day (mean dose 9.1 mgs/day) was used as an add-on for up to 6 months. Aggression reduced in 93% ($p < 0.01$) and SIB in 86% ($p < 0.044$) of 14 participants. Adverse effects were sedation (20%), constipation (10%), gait problem (5%) and weight gain. No increase in serum glucose levels was seen. McDonough and colleagues (2000) [140] reported that olanzapine reduced the stereotyped form of chronic SIB in four of seven individuals with ID. Williams and colleagues (2000) [141] treated 12 adults with mild to moderate ID who had a variety of reasons for giving olanzapine, including psychosis, PB, adverse effects of other medications and inadequate treatment responses. The authors found that 58.3% of the participants 'greatly improved', a proportion essentially same as for those who improved on risperidone treatment.

La Malfa and colleagues (2003) [142] in a prospective case series on the use of quetiapine 300–1200 mgs/day for 6 months on 15

individuals with ID (age 25 to 47 years; 66.7% male) reported significant improvement, particularly among those who had mild to moderate ID.

Cohen and colleagues (2003) [143] have reported adverse effects from ziprasidone (145 mgs/day) among 40 individuals with ID who received other anti-psychotic treatment previously. There was a reduction in behaviour frequency after ziprasidone treatment among 19 individuals who showed mild to moderate frequency of PB, whereas six individuals who showed a high frequency of PB did not show much change after ziprasidone treatment. However, ziprasidone treatment was associated with a significant weight loss of 3.6 kg as well as significant reduction in total cholesterol and triglycerides ($P <= 0.5$). This drug is withdrawn from the UK market for its serious cardiac adverse effects.

There is only one small retrospective chart study of asenapine on PB in people with ID who had psychiatric disorders [144].

11.2.2 Anti-depressants

The evidence on the use of anti-depressants for the management of PB in adults with ID is scant. One RCT on clomipramine showed beneficial effects but the cohort size was very small (n = 10) [145]. However, the response to the SSRIs has been varied whereby some studies reported clear favourable results [146–150], some showed negative effects [150–152], and other studies demonstrated both positive and negative outcomes [153, 154]. King and colleagues (2009) [155] did not find any significant difference in efficacy between citalopram and placebo in a large sample of children with ASD and a high level of repetitive behaviour. This discrepancy in findings, therefore, makes it difficult to draw any definite conclusion regarding the effectiveness of anti-depressants in this context [156].

Improvements were largely reported in SIB and perseverative/compulsive behaviours. It may, therefore, be the case that medications were in effect treating underlying behaviours that are part of the obsessive-compulsive disorder (OCD) spectrum for which SSRIs are indicated anyway. Not surprisingly the anti-depressants were most effective in the management of PB when depression or anxiety was present in the background. In a number of cases, deterioration in behaviour is reported, which may have been caused by the adverse effects of some of the anti-depressants. In general, the majority of the evidence based on open label trials and case series studies is fraught with methodological concerns. The small sample sizes meant that the studies were statistically underpowered and often control groups were not recruited. There was a dearth of validated outcome measures utilised, and where more than one assessor conducted the outcome measurements, inter-rater reliability was not contemplated.

The efficacy of anti-depressants certainly deserves more attention in research, as there is evidence to suggest that these medications are used commonly in the management of PB in individuals with ID [19]. This review does not suggest that they are ineffective but that there is not enough good quality evidence for their efficacy at present.

11.2.3 Mood Stabilisers (Lithium and Anti-epileptic Medications)

Deb and colleagues' (2008) [46] systematic review revealed three small primarily cross-over RCTs of lithium of which only two were published in a peer-reviewed journal [157, 158] and the other one was published as a book chapter [159]. As the participants in these studies were all in-patients, it is difficult to generalise these findings for individuals with ID who live in community settings. Although all these studies showed that a proportion of participants treated with lithium improved, it was not clear whether this improvement was significant compared with the placebo group and also the outcome measures used were not validated and standardised. Also, some studies showed the effectiveness of lithium on PBs but not on others. Therefore, it is difficult to draw any definitive conclusion on the efficacy of lithium for the treatment of PB in people

with ID. There are other concerns regarding the use of lithium such as the need to carry out regular blood tests, which may not be possible for a number of people with ID, narrow therapeutic range between effective and toxic serum level, lithium toxicity and a high chance of relapse if lithium is withdrawn. There is little evidence of the use of lithium by the UK psychiatrists for the ID population [3, 19, 23].

Among other mood stabilisers such as antiepileptics, only a small RCT involving 10 clients is available on the use of carbamazepine [160] and the rest of the studies are case series. There are a small number of small RCTs and non-controlled studies [161, 162] of sodium valproate primarily on children and adolescents with ASD, some of whom also had ID showing some effect on behaviour but nothing is conclusive at the moment [163]. Although there has been anecdotal evidence suggesting that lamotrigine may make behaviour worse in people with ID, a small RCT did not show any change in PB in participants with ASD who were treated with lamotrigine [164] (see also a recent systematic review and meta-analysis by Hirota and colleagues, and Limbu and colleagues [165, 166]). Similarly anecdotal concern about levetiracetam making behaviour worse in people with ID was not always supported [167]. There are no RCTs to support the use of topiramate in people with ID/ASD and PB [168, 169]. Although there is no RCT-based evidence in support of mood stabilisers, that does not mean that these medications are ineffective. As mood stabilisers such as carbamazepine and sodium valproate are used regularly to treat PB in people with ID [23, 53], there is an urgent need to carry out properly designed RCTs involving these drugs notwithstanding the practical difficulties of carrying out RCTs in this population [34, 35].

11.2.4 Opioid Antagonists

The opioid antagonist drug naltrexone has been used to treat PB in children with ASD with or without ID. A systematic review by Roy and colleagues (2014) [49] has found 10 studies that used an RCT design. Two studies used a parallel design ($n = 59$) and the rest of the studies used a cross-over design. The number of children included in these studies ranged from 4 to 20. Less than half of the studies showed a statistically significant improvement in irritability and hyperactivity in the naltrexone group compared with the placebo group, but none has shown any significant effect on the core symptoms of ASD. Most studies either did not observe any adverse effects or report any. Adverse effects included sedation, loss of appetite, vomiting and stereotyped behaviour and in some cases paradoxical increase in aggression.

Roy and colleagues (2014) [50] in another systematic review included 10 cross-over trials of naltrexone for the treatment of primarily SIB in adults with ASD with or without ID. The number of participants included ranged from 4 to 24 and the study period ranged from 4 to 17 weeks. Only two studies found a significant improvement in the naltrexone group compared with the placebo. The dose of naltrexone varied from 50 mgs to 150 mgs per day or 0.5 mg to 2 mgs per kg body weight. Eleven (9%) out of overall 124 participants included in these 10 studies reported minor adverse effects. Adverse effects included weight loss, loss of appetite, mild liver function abnormalities, sleep problems, etc.

There are only a handful of RCTs on naltrexone that included a small number of participants over a relatively short period of follow-up using different doses and crossover design, which has its drawbacks. The findings are equivocal in that some showed a beneficial effect from naltrexone and the others did not. One study showed differential effect depending on the dose, particularly the higher dose being effective and lower dose being non-effective. Therefore, it is difficult to draw any definitive conclusion about the efficacy of naltrexone in the treatment of SIB and other PB among adults with ASD and/or ID.

11.2.5 Anti-anxiety Medications

Findings from a small number ($n = 3$) of small scale uncontrolled studies of buspirone for treatment of PB in adults with ID are mixed [163]. King and Davanzo's (1996) [170] pro-

spective uncontrolled study of 26 adults with ID (age 25–63 years; 46% male) on the effect of buspirone 25–60 mgs/day (average 52 mgs/day) on aggression and/or SIB in people with ID did not show any improvement from buspirone. A recent systematic review and meta-analysis has found two RCTs on buspirone showing some equivocal results on the ASD symptoms of children [156]. There is little evidence currently to recommend any anti-anxiety medication for the long-term management of PB including aggression in people with ID. The benzodiazepine group of medications carries the risk of tolerance and dependence in the long run. In some cases, benzodiazepines may paradoxically increase aggression. The evidence for the effectiveness of buspirone is currently poor, therefore, cannot be recommended. However, for the general population, some SSRIs, selective noradrenaline reuptake inhibitors (SNRIs), an anti-epileptic medication pregabalin and an anti-psychotic quetiapine are now recommended treatment for anxiety-related disorders [171]. In the field of ID, some anti-psychotics are prescribed in a smaller than anti-psychotic dose to manage PB with the assumption that at a lower dose anti-psychotics may work as anti-anxiety medications, although the evidence to support this assumption currently is not available from the literature.

11.2.6 Beta-Blockers

Ward and colleagues (2013) [48] have published a systematic review on the use of beta-blocker medications such as propranolol, nadolol, acebutolol, metoprolol and oxprenolol for the treatment of PB in individuals with ID. They found 14 studies (nine prospective studies and five retrospective chart reviews) primarily on propranolol, dose ranging up to 340 mgs a day, which included between one and 19 participants. The target symptoms included aggression, SIB, irritability, hyperactivity, impulsive behaviour, etc., and in one case anxiety, and in another case somnolence. Target behaviour improved in a number of cases, in some cases the improvement was moderate, and in some cases, there was no improvement, and in a minority of cases, behaviour deteriorated. Not all studies mentioned the proportion of patients reporting adverse effects. Common adverse effects included bradycardia, hypotension, vomiting, sedation as well as insomnia. No study reported any severe adverse effects. However, as none of these studies are RCTs, it is difficult to draw any definitive conclusion on the efficacy of beta-blocker medications in the treatment of PB among people with ID.

11.2.7 Psychostimulants

Psychostimulants' effects on people with ID and ADHD are not well established. Among the psychostimulants, methylphenidate (MPH) is most widely used. A recent systematic review [172] of all the RCTs involving people with ID that assessed effectiveness of MPH on the core ADHD symptoms did not find any RCT on adults with ID. Fifteen papers from 13 studies on children and adolescents with ID were included (315 participants were on MPH and the same number on placebo), 12 of which used a cross over design, and one used a parallel design. On average around 40–50% responded to MPH in the ID group, whereas around 70–80% response rate is reported among the non-ID children. It also seems that overall the effect size of MPH's effectiveness in ID children (average around 0.5) is lower than that in the non-ID children (average around 0.8–1.3). Significant adverse effects included sleep difficulties and poor appetite along with weight loss. Other important adverse effects included irritability, social withdrawal and increased motor activities including tic. The type and rate of adverse effects among ID children seem similar to those in the non-ID children (average around 12–24%). Similarly, the placebo effect seen among ID children seems to be similar to that in the non-ID children (average around 12.5%).

Among the non-ID children, studies have shown MPH's efficacy on overt and covert aggression when associated with ADHD [173]. In Aman and colleagues' pooled data from their three previous studies (n = 90), they reported a significant improvement in the MPH group compared with the placebo group

in (a) Teacher's ABC-Irritability subscale score and (b) Parent's Revised Behaviour Problem Checklist (RBPC) conduct problem subscale rating [174]. However, some other small scale studies did not find any significant difference in problem behaviour between ADHD and non-ADHD children with ID. In practice it is not always easy to tease apart symptom of aggression or other similar problem behaviour in people with ADHD particularly in the presence of ID because of symptom overlap among these conditions [17].

11.2.8 Vitamins and Others

The only study available on the effectiveness of diet (zinc supplement) on the management of PB (pica) did not include a proper placebo control group [175]. Therefore, it is difficult to draw any conclusion from this study on the effectiveness of the diet. Apart from zinc, the other vitamins and supplements that showed some effects in various psychiatric disorders but not specifically in ID are folic acid, S-adenosyl-methionine (SAMe), omega-3 fatty acids, L-tryptophan, methylfolate and Vitamin C and D [176, 177]. Several endocrinal agents such as oxytocin, growth hormone and thyroxin have been used in small scale controlled and uncontrolled studies showing equivocal results on cognition and behaviour in people with ID, particularly with Fragile X syndrome and Prader Willi syndrome [169]. Glutamatergic and γ-aminobutyric acidergic (GABAergic) agents have been investigated in people with ID associated with genetic syndromes with disappointing results [169]. Acetylcholinesterase inhibitors such as donepezil and also other cholinergic agents such as L-carnitine/L-acetylcarnitine returned equivocal results in people with Down's syndrome [169]. Clonidine, a centrally acting α2 adrenergic agonist, has been used in the context of ASD and ADHD showing some early moderate effects [169]. Melatonin has been shown to be effective in regulating sleep disturbance, particularly in the context of ASD [178]. Regularising sleep pattern seems to help to improve behaviour in people with ASD/ID [164]. Other melatonergic agents such as agomelatine are at their very early stage of exploration in people with ASD/ID [164].

11.3 Conclusion

On the basis of the evidence available, it is difficult to either recommend or refute the use of psychotropic medications for the management of PB in people with ID. Furthermore, there is no evidence to show the effectiveness of particular psychotropic medication for a particular type of PB. However, there is now moderate quality RCT-based evidence available on the efficacy of risperidone and some preliminary evidence for the efficacy of aripiprazole particularly for children with ASD but also with ID. In the absence of gold standard evidence, guidelines have been developed in order to provide advice to clinicians when using psychotropic medications for the management of PB in individuals with ID [108, 179–183]. These guides advise that a thorough assessment of the causes and effects of the PB including organic/medical, psychiatric, psychological and social factors should be carried out before a medication is prescribed (see also ▶ Chap. 7 on Problem Behaviour) (see ▶ Case Study 8 and 9). See ◘ Table 11.1 and ◘ Fig. 11.2 for main recommendations.

Case Study 8: The Role of Medical Problem in Causing Problem Behaviour

A 65-year-old lady with severe ID suffered from a stroke that led to paralysis of the right side of her body. She started screaming and shouting regularly and the care staff asked for psychotropic medication to manage her behaviour. However, close examination revealed that the lady developed spasticity in her right hand and she was constantly getting frustrated by not being able to stretch fingers of her right hand, which led to screaming. Instead of psychotropic medication, baclofen was prescribed as a muscle relaxant and the lady's behaviour improved.

Case Study 9: A Medical Symptom Leading to Problem Behaviour

A 75-year-old gentleman with moderate ID and limited communication skills developed symptoms of dementia. He started screaming and head banging. His sister who is his primary carer asked for psychotropic medication but the clinician prescribed paracetamol on a regular dose assuming that the behaviour may have been precipitated by headaches. This has produced a good result and the gentleman stopped screaming and head banging.

Non-medication-based intervention should always be considered first (see ▶ Case Study 10). However, medication may sometimes be used to facilitate non-medication-based intervention. Individuals with ID and their caregivers as well as the multidisciplinary team should be fully involved in the decision-making process from the outset (see ▶ Case Study 11) [184]. There are accessible versions of information leaflets (with audio versions) on psychotropic medications freely available for downloading from the web (▶ www.ld-medication.bham.ac.uk; https://spectrom.wixsite.com/project) [185]. These should be handed over to patients and their carers where appropriate.

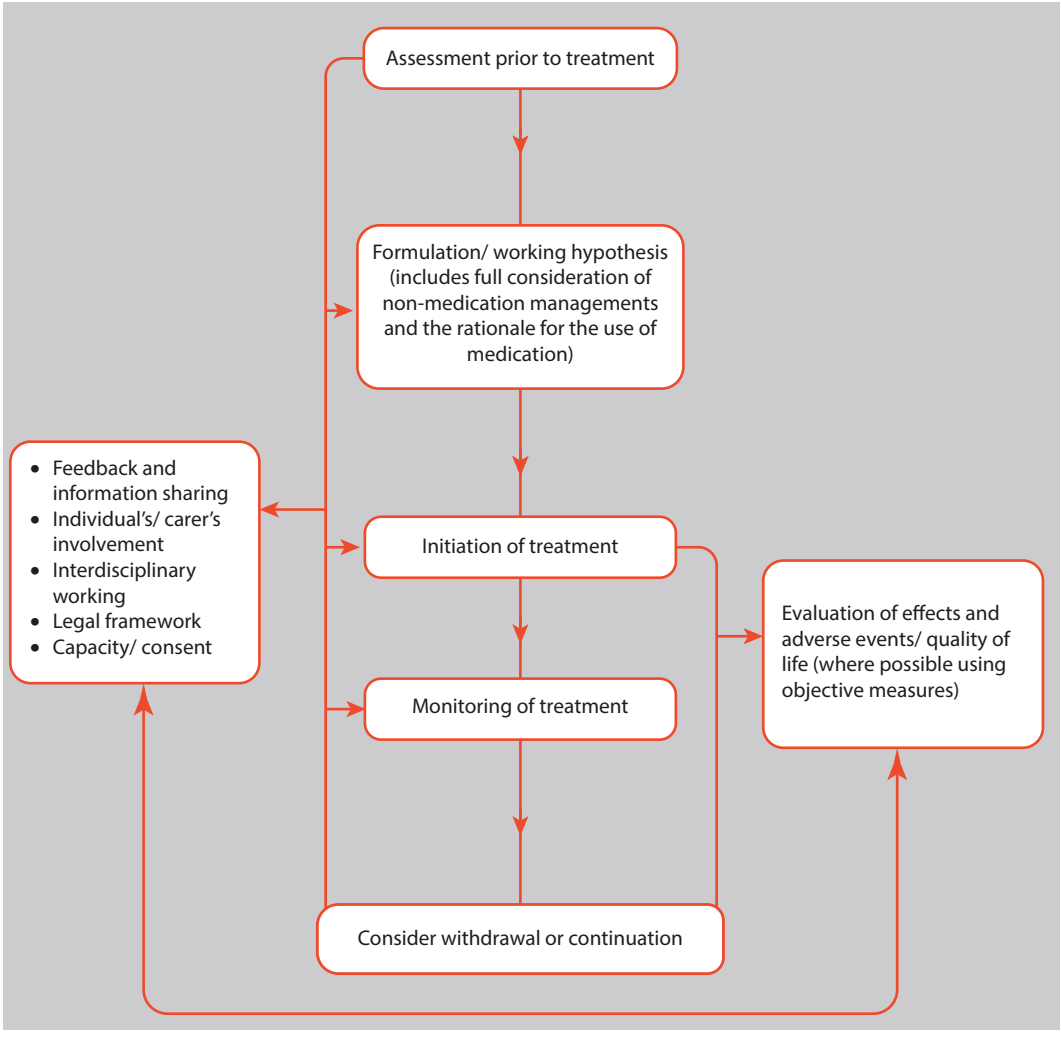

Fig. 11.2 Key processes associated with using medication to manage PB in adults with ID [107]

Case Study 10: The Role of Non-medication Intervention in Problem Behaviour Management

Care staff in a community group home wanted psychotropic medication for a 27-year-old man with severe ID and no speech because of his PB. Further assessment revealed that the person becomes agitated when he is not allowed to go out for a car ride which he enjoys. A Speech and Language Therapy assessment revealed that when the care staff was telling the person, 'you can't go out in a car' and the moment the person heard the word 'car', he thought he was going out and became very frustrated and disturbed as he was not allowed to go out. The Speech Therapist devised a picture board with 'a picture of a car crossed out', which the care staff showed when the person was not allowed to go out and kept him engaged with alternative activities within the house that he enjoys. This strategy was successful, and his behaviour improved without the need of any psychotropic medication.

Case Study 11: The Benefit of Multi-disciplinary Involvement in the Management Programme

Care staff asked for psychotropic medication to control PB in a 36-year-old lady with moderate ID who has no speech. Further assessment by a Speech Therapist revealed that the care staff usually ask the lady what she wants for her dinner and give her two or three choices. She always opts for the last option but when the food is served, she refuses to eat them and becomes disturbed. Speech Therapist suggested that instead of giving her choice verbally, the care staff should show pictures of different meals and will also involve her in the preparation of her meals. This strategy was successful, and her behaviour improved without any psychotropic medication.

Both the impact of the intervention on the behaviour and the adverse events should be assessed as objectively as possible if necessary, using validated instruments (see ► Chap. 7 on Problem behaviour for a list of assessment tools). At each follow-up, the original formulation should be reassessed, and non-medication-based interventions should be considered along with the possibility of withdrawing medication. It is important to recognise the difficulty of monitoring treatment because of difficulty in carrying out necessary investigations (see ► Case Study 12). It is worth keeping in mind that it is possible to successfully withdraw anti-psychotic medications after long-term use in a high proportion of individuals with ID. If medication is withdrawn, a relapse plan should be in place and the possibility of withdrawal symptoms in the form of PB should be considered before taking a decision to reinstate any psychotropic medication. The ultimate aim of the management should be symptom reduction as well as to improve the quality of life of the individual with ID and their caregivers.

Case Study 12: Difficulty in Monitoring Treatment Effect and Carry Out Necessary Investigations

A 33-year-old man with moderate ID and no speech showed PB periodically and the carers raised the issue whether this is part of a bipolar disorder. However, the clinician was not certain as there were no other symptoms to support the diagnosis of bipolar disorder apart from the periodic nature of the behaviour. A small dose of risperidone was used with good effect. The clinician was reluctant to use lithium as another patient in the same clinic developed PB 6 months after withdrawal of lithium after a long-time use. Monitoring of lithium level and other investigations were difficult to carry out in these cases.

Whether medication should be continued will depend on the relationship between efficacy and the undesirable effects, keeping also in mind the potential for contraindications and compliance. Use any medication that was successful in the past and caused no or least harm. Monitor undesirable effects of intervention and interaction with other medication and substances regularly. Discontinue therapy or at least reduce the dose if it does not produce beneficial effects or produces unacceptable adverse effects.

Family history may provide clues to the current problem in the person with ID. The treatment has to be cost-effective. When using more than one medication, evaluate and modify one at a time, assessing the contribution of each individual medication to the observed benefits or adverse effects. Communicate with the person with ID clearly if necessary, using accessible information and other methods such as the use of pictures [185, 186]. It is always prudent to check at the outset patient's and family's expectations from the treatment. For example, some patients may prefer oral over intramuscular therapy or liquid form, perhaps dissolved in their preferred drink. Others may like tablets, perhaps of a certain colour. Assess and document patient's capacity to give an informed consent. If the person cannot consent, then consider involving the legal representative and the family. Any decision taken has to be in the patient's best interests. Re-evaluate the dosage frequently, with the intent of using the lowest possible dose for the shortest possible time, but also for as long as necessary. Adherence to treatment must also be monitored carefully. There are many factors that can influence compliance, such as the expectations of the person with ID and their families and carers or the dynamics involving family members. It is important to communicate with a stable and reliable informant. If no significant beneficial effects have been observed after a period of three to four months after reaching the optimum desirable dose, treatment should be discontinued. Intervention should be an integral part of a person-centred program of care.

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Key Points

- Off-licence psychotropic medications are used commonly to manage problem behaviour in people with intellectual disabilities.
- Evidence to support pharmacological intervention for problem behaviour is equivocal.
- Non-pharmacological interventions should always be considered first for the management of problem behaviour in people with intellectual disabilities.
- The person with intellectual disabilities and their family carers where available should be involved from the outside in the decision of using pharmacological and other interventions along with the multi-disciplinary team.

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Nonpharmacological Interventions

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Learning Objectives

The reader of this chapter will learn which interventions are available for addressing mental health and/or challenging behavior in individuals with intellectual disabilities. S/he will also learn what the evidence-base is of these interventions. Important is that interventions are directed not only toward the client but also toward caregivers. The reader will learn ways of how to adapt interventions to the learning style and characteristics of these individuals.

12.1 Introduction

A range of nonpharmacological interventions have shown promising results in the treatment of behavioral and/or mental health problems in individuals with intellectual disability (ID). This chapter presents a selective overview of interventions that are often applied in clinical practice and that have been documented in the scientific literature. Two main categories of interventions are distinguished: client-oriented and contextual approaches. In client-oriented approaches, the focus of the intervention is the client who is the main recipient of the intervention. We will describe the assumptions, common components, and evidence-base of behavioral interventions, cognitive behavior therapy, and interventions such as mindfulness and relaxation. In contextual approaches, the intervention is primarily directed at persons in the environment of the client such as parents, staff members, and others. The assumption is that a client's well-being will improve and behavioral/mental health problems will be reduced through a change in behavior and attitude of carers and other in the client's environment. Case examples will illustrate the interventions.

12.2 Client-Oriented Approaches

12.2.1 Behavioral Interventions

Behavioral interventions – now referred to as the first wave of behavioral therapies – have been widely used in the treatment of individuals with developmental disabilities. For this population, behavioral interventions have been successfully used to treat a variety of commonly reported excesses and deficits, including (a) aggression, (b) disruption, (c) elopement, (d) feeding disorders, (e) inappropriate sexual behavior, (f) pica, (g) property destruction, (h) rumination and operant vomiting, (i) self-injury, (j) sleeping disorders, (k) stereotyped mannerisms, (l) tantrums, (m) academic failure, and (n) adaptive skill deficits [1–4].

Behavioral intervention has also been successfully used to treat anxiety and mood disorders [2, 4]. Overall, behavioral intervention has a long and continuing history of success in the education, rehabilitation, and treatment of individuals with developmental disabilities. For this purpose, the behavioral approach can be considered a well-established, evidence-based therapeutic model [2, 4].

Behavioral interventions are derived from basic principles of operant conditioning [5]. A considerable amount of research has accumulated since the 1960s showing effective application of behavioral/operant principles for the treatment of individuals with developmental disabilities [2–4].

Key operant principles are listed, defined, and illustrated with examples in Table 12.1.

An important assumption associated with the behavioral treatment model is that human behavior is largely learned and sensitive to environmental contingencies [5]. Another underlying assumption is that the consequences of behavior can have powerful and predictable effects on the topography, frequency, intensity, and patterns of responding [6]. The topography, frequency, intensity, and/or patterns of responding are also assumed to be highly modifiable, provided that the environment, especially the consequences of behavior, can be modified.

In line with these assumptions, contemporary behavioral interventions include an initial assessment phase to identify the operant function(s) of the presenting problem behavior; a procedure that is called functional analysis. This assessment information is then used to design an appropriate course of treatment [7]. Identifying the operant function

Table 12.1 Key operant conditioning principles underlying behavioral interventions

Basic principle	Definition	Example
Positive reinforcement	This term refers to a relation between a response and a consequence in which the future probability of the response increases when it produces or is followed by a specific type of consequence. When a response is followed by a specific consequence and thus becomes more likely to occur in similar conditions, then that consequence can be defined as a positive reinforcer.	Bouts of self-injury by an adult with severe intellectual disability are frequently followed by a caregiver coming over and attending to the person. This attention serves as positive reinforcement and therefore the self-injury is maintained.
Negative reinforcement	This term refers to a relation between a response and a consequence in which the future probability of the response increases when it results in the removal or termination of a specific type of stimulus. When a response results in the removal or termination of a specific stimulus and this becomes more likely to occur in the future, the presence of that stimulus, then the removal or termination of that stimulus can be defined as a negative reinforcer. The stimulus can also be defined as aversive.	An adolescent with autism tantrums and hides under his desk when the teacher tries to engage him in an instructional activity. As a result of this tantrum, the teacher concludes that the student 'needs some alone time.' The teacher therefore removes the task demands and leaves the student alone. The removal of the task demands serves as negative reinforcement for the tantrum behavior.
Punishment	This term refers to a relation between a response and a consequence in which the future probability of the response decreases when it produces or is followed by a specific type of consequence. When a response is followed by a specific consequence and thus becomes less likely to occur in similar conditions, then that consequence can be defined as a punisher.	A child's frequent swearing in the classroom is reinforced by peers laughing and so the teacher decides to punish each instance of swearing by using a 2-min time-out contingent upon each instance of swearing. With this time-out procedure, the frequency of swearing decreases, indicating that the time-out period served as a punishing consequence for swearing.
Extinction	The gradual decrease in the frequency of a response when that response no longer results in reinforcement.	Tantrums that occur at bedtime might be extinguished by making sure that the child is not allowed to stay up later by having a tantrum.
Antecedent control	A stimulus that when present in the environment will either increase (discriminative stimulus) or decrease (inhibiting stimulus) the probability of responding.	A child tantrums when presented with foods that are mixed together on the plate, but does not tantrum when these same foods are arranged in separate piles.
Shaping	The gradual development of new forms of behavior by reinforcing closer and closer approximations of the final desired form.	A child might be taught to request a preferred object, rather than having a tantrum to gain that object by first making any speech-like vocalization (e.g., *ahh*). Over time, the child would be required to produce closer and closer approximations of the correct object label (e.g., *ba*, *ball*).
Chaining	The processes of developing complex skills by linking separate individual responses into an integrated sequence.	A child might be taught to speak in more complete sentences by first teaching and reinforcing single word utterances (e.g., *Ball*), then teaching 2-word utterances (*Want ball.*), and then 3–4 word utterances (*I want the ball.*).

(continued)

Table 12.1 (continued)

Basic principle	Definition	Example
Prompting	Providing an additional cue (e.g., a verbal instruction, gesture, or physical assistance) to increase the probability that a response will occur in the presence of the discriminative stimulus and thus can be reinforced.	During a receptive vocabulary task, if the child does not select the correct picture when instructed (e.g., *Point to the picture of the truck*.), then the teacher could prompt the correct response by pointing to the correct picture or by physically moving the child's finger to the correct picture.
Modeling	A special type of prompt in which the required responses are demonstrated or modeled by another person.	A job coach shows the individual how to operate the photocopier and then gives the person the opportunity to imitate.
Fading	The process of gradually removing prompts so as to promote independent responding.	If a child correctly names pictures only when given a model (*This is a cat, Say cat*), the prompt is faded by giving less and less of the model over successive opportunities (i.e., *Say caa*; *Say ca*; *Say …*).

Table 12.2 Examples of three-term contingencies and operant functions

Antecedent	Response	Maintaining consequence	Operant function or purpose
When an adult is present, but not attending to the child	The child begins to cry and quickly escalates to a full-blown tantrum.	The adult attends to the child in an attempt to calm the child and stop the tantrum.	Social positive reinforcement: Recruit attention
When asked to make his bed	An adult with autism hits the staff person and spits on the bedsheets.	The staff person removes the adult from the room and makes the bed.	Social negative reinforcement: Escape task demands
When greeted by a peer …	The student responds by attempting to bite the peer.	The peer moves away and is less likely to greet the student in the future.	Social negative reinforcement: Avoid social interaction
During playtime …	A child throws objects (e.g., sand, rocks) at peers who play with the child's preferred toys.	Peers are required to give the toy to the child.	Social positive reinforcement: Access preferred objects or activities
On the playground at school …	A child with autism pulls out some weeds and waves these back and forth.	The movement provides visual and vestibular stimulation.	Automatic reinforcement: Gain reinforcing sensory stimulation.

of problem behavior involves an analysis of the environment to isolate the variables that evoke and maintain that behavior. Behavior can be analyzed in terms of the three-term contingency [5, 8]; that is, in terms of the: (a) antecedents that set the occasion for or evoke the behavior, (b) the problematic response forms, and (c) the nature and scheduling of the consequence(s) that are reinforcing problem behavior. Examples of scenarios related to various contingencies are presented in Table 12.2.

The behavioral principles outlined in Tables 12.1 and 12.2 have been applied in numerous flexible ways to the treatment of problem behavior in individuals with developmental disabilities. Three case studies are provided here to illustrate a range of behavioral interventions. These are real cases from our own research or clinical experiences, but some details have been changed to protect confidentiality.

Case Study 1: Treatment of Aggression in an Adult Man with Severe Intellectual Disability by Pairing Staff Presence with Reinforcement and Using a Shaping Procedure

Dean was a 26-year-old man with severe ID living in a community-based group home. He attended a sheltered workshop during the day. When he returned home from the sheltered workshop each weekday, he would typically sit in his favorite chair and attempt to kick or aggressively grab anyone who came near him. Observations over three afternoons suggested that Dean was highly likely to aggress whenever staff approached him, which they often did to request that he engage in some activity, such as asking him to help set the table, come to dinner, or wash his hands. Based on these observations, it was hypothesized that Dean had learned to engage in aggression to escape from these types of staff requests. It was further hypothesized that the approach of a staff person had become a conditioned aversive stimulus that now reliably evoked aggression. To turn this around, the first step was to attempt to establish the approach of staff persons as a signal or cue for positive reinforcement. This was attempted by having staff slowly approach Dean while offering him a preferred snack or beverage. Approximately 10 times each afternoon, while Dean was seated in his favorite chair, a staff person would slowly approach him while holding out a drink or snack that Dean was known to prefer. Staff did not make any demands of Dean at these times. He only had to take the object from the staff person's outstretched hand. With this procedure in place, Dean began to tolerate staff approaching him without attempting to kick or grab them. The next step was to shape up increasingly levels of tolerance by having staff approach more closely to Dean, then approach and actually touch him briefly on the arm or shoulder. Once he was tolerating this, staff then began to approach him and make a simple request (e.g., "Hey Dean, give me a high-five"). When he was tolerating this, staff increased the complexity of the demands, such as by requiring Dean to help set the table. Within a month, aggression had decreased and compliance to staff requests had increased. To maintain these gains, it was agreed that Dean would be allowed to "cool down" for 30 min when he first returned home from the workshop. After this, staff would implement at least three conditioning trials in which they would approach with a preferred snack or beverage and deliver this freely. After this, the general plan was to then approach and present reasonable demands as necessary, which Dean was now highly likely to comply with.

Case Study 2: Treatment of Self-Injury in a Child with Severe Intellectual and Physical Disability by Teaching a Replacement Behavior [9]

Joe was an 11-year-old boy with severe intellectual and physical disability. He could not walk and had no speech and language. He was referred by his classroom teacher for the treatment of self-injury, which had begun when he was about 6 months old. His self-injury consisted of forcibly banging his forehead on the laptray of this wheelchair, hitting his nose with the base of his hand, and – when seated on the floor – banding his head against his knee. His self-injury was frequent and severe. Head banging, for example, was observed to occur at rates

that often exceeded once per second with hundreds of such responses occurring each day. His self-injury had left a permanent welt on his forehead and caused frequent nose bleeds. The first step of this treatment process involved assessing his rate of self-injury under different classroom conditions (e.g., play activity, learning tasks, snack times, independent/alone times). After 90 min of observations conducted over 3 days, a clear pattern had emerged. Joe engaged in almost constant self-injury anytime he was left alone and almost no self-injury when he was engaged in task or interacting with the teacher. Based on this, we hypothesized that Joe has learned to use self-injury as a way of recruiting attention and getting the teacher to engage with him. His intervention program thus focused on teaching him to request attention using a speech-generating device, rather than engaging in self-injury, an intervention approach known as functional communication training [10]. The intervention involved providing Joe with a communication device that required him to press a switch. When he pressed the switch it activated the prerecorded message, "Come here please." Upon production of the message, Joe would receive attention/social interaction. He was taught to press the switch using a discrete-trial training procedure [11]. Specifically, an adult would tell Joe that he had to leave and if Joe wanted to interact he should press the switch. At this point, the adult would walk away and another adult, standing behind Joe, would immediately prompt Joe to press the switch, thus preempting self-injury. Prompting consisted of guiding Joe's hand to press the switch. Over successive trials, the delivery of the prompting was delayed by 3, at which point Joe began to independently press the switch without having to be prompted. As Joe learned to use the switch, he was less likely to show self-injury. In essence, the intervention appears to have replaced self-injury by teaching Joe to engage in a requesting response that seemingly served as same purpose or operant function as his self-injury.

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Case Study 3. Treatment of Depression Disorder and Challenging Behaviors in an Adult with Severe ID

Diana is a 55-year-old nonverbal woman living in an institutional setting since she was 10, with a diagnosis of severe ID and schizophrenia. She was evaluated for treatment because of various challenging behaviors: aggressive (physical and verbal) and destructive behaviors (throwing and destroying objects), refusing to eat, and a range of other dysfunctional behaviors (taking off clothes and wandering around naked, lying on the floor, etc.). The staff members conducted an analog functional analysis, adapting the original conditions introduced by Iwata, Carr, and Durand [12]. They also conducted a preference assessment on single stimuli. Besides, staff members were interviewed on environmental stressors, functional communication skills, support needs, quality of life aspects, and natural/family supports, medical conditions and needs. A psychiatric examination revealed a depressive disorder.

The treatment was aimed at reducing her psychosocial vulnerability and at implementing immediate support by staff on the antecedents of her challenging behaviors (i.e., irritability, low mood, isolation). Once the function (including: avoidance/escape, attention seeking, access to material, and stimulation) of each challenging behavior was assessed, the staff developed a treatment package including the following strategies:

- Modification of antecedents: daily agenda using picture communication, noncontingent reinforcement for not displaying challenging behaviors [13].
- Environmental modifications and protection from threat and physical aggression displayed by others
- Differential reinforcement
- Extinction

More specifically, the treatment was aimed at: increasing her control on environmental variables, behavioral activation (BA), increase of choice-making and self-determination opportunities, demand training (teaching Diana to make more and/or better requests), exposure to success (in order to fade the learned hopelessness), increasing motivational operations, and increasing contact with her family and friends. Diana learned to make more often and more appropriate requests, she accepted to be involved in multiple settings, she showed increased access to available reinforcers and extended her range of preferences, and she agreed to stay with all staff members including male staff members (previously she avoided men). She also began to eat regularly and sleep at night (previously she only slept 2 h per night, spending the rest of the time screaming). A substantial increase in the occurrence of indices of "happiness" was observed, and all the signs of depression decreased in frequency as well as in intensity. Concomitantly, the frequency of her challenging decreased.

12.2.2 Cognitive Behavior Therapy

Cognitive behavior therapy (CBT) – now referred to as the second wave of behavioral therapies – has received increased attention from clinicians and researchers during the past decades (see e.g., Vereenooghe and Langdon [14]). It combines principles from behavioral and cognitive psychology and is referred to as the second wave of behavioral therapies. The assumptions underlying CBT are that behavior, thoughts, and emotions are interconnected, and that cognitive distortions and maladaptive coping strategies increase the risk for psychological and behavioral problems. These problems can be reduced by improving information processing (e.g., through cognitive restructuring – see ► Box 12.1) and the learning of adaptive coping skills.

Contemporary (cognitive) behavioral and other types of interventions should be based on a comprehensive and functional assessment.

Therapists use functional analyses and case formulations to understand the nature and cause of the problems and as a guide to developing treatments (see e.g., Didden [15]). CBT is a composite therapy in which a range of components are used: some of the most common of which are listed in ► Box 12.2.

Box 12.1: Cognitive Restructuring: An Example

- Event: a client who is living in the same community home collides with A.
- Thoughts: He did this on purpose. He does not like me.
- Emotions: Angry, afraid, distressed.
- Alternative thoughts: He did not do this on purpose. He just bumped into me.
- He did not see me. He often comes into my room and starts a friendly chat.
- Outcome: Feels calm, confident.

Box 12.2: Common Components of CBT

- Cognitive restructuring: identifying and challenging negative (automatic) thoughts, also called cognitive distortions. Irrational and/or negative thoughts are replaced by rational and positive thoughts.
- Exposure and response prevention: confront the situation that induces fear or anxiety and decrease avoidance of that situation until anxiety reduces. Response prevention is often used in case of obsessive-compulsive behavior where the client is taught to not carry out his or her impulses to reduce his or her distress.

- Relaxation: any activity or method that leads to a state of calmness if a person feels distressed or tense. Often progressive muscle relaxation techniques are used, as well as breathing exercises and mindfulness.
- Problem-solving and adaptive skill building; the development of conceptual, social, and practical skills that individuals use in daily life. Adaptive skills with which a problem may be solved.
- Functional analysis: assessing the operant function of problem behavior by identifying its antecedents and reinforcers.
- Case formulation: hypothesis or model about the nature and cause of the presenting problems. It provides a framework for treatment.

12.2.2.1 Manualized CBT

CBT is a treatment package containing a large range of techniques that address thoughts, emotions, and behaviors of clients. CBT is a manualized approach in which therapeutic protocols are outlined. At present, there are many protocols available for the treatment of behavioral problems and/or mental health problems in individuals with intellectual or developmental disabilities (IDD). For example, Hassiotis and her colleagues [16] have published a therapist manual for the treatment of anxiety and depression containing case vignettes and examples, exercises, information sheets, resources, and worksheets adapted for use with clients with mild ID. The manual illustrates the process of CBT, from the introduction of CBT and initial assessments to case formulation, treatment planning, intervention, and termination.

Lindsay and his colleagues [17] used a manualized CBT approach for emotional problems (excluding anger). The treatment consists of five modules:

- Module 1: assessment of problems.
- Module 2: preparation phase promoting engagement with CBT. Introducing the concept of CBT, establishing a list with the client's key difficulties that will be addressed, and identifying a family member or carer who will support the client.
- Module 3: recognizing and differentiating between emotions through role-play and scenarios, exploring links between emotions and events that trigger them.
- Module 4: development of a formulation with predisposing factors for the emotional problems, exploring feedback loop from client's behavior to event.
- Module 5: relapse prevention which takes the form of a sort of passport that the client could continue to use following treatment. Key elements of a passport are a simple formulation, adaptive strategies, information about early warning signs, and risk factors. At this stage, the family member or carer was introduced.

12.2.2.2 Adapting CBT to Individuals with ID

Until recently, cognitive and adaptive skill deficits were seen as precluding the use of CBT in individuals with ID. Potential obstacles are limited verbal communication skills, recognizing emotions, executive functioning deficits, and limited working memory, among others. In clinical practice, CBT is often used with individuals with mild ID and CBT is adapted to their skill deficits and needs. Hronis, Roberts, and Kneebone [18] provided suggestions for adapting CBT to children with ID who present with a range of cognitive skill deficits. Below are some examples:

- Attention: use shorter, more frequent sessions, reduce task length (smaller units), and prevent distractions
- Working memory: use memory aids (e.g., visual prompts), present one task at a time, and use short, simple, subject-verb-object sentences

- Executive functions: use structured sessions (e.g., visual schedule), minimize switching between tasks, and redirect uninhibited responses

Cooney, Tunney, and O'Reilly [19] conducted a systematic review of cognitive therapy skills assisting adults with ID to participate in CBT programs. The following skills and prompts were identified that may help adults with ID to benefit from CBT:

- Recognize emotions by using photographs of people's faces.
- Discriminate between thoughts, emotions, and behavior by using personally relevant scenarios and pictorial stimuli.
- Make a connection between events and emotions, and recognize the mediating role of thoughts in the relationship between thoughts, emotions, and behavior by using examples of situations, emotions, and behaviors that are congruent and by using pictorial stimuli.

Willner and his colleagues [20] have adapted the CBT approach to the needs and skill deficits shown by individuals with ID by training their care staff to deliver the CBT. In this multicenter study, a manualized anger management group intervention was compared to a waiting-list control condition. Staff or home carers participated as lay therapists and were trained by a clinical psychologist explaining the principles of anger management and the use of the CBT manual. The outcomes showed that anger management resulted in an improved anger control by participants with ID and an overall reduction in challenging behaviors. Analyses also showed that staff members delivered the CBT intervention with reasonable fidelity.

Dagnan, Jahoda, and Kilbane [21] point to the fact that establishing a therapeutic relationship between client and therapist during CBT is of particular importance, as this has been viewed by the client as one of the most positive aspects of therapy. A positive relationship improves client's motivation to attend the sessions and his or her confidence in learning skills.

Hassiotis and her colleagues [16] have given many suggestions on how to adapt CBT interventions for clients with mild ID.

12.2.2.3 Evidence-Base of CBT

CBT has most often been applied to the treatment of anger and/or aggressive behavior in individuals with ID (see e.g., Willner et al. [20]). Nicoll, Beail, and Saxon [22] have conducted a systematic review of 12 studies assessing the effectiveness of CBT for the treatment of anger problems in adults with ID. Overall, high effect sizes were found for CBT.

However, the authors note that some studies had large confidence intervals, for example, as a result of small sample sizes. CBT has also been applied in individuals with ID who present with mood problems and anxiety. Unwin, Tsimopoulou, Stenfert Kroeze, and Azmi [23] have reviewed 11 studies and concluded that CBT is feasible and may be effective in reducing symptoms of depression in these individuals including negative automatic thoughts. No controlled studies were found for anxiety. Clients and carers perceived CBT in a positive way.

The authors found that most studies had methodological shortcomings such as small sample size and lack of procedural integrity assessments and that data were collected during short-term follow-ups only.

12.2.2.4 CBT Expanding to Other Domains

The range of behaviors or disorders in ID that are being targeted by CBT is extending: substance misuse [24], obsessive-compulsive disorders [25], sexual offending [26], and hoarding. Kellett, Mattuozzo, and Kotecha [27] have assessed the effectiveness of CBT in 14 adults with mild ID who showed hoarding behaviors. They showed excessive acquisition of objects, had a substantially cluttered home environment, and had difficulties with discarding possessions. CBT consisted of 12 individual 2-h sessions via weekly visits to the participant's home. Elements of CBT were goal setting, enhancing motivation, exposure methods, problem-solving, cognitive strategies, and relapse prevention. Carers were used as co-therapists where possible and homework

assignments were given. CBT elements were adapted by reducing the amount and complexity of homework, extending time length of sessions, and simplifying psychoeducation[1] and hoarding formulations. All 14 participants completed the CBT program. Data showed that hoarding was reduced following treatment, a finding that was maintained at follow-up.

12.2.3 Trauma-Focused CBT and Eye Movement Desensitization and Reprocessing

Individuals with ID are at increased risk of experiencing traumatic events, such as sexual and physical abuse, separation, and life-threatening illness, compared to their peers without ID. Processing potentially traumatic events is particularly difficult for individuals with ID. Also, the range of events may be greater for individuals with ID. It is assumed that events, such as early institutionalization and out-of-home placement, few positive experiences in managing negative life events, and limited availability of social support, make individuals with ID vulnerable for developing posttraumatic stress disorder (PTSD) (see Mevissen et al. [29]). Moreover, understanding oneself as having ID may be traumatic in itself. Given the disruptive effects of trauma, PTSD assessment and treatment of PTSD in individuals with ID have gained attention in the literature in recent years. Mevissen et al. [29] have reviewed the literature on two interventions for PTSD in individuals with ID: trauma-focused CBT and eye movement desensitization reprocessing (EMDR), which are evidence-based first-line treatments recommended by the World Health Organization.

Trauma-focused CBT consists of a combination of behavioral and cognitive techniques and focuses on the relationship between events and behavior, cognitions (thoughts, attitudes), and emotions (also see above). Until present, only case studies have been published on the feasibility and outcomes of CBT for the treatment of PTSD symptoms in individuals with ID. A paper by Carrigan and Allez [30] describes the case of Damon who was a 26-year-old who had been diagnosed with mild ID and comorbid autism spectrum disorder (ASD). He lived at home and had no employment. He showed anger outbursts and complained of sleep problems at the time of referral to a community learning disability team. He disclosed that he had been the victim of a sexual assault some years before. He had regularly reexperiences of the event and had nightmares about what happened. He had difficulty concentrating; he had flashbacks and did not want to leave the home because he was afraid he would meet his attackers. In the paper, a case formulation is presented on how his behaviors, cognitions, and emotions are related to Damon's trauma and other symptoms. For example, negative appraisal of the trauma produced a sense of current threat and Damon believed that people wanted to get him and that they are not to be trusted. He blamed himself for what happened. Also, he watched television programs that triggered his memory of the event. His strategies to manage his sense of threat were to avoid people and places where the event had happened and to respond aggressively. He tried to avoid thinking about what happened.

The CBT intervention took 12 one-hour sessions at a hospital. Part of the intervention was anger management for his aggressive outbursts, which was focused on cognitive restructuring of beliefs about the intentions of other people. The intervention had three goals, of

1 Psychoeducation is an intervention for clients, their family members, and/or carers in which information is provided on the symptoms and possible cause of a mental health or behavior problem. Its goal is to understand the problem and provide support, which may help in recovery and well-being. There are very few psychoeducation programs available for individuals with intellectual disabilities and/or autism spectrum disorders. A recently published study by DaWalt, Greenberg, and Mailick [12] showed that an 8-week family group psychoeducation program, called Transitioning Together, resulted in significant improvements in parental depressive symptoms and problem-solving. Social interactions improved for their children.

which the first was to modify negative appraisals of the trauma and its consequences.

Psychoeducation was used to achieve this goal. The second goal was to reduce reexperiencing the event by elaboration of the traumatic memories and discrimination of triggers. For this purpose, imaginal reliving the event was used, whereby Damon was instructed to describe the event in as many details as possible. Thirdly, Damon was helped to drop dysfunctional behaviors and cognitive strategies and to avoid not thinking about the traumatic event. The outcomes showed that trauma-focused CBT – if adapted – is a feasible treatment option for individuals with ID and that the intervention resulted in a reduction of PTSD symptoms.

Eye movement desensitization and reprocessing (EMDR) therapy is an intervention aimed at reducing symptoms that result from traumatic experiences. The theory underlying EMDR is the adaptive information processing theory. Eye movements during the recall of aversive memories reduce their vividness and emotionality. Recalling a traumatic memory is assumed to tax working memory capacity, which is limited. If a second task – for example, eye movements – is executed during recall, less capacity will be available for recalling the distressing event. This makes that the memory is experienced as less vivid and emotional.

Mevissen and her colleagues [29] distinguish eight phases in the planning and execution of EMDR:

- Phase 1: history taking and case formulation, resulting in a treatment plan.
- Phase 2: the client is prepared for the trauma work. Skill building and resource development are typically necessary.
- Phases 3–7: reprocessing of the traumatic memory. The client is asked to bring up the traumatic memory and concentrate on the most distressing image and dysfunctional negative cognition of oneself in relation to that image, as well as the accompanying emotions that go along with it. A core feature of EMDR is the performance of eye movements (typically, the therapist moving his fingers back and forth in front of the client, asking him or her to track the movements while keeping his or her head still), while concentrating on the trauma memory.

  Following the image and negative cognition, access to the emotional and somatic aspects of the memory takes place. Repeatedly, the client is asked to report emotional, cognitive, somatic, and/or imagistic experiences, until intern disturbances reach a SUD (subjective unit of disturbances scale) of zero and adaptive and positive beliefs are rated as strong on a VoC (validity of cognition) scale.
- Phase 8: reevaluation and integration.

Instructions as to how to activate the trauma memory and how to support the client during the desensitization and reprocessing phase are age related and are adjusted to the person's developmental age, taking into account any comorbid disorders such as autism. Task variations might be necessary, for instance, the therapist putting stickers on his fingers to facilitate tracking, using buzzers to vibrate alternately between the person's right and left hand, administering alternating tones via a headphone or audio speakers placed on either side of the person, or tapping on the person's hands or knees.

The evidence-base for EMDR in individuals with ID is still small. In the literature, mostly case descriptions have been published (see Mevissen et al. [29]). Mevissen, Didden, Korzilius, and De Jongh [31] assessed the effectiveness of EMDR in two individuals with mild ID who met the diagnostic criteria of PTSD. Data were collected in a multiple baseline design and EMDR comprised of four 60-min sessions. The results show that for both participants, the number of PTSD symptoms, as measured by the ADIS-C PTSD section (see Mevissen et al. [32]), decreased in response to the treatment and both no longer met the PTSD criteria following treatment.

12.2.4 Relaxation

Relaxation is often a component in a treatment package. There are few studies evaluating relaxation as a stand-alone approach in individuals with ID and/or ASD. Bouvet

and Coulet [33] assessed the effectiveness of relaxation on anxiety, self-esteem, and emotion regulation in 30 adults with mild-to-moderate ID who worked in a French center of supported employment. In 1-h groupwise sessions, clients practiced with relaxation techniques during 10 weeks. They found that, contrary to a waiting-list control condition, relaxation significantly reduced state anxiety and improved the clients' self-esteem and cognitive reappraisal. A recent systematic review by Bellemans and her colleagues [34] showed that relaxation – particularly through progressive muscle relaxation – on average was effective in reducing aggressive behavior in individuals with ID. It should be noted however that the results were somehow mixed. There are relatively many techniques of relaxation. Harris and Robbins [35] used three basic relaxation techniques in the treatment of three clients who suffered from anxiety. For example, relaxation was applied to a female with moderate ID who showed symptoms of anxiety related to the behavior of other clients in her house. The intervention started with controlled breathing (she was able to follow basic instructions regarding her breathing exercises). Unfortunately, because of her communication difficulties the second technique – guided imagery – turned out to be problematic. During guided imagery, the client is guided toward a relaxed and focused state with the help of verbal instructions from a therapist or script. A state of relaxation is reached, if the client succeeds in imaging the details of an event or a safe place, which eventually results in a decrease of tension or anxiety. In her case, the technique was applied in an adapted way, in that a script was developed in which a trip to the zoo was detailed with vivid descriptions of each animal enclosure. This imagery was chosen because she loved animals. It appeared that she responded well to imagery plus controlled breathing. In the third technique – progressive muscle relaxation – she was prompted to use stress balls in each hand to grip and promote tension of the arms. This technique was also adapted in that she was prompted to "squeezing the juice from fruit." The combination of techniques helped her to get some degree of control over her anxiety, particularly regarding her fear of other clients in her house and some of her outreach activities. The case descriptions by Hart and Robbins show that relaxation techniques can be applied to individuals with ID, provided they are adapted to the individual.

12.2.5 Psychodynamic Psychotherapy

Psychodynamic psychotherapy is based on psychoanalysis. At the center of this therapy lies the concept of defense mechanisms which may be defined as the mental operations that remove thoughts and feelings from conscious awareness. Individuals keep something out of thoughts or alter the perception of events as a strategy to manage emotional conflicts or internal and external stressors. The therapy may support individuals to identify what they are doing and to help them to come to terms with why they are doing it. In this way, individuals move beyond the level of "problem statement" (e.g., aggressive behavior). During psychodynamic therapy, a strong focus is placed on the relationship, interpersonal relations, expression of emotions, and exploration of fantasy life (see Beail and Jackson [36]). McInnis [37] reviewed the evidence-base for psychodynamic therapy in adults with ID. On the basis of 14 papers, she concluded that this type of therapy may be of benefit to these individuals. However, the review revealed a range of limitations such as the small number of studies published and the reliance on case studies.

12.2.6 Mindfulness and ACT as Examples of Third Wave of Behavioral Therapies

Behavioral treatments have developed significantly in the last two decades. After the first-generation behavioral therapies – that is, behavior modification – and the spreading of cognitive behavioral approaches (see previous section), recently a "third wave" of behavioral models have been developed. They are mainly focused on the *function* of problematic

cognitions rather than their *content* [38], and put a strong focus on private events without trying to change the form or frequency of these private events (thoughts and emotions). The third wave of behavioral psychotherapies is an important arena of modern psychotherapy development, which has added extensively to the empirically supported treatments for mental suffering [39]. These approaches include a diversity of new techniques and open up possibilities for the treatment of mental disorders. The third wave usually consists of acceptance and mindfulness procedures in combination with behavior modification principles. It is acknowledged that people with ID are more susceptible than others to challenging behavior and mental health problems, but in order to apply a psychological treatment effectively with a person with ID, a conscious effort is needed to adapt the content and the style of presentation to take account of the client's limitations. Table 12.3 [39, 40] summarizes the deficits and potential solutions in four domains of cognition: intellect, emotional literacy, memory, and executive functioning. These issues must be considered during the assessment phase to drive the adaptation of the treatment process.

The main categories of the third wave of behavioral therapies are summarized in Table 12.4.

There is a specific interest in the outcomes of mindfulness-based and ACT interventions for individuals with ID who present with challenging behaviors (mainly aggressive, self-injurious, and stereotypic behavior) and mental health problems because of the negative consequences associated with such problems including the increased risk of exposure to stressful or even dangerous events for staff [39, 49]. ACT can help people with mild-to-severe ID [50], enabling the client to participate in the therapeutic process more readily than in a more traditional CBT approach. In individuals with high-functioning ASD, for example ACT may result in reduced levels of stress, hyperactivity, and emotional distress, and in improved prosocial behavior [51]. The option of treatment using ACT for individuals with ID and/or ASD appears stimulating and convincing, for which a wide range of potential applications are available [52, 53].

Table 12.3 Cognitive deficits and their implications for therapy

Cognitive domains	Specific processes	Implications for therapy
Intellect	Verbal understanding and reasoning	Simple words and short sentences
	Nonverbal understanding and reasoning	Use of nonverbal techniques and materials
Emotional literacy	Emotional vocabulary	Psychoeducation
	CBT skills	Psychoeducation and provision of ideas
Memory	Assimilation	Frequent repetition and more sessions
	Recall of experiences	Involvement of carers
	Prospective memory	Use of reminders and involvement of carers
Executive functioning	Working memory	Chunking of information
	Behavioral inhibition	Greater use of behavioral self-control techniques
	Initiative	Provision of ideas

Research on mindfulness-based interventions and ACT for carers reports positive findings for both staff and the individuals with ID they support (see e.g., Harper, Webb, and Rayner [54], McConachie et al. [55], Noone and Hastings [56], Leoni et al. [49], Ó Donnchadha [57]). Mindfulness-based interventions can also facilitate staff to integrate behavioral and psychopharmacological treatment with more positive outcomes [58], reduce the use of physical restraints [59], and increase indices of happiness in individuals with profound ID [60].

Table 12.4 Categories of third wave of behavioral therapies

Behavioral activation (BA)	BA emerged from studies analyzing the necessary components of classical cognitive therapy [41], and has evolved from a long behavioral tradition seeking to increase positive reinforcement by scheduling appropriate patient behaviors and thus achieving antidepressant action [42].
Dialectical behavioral therapy (DBT)	DBT conceptualizes that skill deficits in the area of emotion regulation are at the center of these disorders [43]. DBT includes the practicing of different skills in the areas of mindfulness, distress tolerance, emotion regulation, and interpersonal effectiveness [[44], 45].
Metacognitive therapy (MCT)	Metacognitive therapy (MCT; [46]) postulates that at the core of depressive and anxiety disorders there is the 'cognitive attentional syndrome' (e.g., worrying, rumination, dysfunctional threat monitoring, and dysfunctional cognitive and behavioral copying). MCT uses attention training techniques to develop skills in cognitive flexibility, teaches detached mindfulness, and guides cognitive and behavioral experiments to change metacognition.
Schema therapy (ST)	ST uses emotion activation techniques originating in Gestalt and Psychodrama; but it is strictly behavioral in the models communicated to the patient [43]. A basic ability reinforced in ST is to recognize the present dysfunctional modes of functioning, such as the detached protector mode, and to have behavior guided by the healthy mode [47].
Mindfulness-based cognitive therapy (MBCT)	The focus of MBCT is to learn to experience dysfunctional thoughts as internal events separated from the self [48]. Like cognitive-behavioral therapy itself, mindfulness-based approaches encompass a wide range of techniques that are customized to the needs of the individual. MBCT uses psychoeducation and encourages patients to practice mindfulness meditation. A core goal is to develop metacognitive awareness, which is the ability to experience cognitions and emotions as mental events that pass through the mind and may or may not be related to external reality [40].
Acceptance and commitment treatment (ACT)	ACT is a behavioral therapy system that is based on functional contextualism and the relational frame theory (RFT). It postulates the following psychopathological processes as central to human suffering and mental disorders: (1) cognitive fusion; (2) experiential avoidance; (3) attachment to a verbally conceptualized self and a verbally conceptualized past; (4) lack of values or confusion of goals with values; and (5) absence of committed behavior that moves in the direction of chosen values. The treatment contains experiential modeling, psychoeducation about key mechanisms, exercises in mindfulness, and cognitive defusion. The value orientation of the patient is elicited and shared, and the patients are reinforced in value-driven behavior in contrast to behavior driven by emotional or experiential avoidance. There are an impressive number of randomized controlled trials (RCTs) testing the efficacy of ACT in heterogeneous clinical conditions. (For a more detailed description of ACT process in individuals with ID, see Leoni et al. [49].)

Case Study 4: Acceptance and Commitment Treatment (ACT) for an Adult with Mild ID

Pietro is a 45-year-old man with mild ID living in a residential setting. Prior to his admission to the residential setting, he lived in the community in a private house, with daily supports. He spent most of his time wandering in the streets, trying to talk with people or to collect money. He was constantly worried about being mislabeled and teased, to the point that he displayed severe challenging behaviors. He was charged for aggressive behavior multiple times and often refused his prescribed psychotropic medications. The community services

decided to refer him to a residential facility, with a diagnosis of psychosis and moderate ID. At admission, he showed severe limitations in communication, inclusive activity, daily routine (self-care, home care), and job activities. He displayed a high frequency of dysfunctional behavior antecedent to problem behavior: he engaged staff members in continuous talk, thereby repeating topics, questions, and phrases, and he was constantly worried about the behaviors displayed by other persons; he felt threatened and was intimidating toward others. Staff initially used a range of behavioral interventions: antecedent modifications (environment management, supports to communication, and structuring of daily activities), and contingency management (token economy), without satisfying results. At some point, the staff introduced a treatment based on acceptance and commitment treatment (ACT), using the Matrix, a tool in which the Hexagon Model used in ACT is simplified; [49, 61] (see Fig. 12.1). Individual sessions of 45 min were held twice a week in order to work with Pietro on the main levels of the Matrix.

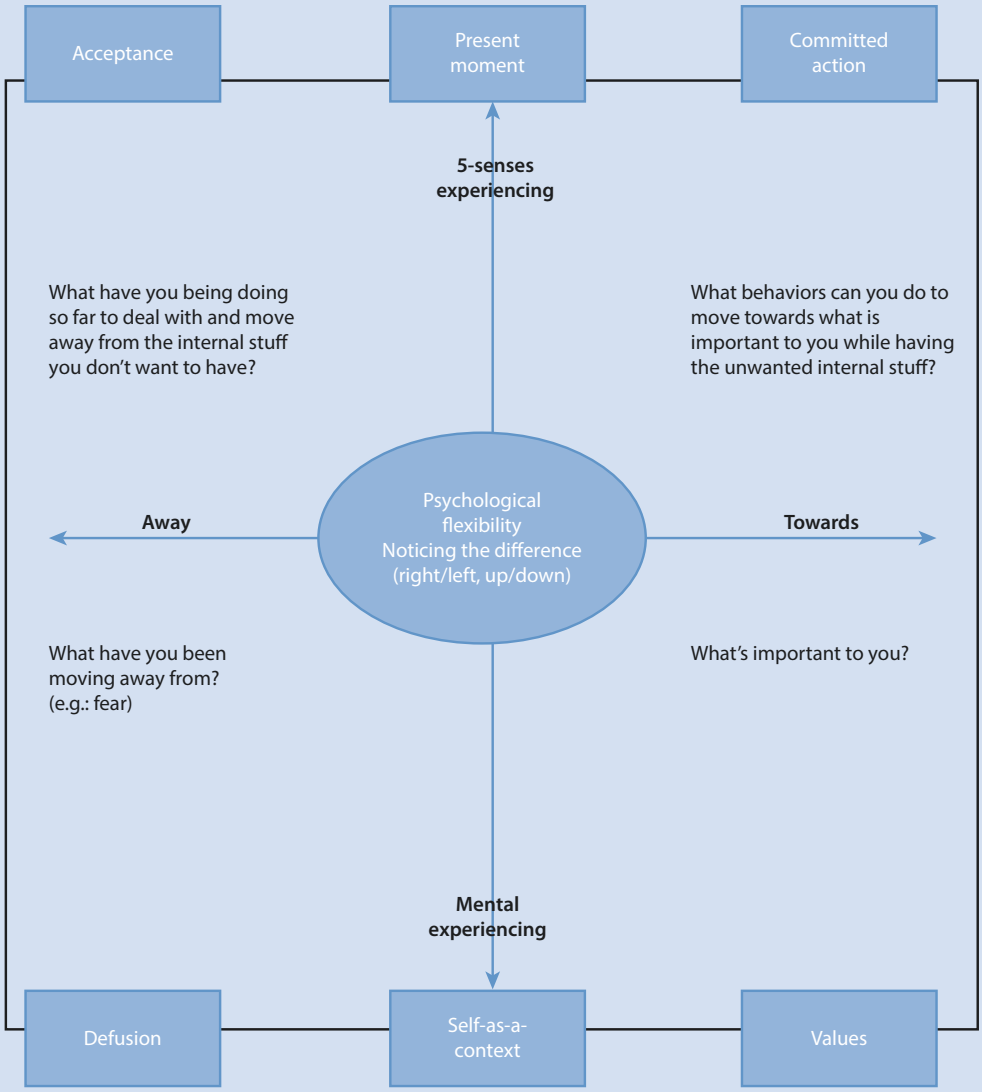

Fig. 12.1 ACT "Hexaflex" model simplified through the Matrix [49]. (Source: Polk and Schoendorff [61])

Firstly, the therapist increased Pietro's awareness of his body sensations and thoughts using instructions and physical exercises. The goal was to identify avoidance patterns in Pietro's behavior. Next to this, a preference assessment was done as well as a life-values analysis (i.e., important directions in life, to be pursued through goals and actions), and connections between values and daily choices were made. Additionally, observation and self-monitoring sheets that Pietro could use were introduced, and behaviors displayed in the previous days were evaluated using the Matrix (see Fig. 12.1), addressing the "away moves" (i.e., avoidance, usual habits, and dysfunctional problem-solving; left part of the Matrix) and the "moving forward" acts (i.e., behaviors aligned with preferences and values; right part of the Matrix). Pietro complied with the treatment (although many adaptations were required, due to his cognitive limitations), and after 4 weeks of treatment, he showed first signs of improvement.

Treatment lasted (with progressive fading of supports) for 24 months, and the staff observed a clear increase in adaptive choices, in reported and observed awareness, in the quality and quantity of the request for supports, and in the frequency and quality of activities. He was able to deal with the grief after his mother passed away, and to move in another group home and negotiate new objectives in the support plan. Challenging behaviors were significantly reduced in terms of frequency, duration, and intensity.

12.3 Contextual Approaches

Contextual approaches are very common in the treatment of mental health problems or challenging behaviors in individuals with intellectual or developmental disabilities (IDD). By contextual, we refer here to approaches that target changes in the *environment* of individuals with IDD. The environment consists of the structural environment (e.g., residential setting and its operational model) and, crucially, the social environment, that is, the people who live and interact with the individual with IDD (paid carers and family carers). The rationale underlying contextual interventions is that challenging behaviors or mental health symptoms of individuals with IDD will improve, if the people who live with them change some aspect of their own behavior (overt or covert behavior) that relates directly or indirectly to challenging behaviors or mental health problems experienced by people with IDD.

Contextual approaches *directly* targeting reductions in challenging behaviors focus on altering overt behavior of carers (i.e., parent training, staff training) so as to alter the contingencies that maintain challenging behavior. This may be combined with changes in the broader system aiming to improve quality of life or activity participation (whole environment interventions such as Active Support or Positive Behavior Support). Contextual approaches that target *indirectly* mental health problems or challenging behavior aim to alter the covert behavior or, else, emotional well-being of carers (family or paid carers). The latter approaches consist of psychological therapy for carers or psychological therapy combined with behavioral skills training.

Figure 12.2 maps some of the contextual factors related to the mental health or challenging behaviors of individuals with IDD using Bronfenbrenner's ecological systems framework. The intervention approaches described below focus on the micro- and mesosystems, though the latter ones actually span across both micro- and mesosystems.

> Challenging behaviors or mental health symptoms will improve, if the people who live with them change some aspect of their own behavior.

12.3.1 Parent Training

As a contextual intervention, parent training is a behaviorally based approach that

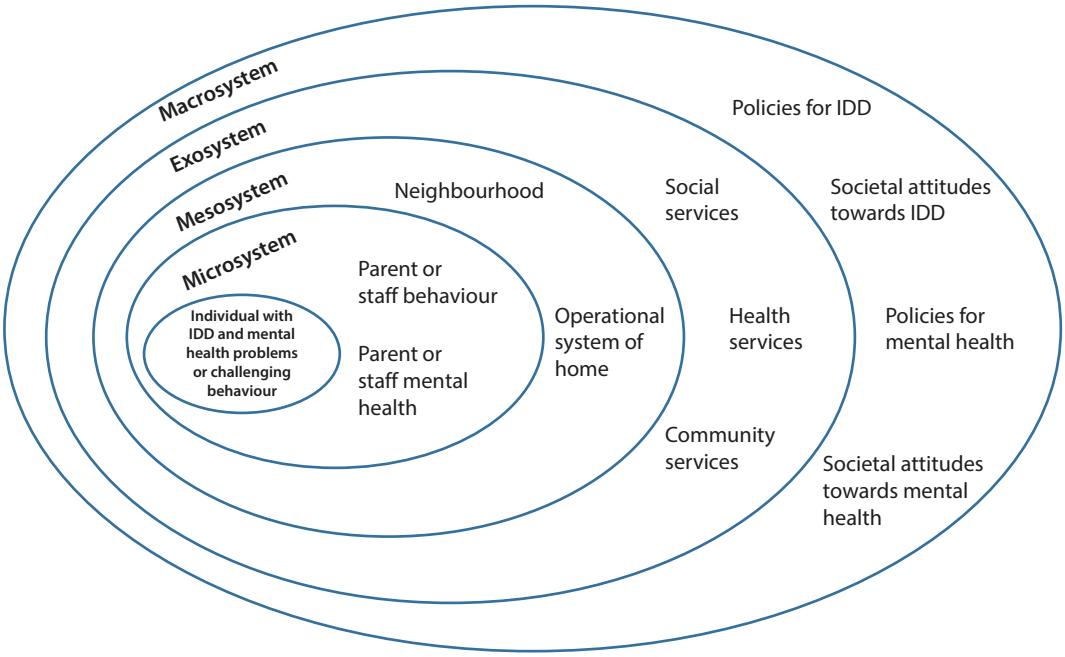

Fig. 12.2 A schematic representation of contextual influences on individuals with IDD and mental health problems or challenging behaviors

targets changes in the parenting behavior of parents of children with IDD, typically in response to emerging or established behavior problems. Most parent training models incorporate approaches from social learning theory and applied behavior analysis to reduce child behavior problems by increasing the use of positive parenting practices (e.g., praise for appropriate child behavior, use of positive behavior management approaches such as differential reinforcement; also see Table 12.1). There are several packaged models, and some may also include other theoretical principles, but social learning and behavioral principles underlie most approaches. Several randomized controlled trials (RCTs) and, subsequently, meta-analyses of RCTs indicated that parent training is effective in reducing child behavior problems (and in particular, conduct problems), increasing positive parenting (e.g., praise), decreasing negative parenting (e.g., ineffective discipline) while also improving maternal mental health, and making parent training an evidence-based (and cost-effective) approach for reducing behavior problems in childhood [62, 63]. There are various parent training models, but there is no evidence of differential effectiveness for different evidence-based models [64]. Notably, some programs, such as Triple P [65] and Incredible Years Parent Training (IYPT; [66]), have a very large evidence-base across several countries.

These parent training models address the general population. They have not been developed for families of children with IDD. However, parent training is gaining momentum as a contextual approach to reducing behavior problems in children with IDD. For parents whose children have IDD, parent training might involve one of the three options: (a) attending a parenting program that has been developed for the general population, for example Triple P or Incredible Years, (b) attending a parenting program that has been *adapted* for use with families with a child with IDD (e.g., Stepping Stones Triple P, [67]; Incredible Years Parent Training for Developmental Disabilities; IYPT-DD, [68]), or (c) a parenting program that has been developed specifically for these families (e.g., Signposts, [69]; Confident Parenting, [70]).

Table 12.5 Some key features of parent training targeting children's challenging behaviors

Content[a]	Format	Additional IDD focus
Positive parenting: when/how to use positive reinforcement; engaging in positive interactions and activities with child Child behaviors to attend to and child behaviors to ignore Behavior management: when/how to use extinction, how to use punishment (time out) Responding to the child in a consistent manner	Small group with/out 1–1 sessions Weekly meetings Facilitator led with/out parent co-facilitator Parents expected to practice new skills with own child and then report back to the group	Behavioral phenotypes of various syndromes Understanding the function of (challenging) behavior; focus on working out functions of own child's challenging behaviors and plan for change Communicative dimension of challenging behavior; adapting communication Changes in the environment to prevent challenging behaviors Supporting alternative behaviors (e.g., teaching a child to communicate a need as replacement for challenging behavior)

[a]See also Kaminski et al. [72] for a component analysis of effective parent training

At the moment, we have no evidence of the relative effectiveness of these different options, as approaches have not been directly compared.

Most parent training models involve parent training in small groups (8–12 parents), attending weekly groups ranging from 4 to 20+ weeks. Therapists are trained and/or accredited in the model delivered, and in some programs they are accompanied by parent expert trainers who have undergone similar training. The latter approach is considered to have high levels of acceptability from parents. Groups typically involve direct teaching, group discussion, modeling, experiential learning, homework, and reflection. Parent training in this population is typically available as a targeted or specialist intervention (i.e., after challenging behavior develops or worsens), whereas preventative approaches are not typically available, despite the fact that challenging behaviors are present from a very young age in children with IDD [71]. Table 12.5 summarizes some of the key features of parent training approaches.

Attending a parent training group that is not adapted or developed specifically for parents of children with IDD might not always be the preferred option for parents of children with IDD but in some areas it might be the only program to offer within a reasonable distance. A degree of inference is applied there, as we assume that families of children with IDD will benefit from programs that do not take into account the child's disability [73]. A recent evaluation with mixed disabilities (families of children with special educational needs) indicated that these families benefit from parent training similarly to families with children without special educational needs [74]. Parents in this evaluation received several programs, with about 80% attending Triple P groups. Clinicians need to be aware that where parents receive generic parent training as part of regular service provision (where services lead on all aspects of delivery including outcome evaluation), longer-term gains (at 12 months past training) are likely not maintained well, and these parents will benefit from top-up or repeat parent training.

Some parenting programs developed for the general population are adapted for families whose child has an IDD. Two such programs are worth noting: Stepping Stones Triple P (SSTP; [67]) and IYPT-DD [68]. Adaptations typically include additional content to provide IDD-specific information (for example on the nature and causes of IDD, behavioral phenotypes in various IDD), a focus on parental

adjustment (what does it mean to have a child with IDD), as well as a focus on the function of behavior problems. Stepping Stones Triple P has been based on Triple P, while IYPT-DD is an adaptation of Webster-Stratton's parent training variant of Incredible Years.

In their adapted versions, both these programs are available in a tiered format moving from less intensive to more intensive (e.g., weekly small parent groups with additional 1–1 meetings) as child need increases. At the moment, most of the evidence available to support the effectiveness of adapted parent training programs refers to small group format that meets on a weekly basis: level 4 for SSTP includes 6 group meetings and 3 1–1 consultations spanning 9 weeks in total, and 12 weekly meetings for IYPT-DD [68, 75]. In the case of IYPT-DD, a small-scale RCT with parents of young children (2- to 5-year-olds) with developmental delays suggested significant gains for observed and parent-rated behavior problems [76]. Incredible Years has also been adapted for families of children with autism and is currently being evaluated in a pilot RCT [77]. The autism adaptation places emphasis on parents helping their children with their communication needs as a functional approach to reducing behavior problems. The evidence-base for SSTP is much larger: a recent meta-analysis of 12 RCTs concluded that reductions in behavior problems of children with IDD are significant and approximately medium-sized, that is, an effect size of about half a standard deviation [75]. Of note, most of the evidence came from level 4 SSTP evaluations (i.e., small group format with 1–1 support meeting on a weekly basis for 9 weeks), whereas gains from level 2 SSTP (one or two large group seminars) were not present for child behavior problems or any other outcomes. Overall, IDD-adapted parent training is the parent training option with the largest evidence supporting its effectiveness in reducing behavior problems, though clinicians should be mindful that reductions in children's behavior problems are evident following more intense input; there might be little value in less-intense approaches.

Last, attending parent training, specifically developed for parents of children with IDD, is a very appealing option for parents who feel that being surrounded by people who experience the same challenges as them is a very important feature of parenting groups [78]. Parents value opportunities to share while not feeling judged or criticized by the other parents, and they also feel that they learn more by other parents in their group. Examples of such programs are Signposts in Australia [69], Confident Parenting in the UK [70], and (a soon to be evaluated by an RCT) Early Positive Approaches to Support in the UK (E-PAtS; Coulman et al. [79]). Some programs specifically target child challenging behavior (Signposts, Confident Parenting), but mostly take a broader view of family needs, by also addressing parental psychological well-being and other child needs, such as sleep or adaptive skills (Confident Parenting, E-PAtS). To date, we have no rigorous evidence that IDD-specific parent training improves challenging behavior or child mental health problems.

Therefore, while the option of attending an IDD-specific parent training group might be valued by parents for the social support networks, it is uncertain whether benefits in challenging behaviors or child adaptive skills will follow.

12.3.2 Parent Therapy

Parents of individuals with IDD experience significantly worse mental health than other parents, and this increases the risk for behavior and mental health problems in their offspring with IDD [80, 81]. Therapy to improve mental health in parents has focused on decreasing negative affect (e.g., CBT) or increasing mental well-being (e.g., mindfulness, positive psychology approaches). Overall, parent therapy is effective in reducing psychological distress in parents, especially mothers, of children with IDD, and the most effective approaches are the ones that combine behavioral parent training with therapy [82].

However, there is little direct evidence that parent therapy is associated with improvements in offspring mental health, as parents

tend to receive therapy because they experience poor mental health themselves (therefore, services or researchers tend not to measure offspring mental health). A contextual approach with some evidence of reduction of child aggressive behavior is mindfulness. Parents who receive mindfulness therapy tend to report lower aggression, more compliant/easier behavior for their children with IDD [83, 84], despite the fact that mindfulness therapy does not directly target parenting skills or child behavior at all. It is thought that the practice of mindfulness increases acceptance of the child with IDD and its behavior, and therefore increases positive interactions between the parent and the child. It is likely that parents who practice mindfulness become more aware of the ways they respond to their child's aggression and, as such, better able to interrupt unhelpful responses. It should be noted that other mindfulness evaluations have replicated the gains in parental mental health but did not find gains in child challenging behaviors [52].

12.3.3 Staff Behavioral Training

As with parents, staff behavioral training aims to improve staff skills in managing challenging behaviors using a variety of principles derived from applied behavior analysis. Staff behavioral training is an evidence-based approach for reducing challenging behavior in adults and children with IDD [4, 85]. See ▶ Sect. 12.3 outlining how behavioral training can support reductions in aggression and self-injury as well as anxiety. Overall, systematic evidence on the effectiveness of stand-alone carer behavioral training on the mental health of people with IDD is scarce (compared to effects on challenging behavior), and clinicians tend to implement direct interventions for mental health problems in this population (see ▶ Sect. 12.2). In the case of severe IDD, there is no evidence on how staff stand-alone behavioral training might impact on mental health problems with the exception of single case evaluations of enriched environment for low mood, and relaxation with interruption for treatment of tics [86]. More evidence is available for sleep problems. A recent meta-analysis demonstrated that carer behavioral training is a promising evidence-based approach for sleep problems, which can be symptoms of broader mental health difficulties [87]. Training caregivers on extinction, and positive reinforcement of appropriate behavior (e.g., going to bed), while also implementing environmental modifications that promote sleep (e.g., no naps during the day, exercise, no television in bedroom) were associated with very large increases in both sleep initiation and sleep maintenance behaviors [87].

12.3.4 Staff Psychological Therapy

Staff psychological therapy is a contextual approach that primarily aims to improve staff mental health. Staff who support adults with IDD and challenging behaviors, and in particular aggression, experience higher levels of burnout and emotional exhaustion [88]. Job-related stress is one of the main reasons staff in IDD services leave their jobs [89]. At the same time, it is considered that burnout and stress lead to a worsening of staff attitudes regarding challenging behaviors, which in turn has been associated with more restrictive staff practices [90]. Two staff therapy approaches that have attracted attention in the past few years are acceptance and commitment therapy (ACT) and mindfulness-based approaches. At the core of these approaches is that the increase in psychological acceptance that comes through ACT or the practice of mindfulness will lead to more positive and less avoidant interactions with adults with IDD and challenging behaviors [91]. Most of the (still very limited) evidence to date suggests that ACT and mindfulness-based approaches reduce psychological distress in staff working in IDD services [55, 56, 92]. However, despite the hypothesized relationship with challenging behaviors, there is very little evidence that staff ACT or mindfulness-based therapy impacts on challenging behaviors of adults with IDD, mostly because studies have not collected such data. There is some evidence that mindfulness-based therapy for staff leads

to a reduction in the use of restraint or similar restrictive practices from staff, especially when combined with systemic approaches (see below).

12.3.5 Systemic Approaches

Systemic approaches target at once several aspects of the environment of the person with IDD. Systemic approaches are warranted when there are concerns about the overall quality of life of people with IDD, not just their levels of challenging behavior or mental health problems.

Active Support aims to improve the quality of life of adults with IDD by increasing participation in meaningful daily life activities, with appropriate support from staff [93, 94]. Active Support was developed to provide an operational model for community homes for adults with IDD. As a philosophy of care, it promotes active participation, choice, and independence. As a systemic intervention, it focuses on each resident at a time, while changing the way the whole home operates. Active Support draws on behavior analytic principles and techniques, teaching staff behavioral skills to support resident engagement in activities, and also using behavioral technologies for organizing life in the home in such a way that opportunities for activity participation are always available along with staff support, that resident experience is monitored through ongoing data collection, and that increased independence is achieved by practicing and learning new skills [95, 96]. In community settings, Active Support significantly increases the amount of time residents spend engaged in daily life activities, and the amount of time staff support them to do so [97].

Active Support does not directly target challenging behaviors, instead it promotes engagement in activities *despite* challenging behaviors, that is, challenging behaviors should not be put forward as a reason not to support a person to participate in daily life activities, such as shopping, cooking, and looking after their own home. Active Support is considered to impact indirectly on challenging behaviors by changing the establishing or abolishing operations that facilitate them (i.e., by changing the motivation to engage in challenging behaviors): Active Support creates a "helpful environment," one where staff attention is available and contingent on appropriate resident behavior, where access to activities and food/drink is always available, where residents know their schedule, and where staff support matches resident skill, so that the "need" to escape difficult instructions or activities is minimized [94]. In other words, Active Support is expected to impact on challenging behaviors by indirectly impacting on their function(s). Active Support does not target mental health problems directly. However, increasing levels of activity should reduce depressive symptoms in adults with IDD [98].

A recent meta-analysis indicated that there was actually very little change in resident challenging behaviors following Active Support implementation, and unclear change in depressive symptomatology [97]. Overall, however, the quality of included studies was moderate to low, and until better quality studies are available it will be difficult to determine whether the hypothesized effects on challenging behavior and depression can be brought about by a systemic intervention that only indirectly targets these outcomes. Clinical case work has shown that when Active Support is combined with Positive Behavior Support (PBS), significant reductions can be achieved [99].

This raises the possibility that systemic approaches work best to improve mental health and challenging behaviors when combined with direct interventions for reducing challenging behavior. Positive Behavior Support (PBS) is a multicomponent intervention that targets change in the environment, in which challenging behavior is displayed as well as change in the individual who presents with these behaviors. Contextual approaches within PBS can take many forms (from behavioral staff training to ensuring that daycare placements are available), and this depends on (a) preferences of the person with IDD and (b) the results of a systematic, in-depth analysis of the function of challenging behaviors (i.e., functional analysis; [100]). As a systemic intervention in community settings

for adults with IDD who already present challenging behavior, PBS can reduce challenging behaviors and lower the probability of mental health disorders (namely affective, psychotic, and organic disorders; [101]). Contextual interventions within PBS are variable, and not clearly documented, whereas PBS approaches directly targeting challenging behavior reductions are more standardized, that is, functional analysis and function-based interventions. In the absence of a component analysis, it is unclear which PBS components (the contextual approaches vs the direct ones) are associated with improvements. Systemic approaches also work well when coupled with other contextual interventions, such as staff therapy. Offering staff mindfulness training (with a focus on personal meditation) alongside training in PBS seems to produce higher reductions in aggression (and staff use of restrictive practices) than just training staff in PBS [102]. It is not clear why this happens, but if PBS focuses on the reduction of challenging behavior through the implementation of positive interventions, it may improve staff skill in managing resident behavior, but does not necessarily improve staff skill in managing their own behavior consistently; the addition of mindfulness strengthens staff awareness of own thoughts and emotions leading to increased psychological flexibility, which in turn may enable a more adaptive behavioral response to resident aggression [103].

Overall, systemic approaches are not the first port of call for reducing mental health problems or challenging behaviors that are already present. Systemic approaches aim primarily to improve quality of life, but where challenging behaviors are present, a whole environment approach that combines direct work to reduce challenging behavior, environmental modifications to improve quality of life and reduce opportunities or motivation for challenging behavior, as well as support for the mental well-being of the social environment appear to be particularly beneficial for challenging behaviors and mental health problems of people with IDD.

12.4 Conclusion

In this chapter, we have summarized nonpharmacological treatments for behavioral and/or mental health problems in individuals with IDD. We provide a selective review of strategies that are often used in clinical practice and for which there is some evidence on their effectiveness for such problems. This chapter provides practical guidelines for adapting interventions to these individuals.

Adapting interventions to individuals with ID remains challenging for clinicians and established first-line interventions such as CBT may not always be accessible for these individuals, given their communication difficulties and cognitive and adaptive impairments. Clinicians and researchers continue searching for interventions that are focused on behavior change rather than cognitions. Behavioral activation may be an interesting development, in that this strategy is relatively easy to implement and it is less cognitively demanding. Recently, Jahoda and his colleagues [98] used an intervention called BeatIt in adults with moderate-to-mild ID for the treatment of depressive symptoms. The focus of BeatIt was on increasing activity, scheduling activity, and addressing barriers to activity. The results showed that behavioral activation was effective in reducing depressive symptoms, although the treatment was not more effective than a comparison condition of guided self-help intervention. A strong feature was that the intervention was delivered on an outreach basis by nurses and health professionals supporting the clients.

On the basis of this selective review and other reviews, we may conclude that a range of behavioral, cognitive, and other types of interventions are promising in the treatment of challenging behaviors and/or mental health problems in individuals with ID and comorbid conditions. The evidence-base has grown during the past two decades but is still relatively small when compared to individuals without IDD. Most studies have addressed challenging behaviors in individuals with

ID. Particularly, interventions for mental health problems in individuals with severe ID are lacking (see Vereenooghe et al. [86]). This chapter also shows that many studies reflect methodological shortcomings and that there remains a lot of work to be done by practitioners and researchers. We would like to paraphrase Vereenooghe and colleagues who state that bidirectional knowledge transfer is particularly important: research into ID making its way into the training of practitioners, as well as practitioners highlighting difficulties in assessment and treatment that need addressing (p. 18).

Tip

Adapt the intervention to the characteristics and learning style of individuals with intellectual disabilities.

Key Points

There are a range of interventions that may be effective in reducing challenging behaviors and/or mental health problems in individuals with intellectual disabilities.

Contextual interventions are just as important – and sometimes even more important – as client-directed interventions.

In both cases, interventions should be adapted to the characteristics and learning style of the client or his or her caregivers.

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Integrated Care

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Contents

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Learning Objectives

- To achieve a holistic consideration of physical, behavioural and mental health issues related to persons with intellectual and developmental disabilities, involving in their care a range of disciplines and health professionals.
- Integration across care may have many real and potential advantages, including identification of mental health issues, increased accessibility to mental health services, reduced costs and better outcomes.
- Integration requires the adoption of a person-focused approach which in turn is defined by a bio-psycho-social model.
- Given the high prevalence of physical and mental disorders in persons with intellectual and developmental disabilities and the high rates of physical–mental comorbidity, integrated care and approaches based on inter-disciplinarily input are urgently needed.

13.1 Introduction

Guaranteeing effective and appropriate integrated assistance for people with complex social and health needs represents one of the major challenges of healthcare systems. In Western countries, we are witnessing an increasing ageing of the population and a growing increase in the costs of services. The interest of researchers, service providers and policy makers in integration is not only limited to the elderly, but also to those who simultaneously use services deriving from multiple assistance systems, such as individuals with physical and intellectual disabilities or mental illnesses.

People with intellectual and developmental disabilities (IDD) are more vulnerable to physical and mental health problems than the general population. Besides this higher predisposition, they are often confronted with fragmented healthcare systems in which primary and specialty care is unable to meet their needs. Traditionally, healthcare for these persons has been parsed out to multiple providers and/or agencies and their physical and mental health are managed by different entities.

This lack of integration is made worse by a range of complicating factors, including barriers to the access and healthcare professionals who have received limited trainings about the peculiarities of these disabilities. Furthermore, several disparities emerge between the health sector and the social and education sectors which have significant implications for service planning and delivery. Changes need to be made in order to improve accessibility and integration of services necessary to meet the complex health needs of this population.

The development of integrated care pathways involving different professionals, family and life environments will help ensure that the needs of people with IDD are met. The necessity to address individual needs considering them in their entirety requires a planning of interventions which must take in account the principles of coordination, integration and cooperation between the various institutional levels that participate in the implementation of healthcare and social services. Integration allows the connection between healthcare (including acute care, primary and specialist care) and other assistance systems, improving outcomes both in terms of clinical results and quality of life. Identifying the links between different systems requires the adoption of a holistic approach which considers all major aspects of a person's life interrelated.

To date, the promotion of integrated and person-centred care represents an obligatory step for the improvement of public health policies outcomes. In order to achieve this goal, general understandings about integrated care should be converted into practical terms, in order to make available more effective health services able to improve quality of care and quality of life for the individuals. In this chapter, we discuss the notion of 'integrated care' and its features, exploring the many advantages deriving from its adoption in the field of IDD.

13.2 Integrated Care and Person-Centred Approach in Intellectual and Developmental Disabilities

Integrated care has more than one meaning and it is often used by different people to mean different things. The word 'integration' comes directly to us from the Latin verb 'integer', which means 'to complete'. Consequently, the definition of the adjective 'integrated' is 'combining or coordinating separate elements so as to provide a harmonious, interrelated whole'. It is mostly used to express the bringing together or merging of elements that were formerly separated [1].

The notion of integration in the fields of physical and mental health was first introduced more than a century ago by psychologist Alfred Adler (1870–1937) who codified concepts at the basis of individual psychology. According to this theoretical framework, the relationship between physical health and mental health is inextricable. The adoption of this concept in healthcare allows professionals to examine health status as a whole.

In its joint position statement on health, mental health, vision and dental care (as adopted in February 2013), the American Association on Intellectual and Developmental Disabilities has declared that 'all people, including people with IDD, should have timely access to high quality, comprehensive, accessible, affordable, appropriate healthcare that meets their individual needs, maximizes health, well-being and function, and increases independence and community participation'. Healthcare and social services face patients who have composite problems and this holds particularly true for people with IDD who are far more likely to have multiple comorbidities. People with IDD represent a vulnerable group of patients requiring well-timed, adequate and sensitive care interventions but actually, in several European countries, healthcare does not succeed in providing adequate services for them. Major difficulties refer to access to primary care, medical prescriptions, return of information, treatments for serious mental illnesses, communication between health and social services, lack of clear lines of responsibility and accountability for implementing care, and shortage of financial supports and resources [2, 3].

In the last two decades, the average life expectancy for persons with IDD living in high-income countries has increased, due to the improvement of life conditions and healthcare practices, although it remains lower than in the general population [4–6]. According to some studies, average age of death for persons with IDD is now 63.3 years for males and 69.9 years for females [7, 8]. Simultaneously, a rise of ageing-related diseases has been recorded [9]. These demographic trends suggest that the need for healthcare delivery systems and approaches should be able to coordinate primary, mental and behavioural care in order to address a variety of complex healthcare needs [10]. Nevertheless, people with IDD are still frequently confronted with fragmented healthcare in which primary and specialty care are unable to meet their peculiar needs [11, 12]. In fact, there are very few systems in which different components are integrated and this lack of integration is made worse by a number of complicating factors, such as the limited training received by healthcare providers about issues related to IDD [10, 13]. As a result, unmet physical and mental health needs are common across the lifespan of persons with IDD, and the challenges arise accordingly with increased severity levels of the impairments [14].

To date, the scientific evidence suggests that integration across care may have many real and potential advantages [15, 16]. Integrating primary healthcare with mental and behavioural healthcare entails a number of benefits which includes identification of mental health issues, increased accessibility to mental health services, reduced costs and better outcomes [17–20]. Integrated care was described by

Leutz as a broad inter-sectorial system approach that aims to connect the healthcare system with other human service systems in order to improve outcomes (clinical, satisfaction and efficiency) [21]. Over the last few years, the concept seems to have received attention in the field of IDD and now it is widely supported for improving accessibility and quality of healthcare. Nevertheless, a comprehensive shared knowledge about issues related to integrated care is still lacking. According to Dowling and collaborators, the barriers perceived to prevent its successful implementation could be due to philosophy or culture of care, power and funding structures, high levels of staff turnover and lack of training, lack of experience among service management, inadequate staff supervision and ambiguity among some stakeholders [22].

From a clinical point of view, integration requires the adoption of a person-focused approach rather than a disease-focused one. The main characteristic of person-focused care is defined by a bio-psycho-social approach applied to health which is aimed at comprehending the personal meaning of a disease attempting to comply with individual needs and preferences. From this point of view, diseases are simultaneously a medical, psychological and social problem [23]. Focusing on the disease reveals a clinical perspective that links the specific needs of an individual to separated biological entities [24, 25]. Commonly, health systems in Western countries adopt this perspective which often disregards the implicit reasons of illness. Nevertheless, a disease-focused approach seems to be inappropriate for patients with chronic and overlapping diseases [26]; in these cases, adopting a person-focused approach is more functional as it allows a holistic vision where most health and social issues are interrelated and the identification of the links. Integration is also required to supply an ongoing and comprehensive provision of services matched to the needs of the users. The major challenge is to convert general understanding about integrated care into practical terms to make available more effective health services able to improve quality of care and quality of life for the individuals [27].

An important contribution has been made by the International College of Person-centred Medicine (ICPCM) and the Person-centred Integrative Diagnosis (PID) multi-dimensional matrix [28]. The PID focuses the attention to the importance of involvement and partnership in the clinical care process and supports the patients' autonomy, responsibility and dignity while advancing the recovery and promotion of well-being. The construction of this theoretical model has been carried out by the World Psychiatric Association (WPA) through an Institutional Program on Psychiatry for the Person (IPPP) [29, 30]. Emerging features of the PID model include them being a diagnosis of health (of both illness and positive aspects of health), involving collaborative and empowering engagement of patients and serving as informational basis for prevention, treatment, rehabilitation and health promotion [31]. PID uses descriptive categories, dimensions and narratives, to manage patient-family-clinician partnerships for achieving shared diagnostic understanding and shared commitment to care. The application of this model to the assessment of personal well-being, experiences, satisfaction and aspirations of persons with IDD faces significant challenges as the self-reported assessment of these complex concepts is limited due to the cognitive and communication impairments which frequently occur in persons with IDD [32].

IDD may be a good example of how the holistic and comprehensive approach is useful for comprehending these complex constructs in healthcare. Integrated care and person-centred approach lay on the same conceptual frame of practice-based research. What is effective in evidence-based studies, including first-level studies, does not always have the same efficacy in the real world of everyday practice [33, 34]. The same applies to the relationship between costs and benefits of interventions: the dynamic interaction of the many factors conditioning the psychic state of persons with IDD and those who implement professional interventions can substantially change what is indicated by evidence-based studies. Moreover, some researchers have identified that even in organic medicine – in which the variables involved in the genesis and in the

resolution of the disorders are lower than those of the psychological and psychiatric clinic – the portion of patients to whom evidence-based guidelines are applicable without major adaptations often remains below 50% [35, 36].

An attempt to overcome these limitations is represented by research based on practice (practice-based research (PBR)), or by a collection and analysis of data derived from daily clinical practice and aimed directly at improving it. PBR is applied to professional studies, in outpatient clinics, in schools, in rehabilitation centres and in all other contexts in which people with IDD actually receive the interventions they need, becoming themselves an active part of the research process [37, 38]. The application of PBR can identify and solve the problems of applicability of the evidence from the scientific literature to the practices in various care settings [39, 40].

Integrated care promotes a holistic vision in which health and social problems are interconnected, allowing the identification of links between the different systems. Multiple perspectives may lead to the development of new intervention models based on person-oriented approaches to address individual characteristics and needs.

Tip

- Integrated care may be defined as a coherent set of methods and models regarding the provision of administrative-organizational and clinical services designed to create connectivity and collaboration within the healthcare contexts.
- From a clinical point of view, integrated care models require the adoption of a person-focused approach, in order to improve the patient's general well-being and take into account his/her needs.
- The integration of different health disciplines in the treatment of persons with IDD is increasingly indicated as a useful method, which may result in improved accessibility, appropriate use of healthcare services and higher quality of healthcare.

13.3 Integrated Care and Physical Ill-Health

It is widely proven that persons with IDD present a higher vulnerability to mental and physical health problems [41] (see ▶ Chap. 3) and face a much greater burden and earlier onset of physical ill-health than the general adult population [42, 43]. Given the high prevalence of physical and mental disorders in persons with IDD, they often tend to coexist. According to the findings of the Adult Psychiatric Morbidity Survey 2014 [44], chronic physical conditions such as asthma, cardiovascular disease, epilepsy and diabetes seem to be associated with common mental disorders (e.g. anxiety and depression) and poorer mental well-being in the general adult population. Barnett and colleagues [45] found the prevalence of physical–mental comorbidity to be 8.3% in the general population, and the likelihood of presenting mental disorders seemed to go up as the number of physical illnesses increased. It is widely known that physical–mental comorbidity is associated with poorer outcomes compared with the presence of either physical or psychiatric conditions alone [46, 47]. These high rates of comorbidity have some serious consequences, such as higher mortality [48], increased risk of suicide [47], functional impairments [49], poorer quality of life [50] and higher medical care costs [48].

To date, only few researches have examined the relationship between physical and mental ill-health in adults with IDD and those which have been identified have some limitations. Three studies have examined this relation focusing on older adults with IDD [51–53]. Cooper [52] observed a positive association between physical ill-health and dementia in a sample of 134 of older adults (aged 65 or over) but her study did not show further connections with other mental disorders. A complex relationship between the presence of epilepsy and mental health among adults with intellectual disabilities has been described [54], while McCarron and collaborators [53] have identified a significant

association between gastrointestinal disease – but no other physical conditions – and mental ill-health in a sample of 753 adults with IDD over the age of 40. More recently, Dunham and colleagues have found no significant correlations between physical multimorbidity and mental ill-health in adults with intellectual disability [55]. Nevertheless, this result could be due to the width of physical multi-morbidity (99.2% of the sample had physical conditions), which was so high to compromise the chance to identify associations.

Findings of some recent studies have shown that obesity is associated with mental ill-health and aggression [56–58]. Endocrine disorders were found to increase the risk of having problem behaviours by 22.4%; in particular, hyperthyroidism has been shown to be associated with psychosis, irritability/behaviour changes, restlessness and hyperkinesis [59]. Respiratory disorders have been identified as protective factors against problem behaviours and every type of mental disorders; this finding could be attributed to the ease of detection of symptoms (e.g. cough, breathlessness and wheeze), resulting in more attention by family members and/or carers which in turn produces a positive psychosocial effect [55].

Another significant association was found with reference to musculoskeletal problems. Actually, their presence seems to reduce the risk of mental disorders of any kind by 15.8%; this may be due to the anti-inflammatory effect of the drugs prescribed to treat these conditions. Some researchers have suggested that inflammation can induce depression. Makhija and Karunakaran have demonstrated that increased inflammatory cytokines are mechanistically linked to a number of interacting neural pathways that may contribute to the onset of depressive symptoms [60]. Other important findings concern ischaemic heart disease that has been found to increase the risk for the onset of problem behaviours by three-fold. Thombs and colleagues have shown that depression is around three times more common in patients after an acute myocardial infarction than in the general population [61]. The authors have also suggested that the mechanisms behind these associations in adults with IDD may be similar to those seen in adults who do not have IDD.

Given the high prevalence of physical and mental disorders in persons with IDD and the high rates of physical–mental comorbidity, integrated care and approaches based on interdisciplinarily input are urgently needed. The collaboration between psychiatrists and other professionals such as general practitioners, family doctors, neurologists, dental practitioners or orthopaedic surgeons [62–64] is, therefore, recommended. Good interdisciplinary communication seems to be associated with improvements in personal outcomes and in diagnostic and prognostic abilities of health professionals [65]. Providing specific trainings in the field of IDD for all health professionals seems to be fundamental in order to achieve a better knowledge of issues related to the identification and treatment for chronic health conditions which affect people with these conditions. Moreover, greater efforts should be made in order to improve the understanding of how to monitor mental and physical health among ageing IDD population.

13.4 Integrated Care and Mental Ill-Health

Diagnosing psychiatric symptoms in persons with IDD faces several difficulties, particularly in evaluating the impact of the specific symptoms on daily functioning. Assessment, diagnosis and treatment of mental problems in this group of patients demand specific adjustments due to the cognitive impairment, limited communication, sensory impairments, skill deficits, difficulties in adaptation and other disabilities [66]. Despite their complex and specific general health and mental health needs, people with IDD continue to experience disparities in healthcare provision. Unmet mental health needs are common across the lifespan of IDD, and the challenges increase accordingly with increased severity levels of the condition [14]. Particularly, the most urgent intervention required is the reduction of health

disparities, addressing to those issues which represent barriers to their access to services and appropriate treatments. Adopting adequate strategies is necessary to include people with IDD in health prevention programs, through an early identification of such problems, and to develop structured assessment tools coupled with tailored interventions [27].

As mentioned above, IDD presents several positive implications for psychiatry, such as providing assessment models, support system and diagnostic frameworks in severe mental and cognitive disorders. As suggested by Bertelli and colleagues, models of care – such as residential care, respite care and multi-disciplinary approach to care – as well as social issues of health – such as stigma and labelling and self-advocacy – were developed first in the IDD field, and now they are widely used in general psychiatry [27]. The usefulness of an integrated psychiatric assessment results from the consideration of several factors. Taking into account the high vulnerability of persons with IDD and the significant prevalence of mental health problems in this population, the adoption of a holistic approach that comprehends as many points of views as possible seems to be the most appropriate in their assessment.

Problem behaviours may be a significant example of the usefulness of an integrated assessment. In persons with IDD, the first choice treatment for problem behaviours is often pharmacotherapy and the search for a non-pharmacological therapy which considers the individual-specific conditions and individual quality of life as an outcome is often ignored. Actually, clinical practice suggests that an effective intervention of problem behaviours should be characterized by a simultaneous consideration of organic, psychiatric and socio-functional aspects and their pathogenetic contribution, on the basis of a multimodal analytical approach [27]. According to NICE guidelines [67], assessment on problem behaviours should be integrated into a comprehensive assessment for adults with autism. When assessing problem behaviours, practitioners should undertake a functional analysis in order to identify and evaluate any factors that may trigger or maintain the behaviour, including any physical health problems, the social environment (including relationships with friends, families and carers), the physical environment, coexisting mental health disorders, communication problems, changes to routine or personal circumstances.

The involvement of various disciplines in an integrated assessment might be useful in the identification of problems in the classification systems, strictly connected to clinical practices, and in identifying more and more sensitive diagnostic tools, instead of starting from very generic symptoms in assessing skills and performances [27]. Persons with IDD are uniquely positioned to benefit from the coordination of care that originates from integrated medical and mental and behavioural healthcare [10]. In fact, more than any other mental health condition, IDD provides opportunities to explore the clinical expression of the body-mind link. In order to have a clear understanding of it, all parameters of a quality mental healthcare such as holistic consideration of individual and sensitive diagnostic methods are highly relevant [32]. Multiple perspectives may lead to the development of new intervention models based on person-oriented approaches to address individual characteristics and needs.

The adoption of Patient-Reported Outcomes (PROs) is widely encouraged in assessing patients' performances and evaluating the efficacy of the treatments. These measures include the model of Quality of Life, widely applied in the field of IDD. In the field of IDD, since it is difficult to think about rehabilitative, educational and medical objectives that restore functional capabilities similar to those of most people, Quality of Life becomes a fundamental reference. It allows the assessment of the distance between individual expectations in the different areas of life and the results that are actually achievable; it helps to identify the most important and satisfying areas among the various interests and capabilities of individuals. Thus, a contextualized multi-modal assessment and a multi-disciplinary integrated intervention, involving different professionals, family and life environments, have several positive implications in achieving a comprehensive consideration of physical, behavioural and mental health issues related to persons with IDD, involving in their care a range of disciplines and

health professionals [27]. ► Box 13.1 summarizes the main factors supporting the relevance of an integrated care in psychiatry for IDD.

Tip

- Compared to the general population, persons with intellectual and developmental disabilities show a higher prevalence rate of both mental disorders and physical disorders.
- Due to their clinical peculiarities, persons with intellectual and developmental disabilities could particularly benefit from the coordination of multiple interventions deriving from the involvement of several specialists in the medical, mental and behavioural fields, as well as from different disciplines, services and supports for health promotion.

People with intellectual and developmental disabilities have complicated and ongoing needs and require a mix of services delivered by multiple providers. Integrated care improves mental and physical health treatments for people with intellectual and developmental disabilities establishing a comprehensive approach to patient, which enhances both quality of healthcare and quality of life.

Box 13.1: Reasons for Integrated Psychiatric Care in Persons with IDD Experiencing Mental Disorders

- Higher prevalence of psychiatric disorders than in the general population
- Identification of problems in the classification systems (i.e. ICD)
- New understanding for intelligence
- Models for the assessment of problem behaviours in severe mental disorders and cognitive deficit
- Genetic models for psychiatric disorders
- Direct clinical expression of the body-mind link
- Changes and adaptation of diagnostic criteria and diagnostic process for psychiatric disorders
- Sensitive diagnostic skills and tools (one often has to start from very generic symptoms, like behavioural changes or problems)
- Models for the assessment of adaptive skills as well as supports
- Life-span approach
- Holistic consideration of the person which requires multi-disciplinary intervention
- Model for high vulnerability to distress
- Person-related outcome measures, like generic quality of life

Bertelli et al. [27]

13.5 Conclusion

IDD represents an interesting area for the exploration and the understanding of the implementation of person-centred integrated care in classification, assessment, interventions, care delivery and policy planning [27]. In recent years, interest in healthcare integration has grown, with increasing evidence of its role in achieving better outcomes for mental health services' users. Nevertheless, reliable models and guidelines for daily practice still seem unclear, although persons with IDD, their families and an increasing number of healthcare providers are demanding them. To this purpose, health, social and educational professionals should collaborate to produce or enhance inter-disciplinary model of intervention and support, as well as to prove them through research trials and include them in the training curricula. This should be addressed also to primary care [10]. The necessity to encompass the existing models developed for the general population with the specific characteristics of IDD needs immediate attention [68].

Key Points

- Integrating primary healthcare with mental and behavioural healthcare entails a number of benefits.

- The early development of strategies of both person-centred care and integrated care in the field of intellectual disability and autism spectrum disorder may contribute to a better knowledge of the challenges of developing integrated care both in the interaction between primary care and secondary care and in the integration of health and social care.

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Outcome Measures and Inclusion

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Contents

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Learning Objectives

- Understand the difference between various outcome measures, especially between traditional medical outcome measures and person-centered outcome measures
- Understand the difficulties in assessing the psychometric properties and validity of outcome measures
- Understand the meaning and evolution of functioning, empowerment, subjective well-being, and quality of life as outcome measures in the field of ID and/or ASD
- Comprehend the articulation of inclusion outcomes and the issues associated with their measurement

14.1 Introduction

Since the end of the last century, outcome frameworks for mental health services, social care, and education have considerably changed and the last decade's policy of the World Psychiatric Association, "No Health Without Mental Health" [1], identified new high-level outcomes related to the aims of greater prevention, recovery, quality of life, and social inclusion. This shift in focus started earlier in the field of neurodevelopmental disabilities than in other areas of mental health as a key strategy to promote quality of patient's life and equity of healthcare.

The criteria to identify the most suitable interventions and to evaluate their effectiveness are extremely variable among different studies, or even in a single study, and differ depending on the purpose and the specific contexts, the level of assistance to be assessed, clinical characteristics of participants, and the sources of the available data. Many outcome measures were developed for use in population surveys or clinical trials and not for monitoring individual patients in daily practice, other measures were thought of as performance or cost indicators in mental health services (payment-by-results model) [2, 3]. Therefore, outcome measures and related tools are extremely heterogeneous, including assessment of symptom reduction, adaptive skills, functioning, specific abilities, intensity and pervasiveness of problem behaviors, level of use of services, and quality of service provision.

Measuring outcomes in persons with intellectual disability (ID) and autism spectrum disorder (ASD) poses even greater problems than in the general population because of the differences in sources of information and raters, the availability of specifically adapted tools, influence of cultural aspects, such as the conceptualization of developmental disabilities and their possibilities of improvement across the life span.

Research and clinical practice in this area have been scarcely developed and tended to generate outcome measures on their own rather than contributing to create and improve standard ones, although an excess of standardization could limit the applicability to such a wide range of individual variability as in ID and ASD.

Outcome measures are related to the very meaning of social health and educational practice and their optimization should represent a research priority in the field of mental health of neurodevelopmental disorders.

14.2 Outcome Measures

14.2.1 Heterogeneity of Outcome Measures

The evaluation of outcomes of therapeutic, rehabilitative, and educational interventions is becoming a key requirement for reliable and effective clinical practice and social-health services. However, the parameters that should be considered to evaluate the final result of an intervention are still widely discussed by the clinical and scientific community, and a consensus definition of the adequacy of the outcome measures and efficacy criteria has not yet been reached.

According to historical definitions, such as the one proposed by Donabedian [4, 5], outcome measures represent a part of a quality assessment system which also includes struc-

ture and process indicators, considering them a fundamental aspect of the assessment of the quality of interventions.

Outcome measures have been grouped by Epstein and Sherwood [6] into clinical, economic, and humanistic, with the clinical ones being the most frequently used in practice and research. Authors of the present chapters (in particular Bertelli and Bianco) propose a classification in four groups, clinical, economic, social, and person-centered, as in Table 14.1, although they believe that some measures straddle two groups, such as functioning with respect to the clinical and social groups or empowerment with respect to social and person-centered groups.

In addition to type, outcome measures can be framed on the basis of the source of information, mostly as clinician-rated outcome measures (CROM) and patient-reported outcome measures (PROM). Process measures, such as patient-reported experience measures (PREM), represent another important resource in the evaluation of healthcare quality, but they cannot be considered as a subtype of outcome measures and must be distinguished from them. PROMs allow evaluations of patients' perceptions of the efficacy of a clinical intervention, general health condition, or health condition related to a specific disease, while PREMs gather information on patients' views of their experience while receiving care (e.g., communication and timeliness of assistance).

In the field of ID and ASD, the term "outcome" refers in most cases to the final result of a process simplistically aimed to the reduction of the symptoms and dysfunctions related to the developmental disorder, mostly in terms of quantitative deviation from a standard (general population mean). However, there is considerable variability of outcomes definition and measurement, also according to numerous factors, such as the type and goal of the intervention they refer to, the clinical condition they address, and the context in which they are set. The age of the person to whom an intervention is directed is

Table 14.1 Main outcome measures by type

Clinical	Economic	Social	Person-centered
Intensity and pervasiveness of symptoms (by both patient and clinician reports)	Direct and indirect medical cost	Inclusion	Individual perception of symptoms impact on life quality
Functioning (by both patient and clinician reports)	Reduction of the need for admission to hospital	Occupation	Subjective well-being
Mortality	Utility (preference-weighted scales used in economic evaluation)	Caregiver burden	Quality of life
Life expectancy	Cost–benefit ratio	Education level	Patient satisfaction
Laboratory and instrumental examinations	Quality-adjusted life-years	Empowerment	
Intensity, frequency, and duration of problem behaviors			
Transition outcomes: engagement, adaptation, and continued growth and development			
Staying on treatment			
Safety and tolerability			

also important for the choice of the outcome measure, especially during the developmental age, in which the acquisition of nonspecific basic skills is relevant for all individuals.

This heterogeneity can also be found in scientific research, even in relation to the same intervention [7, 8]. Provenzani and collaborators have recently observed that 69% of the outcome measures found in the literature for ASD had been used only once and that only seven assessments had been included in over 5% of the studies. They also outlined that many of the assessments were not developed as outcome measures nor as specific for ASD [9].

As an example, the efficacy of psychopharmacological treatments for ASD has been alternatively evaluated in different studies in terms of overall clinical improvement, reduction of severity of core symptoms (i.e., social withdrawal or stereotypies), reduction of "non-core" (also called "associated") symptoms (i.e., aggressiveness, irritability, or hyperactivity), functioning, adaptive skills, and quality of life, to the point that precluded the possibility for providing precise conclusions in meta-analytic reviews [10–12].

Therefore, it is very complex to define whether an intervention is reasonable, useful, successful, and of good quality, especially when referring to persons with ID/ASD and co-occurrence of psychiatric disorders.

14.2.2 Validity and Reliability

Most current outcome measures were developed primarily for research purposes and with a consequent preoccupation with reliability and validity. Reliability is best interpreted as the repeatability of an assessment both over time and by different raters. If the same measure is used more than once on the same patient or the same group of patients within an appropriate time period, then the agreement between the two measures is an estimate of the stability over time, also called test–retest reliability. Similarly, the agreement between different raters assessing the same patient or group of patients is defined as interrater reliability. The more reliable the measure, the more stable and consistent should the various assessments result be.

A measure can be extremely reliable but have no validity whatsoever. Validity is the extent to which the scores from a measure represent the outcome they are intended to measure and is commonly judged through three main aspects: face, content, and criterion. Face validity is the extent to which a measure appears "on its face" to assess the outcome of interest and represents a very weak kind of evidence that a measure is measuring what it is supposed to. Construct validity is the extent to which a measure "covers" the outcome of interest and is assessed by carefully checking the measurement method against the conceptual definition of the construct. Criterion validity is the extent to which scores on a measure are correlated with variables that one would expect them to be correlated within reasons of their interconnection with the outcome of interest. When the criterion is measured at the same time as the construct, criterion validity is referred to as concurrent validity, while when the criterion is measured to predict a future outcome, it is referred to as predictive validity. Criteria can also refer to other measures of the same outcome, which is known as convergent validity.

Although in the last two decades, new outcome measures have been developed with increasing reliability and validity, practices for persons with ID/ASD have continued to rely on traditional ones concerning objective and standardized aspects of health, such as symptoms intensity and pervasiveness, biomarkers (blood tests or radiographic images), behavior, functioning, mortality, hospital, and services utilization, or other aspects related to morphological and functional integrity [13, 14]. The theoretical model of these traditional measures was developed for the general population and foresees that therapeutic, rehabilitative, and educational interventions tend toward normalization, that is to make a person as close as possible to the structure and functioning of most people. These measures have the advantage of being easy to measure, highly reliable, and suitable to a system for costing mental health services that links quality and outcome to funding, but they lack

appropriateness, responsiveness, precision, acceptability, and feasibility for ID and ASD population, in particular, they do not include actual possible goals of many conditions of disability and fail to capture the patient's experience [2, 7].

Traditional outcome measures concerning objective and standardized aspects of health have the advantage of being easy to measure, highly reliable, and suitable to link healthcare quality and outcome to funding, but they do not include actual possible goals of many conditions of disability and fail to capture the patient's experience.

14.2.3 Functioning

Individual "functioning" is an umbrella term which has been used across time with very different meanings and in reference to very different characteristics of human beings and their environment as well as life activities. The focus of functioning as one of the last 25 years' most important outcome measures of interventions for persons with ID and/or ASD [15] reflects the 1980s' and 1990's movement away from a primary emphasis on diagnosis and classification to a prevalent consideration of planning and provision of individualized supports [16]. In fact, research findings of the last few decades showed that medical diagnosis alone does not predict service needs, level of care, work potential, the likelihood of social integration or functional outcomes, and that the incidence and severity of disability in a population can be reduced by modifying those features of the social and physical environment that can enhance the functional capacity of persons and improve their performance [17]. This increasing consideration in healthcare of people with ID/ASD and other lifelong disabilities of aspects other than those defined by the impairment has implied the medical and the social models to interact with each other and try to come to a common ground.

The medical model deals with disabled bodies and minds for their part that is considered to be pathological or defective, while the social model approaches disability for what results from attitudes and other features of the social environment.

A relevant attempt to measure the impact of illness and its consequences on individual functioning was realized in 1980 by the World Health Organization (WHO) through the International Classification of Impairments, Disabilities and Handicaps (ICIDH) [18]. The ICIDH is based on a tripartite distinction between impairment, disability, and handicap, where impairment refers to a loss of structure, or abnormality of function at the organ level, disability to a restriction of actions at the person level, and handicap as a set of disadvantages within the individual's social context [19]. Comparison of impairment and disability profiles highlights the role of functional limitation in mediating between disease (medical model) and disability (social model).

Another interesting perspective toward understanding the implication of human functioning in the definition of disability, specifically ID, was proposed by the American Association on Mental Retardation (currently American Association on Intellectual and Developmental Disabilities; AAIDD) in its 1992 manual [20] and further refined in 2002 [21]. In this view, substantial limitations of functioning and associated dimensions, determined by the dynamic interaction between health condition and environmental factors, represent the main diagnostic criterion for ID and the main outcomes to be pursued through an adequate system of interventions and supports. This enhancement of human functioning has to address all its dimensions, which are identified in intellectual abilities, adaptive behavior, health, participation, and context [22]. Intellectual abilities include reasoning, planning, problem-solving, thinking abstractly, comprehending complex ideas, learning quickly, and learning from experience; adaptive behavior, on the other hand, refers to the collection of conceptual, social, and practical skills that have been learned and are performed by persons in their everyday lives. However, health describes a state of complete physical, mental, and social well-being; participation

addresses the performance in actual activities of social life domains; context refers to all the interrelated conditions within which persons with ID live their everyday lives; and context represents an ecological perspective that involves the micro (family and immediate social setting), meso (neighborhood, community, and support services), and macrosystems (overarching patterns of culture and socio-political influences) around the person. Furthermore, as mainly defined by the level of individual functioning, ID is no longer seen as necessarily a lifelong condition, but its existence or degree is dependent on the provision of appropriate supports [21].

A similar framework was proposed around the same time by the WHO in the International Classification of Functioning, Disability, and Health (ICF) [17], which represents the evolution of the ICIDH and the main reference system to date for the definition of functioning as a descriptor of health and health-related states. In ICF, human functioning is articulated in the three levels of body or body part, the whole person, and the whole person in a social context, while disability refers to impairments, activity limitations, and participation restrictions (see Table 14.2).

The ICF "mainstreams" the conditions of ill-health and disability and redefines them as universal human experiences by acknowledging that every human being, not only a minority of the population, can experience a decrement in health and thereby some disability. By shifting the focus from causes to impact, it also places all health conditions on an equal footing allowing them to be compared using a common metric.

The ICF is based on the biopsychosocial model [23] and provides a coherent view of the biological, individual, and social perspectives of health. As such, it complements the International Classification of Diseases [24] to create a broader and more meaningful picture of the experience of the health of individuals and populations.

At the beginning of the last decade, the AAIDD produced an upgrade of its systems approach to human functioning, whose input and impact (output) could be evaluated based on three human functioning outcomes: social-economic status, health status, and subjective well-being [25, 26]. This model reflects the twenty-first-century style of systems thinking, synthesis, and alignment, which focuses on the factors that affect human functioning, integrates information from multiple sources, and places critical organization and system functions into a logical sequence. These out-

Table 14.2 Levels of human functioning classified by the ICF

Level	ICF component	Definition
Body or body part	Body structures	Anatomical parts of the body such as organs, limbs and their components
	Body functions	Physiological functions of body systems (including neuropsychological and psychological functions)
	Impairments	Problems in body function or structure such as a significant deviation or loss
Whole person	Personal factors	Particular background of an individual's life and living, such as gender, age, coping styles, social background, education, profession, past and current experience, overall behavior pattern, character
	Activity	Execution of a task or action by an individual
Whole person in a social context	Environmental factors	Physical, social, and attitudinal environment in which people live and conduct their lives
	Participation	Involvement in a life situation

come measures were proposed in responsive to the move toward evidence-based practice, quality management of health and social services, and the need for indicators associated with subjective well-being and human rights [25, 27]. A further extension of this concept has been developed recently, to address the current scientific and cultural shift from a person-environment paradigm to a multidimensional contextual paradigm. According to this update, context is defined as "a concept that integrates the totality of circumstances that comprise the milieu of human functioning" and is conceptualized as being multilevel, multifactorial, and interactive. The multilevel component describes the ecological systems within which people live, are educated, work, and recreate. The multifactorial component refers to the potentially influential factors within the ecological systems, and the interactive component identifies the variety of ways in which levels and factors interact to influence personal outcomes [28].

In the ICF, functioning is conceptualized as the result of a dynamic interaction between individual health conditions, environmental factors, and personal factors. This conceptualization is aligned with a biopsychosocial model of disability, based on an integration of the social and medical models of disability.

14.2.4 Empowerment

Since the 1980s, empowerment has been recognized by the WHO and other health agencies around the world as a core concept in health promotion [29] and has been used widely in the disability field to describe both a desirable process and a desirable outcome for health and social policies and services. It reflected a parallel development of the areas of advocacy, consumer rights, and professional practice and integrates the achievement of social equity.

One major contributor to the development of the concept of empowerment was the social scientist Julian Rappaport, who defined empowerment as a "concept that suggests both individual determination over one's life and democratic participation in the life of one's community … both a psychological sense of personal control or influence and a concern with actual social influence, political power, and legal rights" [30]. Furthermore, he indicated that "empowerment is a process by which people, organizations, and communities gain mastery over their affairs. Consequently, empowerment will look different in its manifest content for different people, organizations, and settings" [30].

Empowerment appears in the literature of several disciplines such as psychology [30–33], social welfare [34–40], education [41–43], and medicine [44, 45], especially psychiatry [46–48].

Despite its widespread use across decades, a systematic definition of the concept is still lacking. It has much intuitive appeal and arouses images of growth, fulfillment, triumph, righteousness, and achievement of justice despite circumstances that may hinder such an outcome. Empowerment is more often defined according to some of its anticipated outcomes rather than its very nature [49]. In the field of mental health, empowerment refers to the level of choice, influence, and control that users of mental health services can exercise over events in their lives. The keys to empowerment are the removal of barriers between individuals, communities, services, and governments, the attention to self-help and self-reliance of service users, and the recognition that services may be delivered in ways that promote, rather than inhibit, personal growth and focus on individual strengths instead of deficiencies [37, 50, 51]. Rappaport argued that empowerment may be easier to define by its absence (e.g., alienation and powerlessness) than by its presence [52].

Dunst and collaborators asserted that the term empowerment has been used in six interrelated ways, such as philosophy, paradigm, process, partnership, performance, and perceptions, which can be further grouped into the three key elements of ideology (comprising philosophy and paradigm), participatory activities (including process and partnership), and outcome indicators (comprising performance and perceptions) [53].

Table 14.3 Key components of empowerment

Component	Description
Self-efficacy	Belief in the ability to produce intended results; self-perception of being capable of exercising some control over the quality of life
Participation and collaboration	A collaborative relationship between help-seeker and help-giver in which the former is encouraged to assume as much responsibility as possible in decision-making
Sense of control	The attribution of attitude or behavior change to one's own actions; it is rather near to the concept of *locus of control* (see ► Chap. 10), which represents an allied construct of empowerment
Meeting personal needs	Empowering interventions must address the needs and aspirations of individuals in ways that may make the individual more capable and competent
Understanding the environment	Aspects of the environment that an individual may utilize to meet his/her own needs
Personal action	Taking personal actions to address a need. Several authors pointed out that it may not be an essential component of empowerment, since empowerment may also be expressed in ways other than personal action such as an improvement in attitude about dealing with a problem or an increased awareness of the options that may be pursued to meet a personal need
Access to resources	Personal influence on the possession of relevant resources, information, and skills. Resources include sources of support such as friends and relatives, community groups and organizations, government assistance, and self-help groups

At the end of the last century, a literature review by Dempsey and Foreman [54] found some explicit and implicit similarities between various definitions of empowerment as an outcome of disability service provision and some general consensus on its components. They also identified key components of these definitions of empowerment through a process of content analysis, which are summarized in Table 14.3.

Several attempts have been made to integrate the components of empowerment into a consolidated model, with three component models being the most common which includes personal, interpersonal, and community (or political, or social, or behavioral) level engagement [55–57]. Koren and colleagues proposed to add a second dimension of classification referred to as individual expressions of empowerment comprising three components, attitudes, knowledge, and behavior [58]. A very comprehensive model has been developed by Dunst and colleagues, which includes three dimensions (levels, form, and context) articulated in a 96-cell matrix. It provides a precise framework of the concept of empowerment and offers a schema to conduct research that may systematically refine theoretical assumptions [53].

A wide range of scales have been developed for the measurement of empowerment, but the qualities of these have not been rigorously assessed. A systematic evaluation was carried out by Cyril and colleagues using the following six criteria: use of an a priori explicit theoretical framework, content validity, internal reliability, structural validity, confirmatory factor analyses to determine internal construct validity, and external construct validity. Only 9 (45%) studies on as many tools were rated as high quality (15% of studies scored 6 and 30% scored 5), 10 (50%) studies were rated as medium quality (30% scored 4 and 20% scored 3), and 1 study was rated as poor

quality. The vast majority of studies measured individual empowerment, only 15% measured community (10%) and organizational (5%) empowerment [59].

In the field of ID and ASD, empowerment has received a lot of attention in research and social-health policy papers over the last decades and it has been reported as the main outcome of self-advocacy [60]. Some studies found support and assistance to families, programs of rehabilitation from education services and non-governmental organizations to be key aspects in the development of social empowerment of persons with ID/ASD [61–69]. Other studies indicated a significant contribution of social empowerment on life quality, social well-being, and social welfare, while communicative empowerment has been shown to improve personal communication skills [61, 69–74]. Some authors have also suggested standards and indicators to facilitate the social empowerment of individuals with ID related to social justice, healthcare, social rehabilitation, and job opportunities [61].

Many important aspects have yet to be explored or addressed, some of which have great implications for applying empowerment to daily practices such as the extent to which empowerment is influenced by the severity of cognitive and other individual characteristics, stability of empowerment over time, the importance of behavioral versus attitudinal components of empowerment, the nature of the relationship between empowerment and community intervention, the overlap and the differences between empowerment and whole-person quality of life (see ▶ Chap. 15), and the extent to which persons with more severe ID may become empowered.

Despite its widespread use, a systematic definition of empowerment is still lacking. In the field of mental health, it refers to the level of choice, influence, and control that users of services can exercise over events in their lives. Fundamental aspects are represented by the promotion rather than inhibition of personal growth and the focus on individual strengths instead of deficiencies. Research on ID and ASD found improvement of communication skills, support and assistance to families, programs of rehabilitation from education services and non-governmental organizations to have a major positive impact on personal empowerment.

14.2.5 Person-Centered Outcome

During the last four decades, there has been a profound transformation of practices and interventions addressed to persons with ID and ASD. The typical models of treatment and rehabilitation, mainly aimed at restoring structures and functions that have been affected by the developmental disorders and any co-occurrent morbid conditions, have progressively been shown as inadequate to these persons and gave way to new multidimensional and person-oriented approaches, which pointed at promoting satisfaction with life regardless of the severity of disability and co-occurrent physical and mental health issues. In fact, setting the goal of therapeutic interventions for people with ID/ASD on the achievement of a personal functioning that is as similar as possible to that of people with typical development is not just unreasonable but may favor the development of prejudices of irrecoverability and health disparities [75].

This progressive, sometimes nonlinear, change of models also involved the definitions of ID and ASD, leading to the adaptation of the main nosographic systems [76, 77], the legislative framework, the services organization, the planning of therapeutic-rehabilitative interventions, and the consideration of the person with ID and/or ASD as part of society.

The adoption of a "person-centered" perspective, based on holistic, multidimensional, and bio-psychosocial definitions, is fundamental in the evaluation of the outcomes of the interventions and the state of health of persons with ID and/or ASD. In fact, the physical and mental health needs of this population address an area of care that straddles health and social services, general and specialized medicine, services for the developmental age, adulthood, and aging.

This broadening of focus in the measurement of health, beyond traditional health

indicators started in the second half of the last century, when the increasingly mechanistic model of medicine, concerned only with the eradication of disease and symptoms, raised the need for the introduction of a humanistic element into healthcare (revalued in its component of humanistic transaction) and the consideration of the patient's well-being as a primary aim. In that time, the WHO expanded the definition of health to "a state of physical, mental and social well-being, not merely the absence of disease and infirmity" [78, 79] and suggested to policy makers to increasingly include measures of the impact of disease and impairment on daily activities and behavior, perceived health measures, and measures of functional status. Nevertheless, while beginning to provide a more articulated perspective of the impact of diseases and their treatment on patients' life, these measures did not include patients' desires, values, family situations, social circumstances, and lifestyles [80].

The increasing emphasis of the last two decades upon person-centered outcomes in healthcare, and related disability and rehabilitation literature, seems to have represented an attempt to contrast the persistence of the tendency to focus outcome measures of interventions on illness and disability, rather than health and ability. That is, health continues to be seen primarily as the absence of illness or disability rather than a resource for daily living [81]. On the contrary, person-centered care, especially mental healthcare, see patients as individuals to work with rather than passive recipients of aid and tries to help them to feel satisfied with their own lives in ways that are customary to them and valued by them [14]. These and other main principles underlying person-centered care that have been found by research to be most important for affecting outcomes are summarized below:

- Approaching the patient as a person
- Identifying patient's values, interests, preferences, and goals
- Seeing the patient as an expert about their own health and care
- Providing good communication, information, education, and emotional support
- Involving family and friends
- Sharing power and responsibility
- Taking a multidimensional approach to assessing people's needs and providing care (continuity between and within services).

In this sense, person-centered outcome measures do not coincide with subjective outcome measures but represent only a part of them. In fact, as mentioned earlier in the text, many subjective outcome measures refer to the patients' perception of their own clinical condition or functioning compared to that expected by the norm. On the other hand, person-centered outcomes include some objective measures, whose relationship with subjective ones has yet to be clarified as well as the impact of personal characteristics (i.e., cognitive profile, adaptive skills, and problem behaviors) and environmental variables (i.e., living arrangement, family, professional caregivers, services) on the variation of either objective or subjective outcomes.

Perry and collaborators found person-centered outcome measures, such as satisfaction, to correlate little with objective indicators and to be not predicted by personal and environmental characteristics. In contrast, objective measures of other subjective outcomes, such as choice, involvement in activities, and social and community affiliation were significantly dependent on personal and environmental characteristics [82].

The literature is consistent in pointing out that person-centered outcome measures can promote the development of a more accessible, efficient, and cost-effective service network to provide effective and satisfying assistance.

In the United States, Australia, and European countries, where the implementation of healthcare models based on this type of approach is prevalent for a long time, the most important obstacles have been the resistance to change in the philosophy and culture of care, problems related to financing, the high turnover of staff, and the lack of experience of professionals involved in management and organization [83–85]. A very interesting contribution was provided by the International Network of Person-Centered

Medicine (INPCM), which proposed the use of evaluation and intervention procedures based on the collaboration between clinicians, patients, and family members, in perspective which is complementary to the traditional nosological one [86].

Contrary to what is commonly believed, person-centered outcome measures can favor the development of a more accessible, efficient, and economical services network.

Person-centered outcomes focus on holistic and multidimensional aspects of health than symptoms intensity or pervasiveness of a disease or a disorder. Thus, person-centered outcome measures do not coincide with subjective outcome measures, which instead may refer to traditional clinical indicators.

14.2.6 Subjective Well-Being

The term subjective well-being (SWB) refers to a general area of scientific interest rather than a single specific construct as it is commonly used to describe a broad category of psychological phenomena that include individual emotional responses, feeling, and mood (e.g., contentment, happiness, absence of depression, or anxiety), satisfaction with different aspects and situations of life, fulfillment and positive functioning, and global judgments of life satisfaction [88–90]. Thus, SWB is often subdivided into different constructs, which correlate substantially with each other and need to be specified in their own [90]. In some contexts, SWB is used in a restrictive way with a specific reference to good living conditions (e.g., housing, employment). Tracking these conditions is important for public policy. However, many indicators that measure living conditions fail to measure what people think and feel about their lives. According to Knight and McNaught, SWB has to be understood as dynamically constructed interconnections among people's circumstances, locality, activities, and psychological resources, including family and wider interpersonal relations [91].

In simple terms, well-being can be described as judging different aspects of life positively and feeling good. In the field of mental healthcare, emotional well-being and psychological well-being (e.g., feeling very happy and full of energy) are viewed as critical to overall well-being. The different aspects of well-being that have identified and examined by researchers from different disciplines [87, 89, 92–96] are summarized here down:

- Physical well-being
- Economic well-being
- Social well-being
- Development and activity
- Emotional well-being
- Psychological well-being
- Life satisfaction
- Happiness
- Domain-specific satisfaction
- Engaging in activities and work

Emotional well-being, happiness, and satisfaction are closely related. Cummins asserted that "some combination of happiness, or positive affect, and life satisfaction" are frequently involved in the definition of SWB, "but while such a view has intuitive appeal and face validity, closer inspection reveals very different ideas on the nature of the component parts and almost no information on how the components combine. Some authors simply declare happiness and life satisfaction to be synonymous, but this view is not supported by a considerable body of literature. Measures of happiness and satisfaction share a maximum of 50–60 common variance. These data confirm the view expressed by others that while happiness and satisfaction may indeed form part of the SWB construct, it is heuristically useful to measure and analyze them separately" [97]. According to Helm, happiness represents one component of satisfaction and is more transitory than the latter, as it reflects the positive and negative affects of positive or negative emotions and moods [98]. Satisfaction has been appointed by Diener, Cummins, and other authors to express global judgments about one's life overall or specific life domains, to show trait-like stability over time, and to be not predicted by environmental situations or personal characteristics such as skills or challenging behaviors [87, 99–101].

Considerable research involving the predictors of SWB in the general population has been done by Myers and collaborators, and Cummins, who have found that people are happier with overall life than one might expect (average of 6.5 on a scale from 0 to 10) and that happiness seems to be associated more with a combination of personal characteristics and sociocultural factors, such as extroversion, meeting intellectual challenges, social belonging, social support, health, religiosity, personal freedom, and income, than with external circumstances and individual backgrounds such as age, economic class, race, and education level [97, 102, 103].

One aspect of high importance is the individual capacity to change the way of thinking about one's own life. Brickman and Campbell argue that an individual can learn how to be more optimistic by changing how they think about themselves [104].

The development of the concept and application of SWB started in the process of implementing research on the quality of life (QoL) with consequent confusion between the two concepts, which is still frequently found. SWB is often regarded as a subjective or psychological aspect of QoL [105]. Actually, the two concepts, although presenting some overlap and interconnection, have been progressively distinguished. QoL represents a complex evaluation of the quality level reached by an individual in those aspects of life that can apply to anybody's life, whereas SWB refers to an affective state toward any aspect of life. Cummins [106] claims that the QoL variables that are closest to SWB are the least sensitive, while Diener and Suh [107] think that measures of SWB, economic indices, and social indicators can complement each other in assessing QoL.

Despite a voluminous literature on the general population, there are only a handful of studies on SWB on individuals with disabilities. Results from available studies show that the persons with disability have a significantly lower SWB than persons without disability and among the former, the lowest level of SWB is reported by individuals between the age of 45 and 54 and by those with physical health impairments as compared with those with mental disabilities and sensory impairment [108]. In the field of ID, research appears to have been limited by measuring issues such as the construction and administration of scales, and validity or reliability of self-reports. Proxy reporting, which involves the provision of responses by another person (e.g., a relative, friend, healthcare professional) on behalf of the person with ID, has also raised serious concerns about its reliability and validity and whether it accurately reflects the perceptions of the individual concerned [109, 110].

An interesting line of research was conducted by Cummins and colleagues, who also adapted for mild and moderate ID a tool widely used in the general population, the Personal Wellbeing Index [111]. This scale includes eight life domains (standard of living, personal health, achievement in life, personal relationships, personal safety, community connectedness, future security, and religion/spirituality), which represent the first-level deconstruction of "life as a whole" and provide insights into the various aspects that shape SWB [99, 112, 113]. The authors of these studies also formulated a theory to explain a possible cause of the repeated finding of similar levels of SWB in individuals with and without ID [113–115]. This theory, named "theory of SWB homeostasis," posits that all humans, independently form their cognitive characteristics, have the psychological ability to homeostatically maintain levels of SWB within a relatively narrow range even in the presence of persistent adverse factors [116–118]. A similar theory was developed by Brickman and Campbell with the name of "hedonic treadmill," which is focused on the idea that individuals tend to fast adapt to changes in life and return to the baseline level of happiness [104].

In persons with ASD, IQ and happiness emerged as highly interconnected central factors that can funnel and influence the network of factors more than others [119, 120]. Self-reported IQ appears to be connected to SWB indirectly, as a resource that allows the individual to engage in social relations and adaptive functioning and increase in turn self-esteem, self-confidence, and self-efficacy [120, 121].

Also, social satisfaction emerges as highly important for SWB in persons with ASD. Nevertheless, the number of social contacts from different contexts (such as family, work, leisure activities) does not seem to influence SWB directly, but merely through one's satisfaction with these contacts [120, 122]. Moderate use of Facebook has resulted to enhance SWB and has a protective action against co-occurrence of anxiety and other mental health issues [123].

One of the strongest risk factors for low SWB in ASD has been reported to be the number of physical problems [120]. Pain and physical stress also act as causes or triggers for problem behaviors that, in turn, seem to influence an individual's level of well-being [124]. Another factor with a considerable negative impact on SWB is the co-occurrence of other neurodevelopmental disorders or other psychiatric disorders [121].

- SWB is commonly used to describe a broad category of psychological phenomena that includes emotional responses, feeling, and mood, satisfaction with different aspects and situations of life, fulfillment and positive functioning, and global judgments of life satisfaction.

- Despite a voluminous literature on the general population, there are only a handful of studies on SWB on individuals with disabilities. In the field of ID, research has been limited by measuring issues such as the construction and administration of scales, and validity or reliability of self and proxy reports.

- In persons with ASD, IQ, social relations, adaptive functioning, and physical problems have been reported among the factors with the highest impact on SWB.

14.2.7 Quality of Life

Among person-centered approaches, Quality of Life (QoL) has raised particular interest in the scientific community in the last three decades with a large number of publications of research and conceptual articles as well as textbooks. At present, QoL is an important and commonly held goal in services and supports for people with ID and ASD and takes place across all interventions such as social care, education, and medicine, especially in psychiatry.

Despite its long history in the literature and the last decade's shift of scientific interest from theoretical issues and implications to ways of measurement, there is still disagreement as to how QoL should be defined and measured.

The quality of existence has been an important human concern since antiquity and can be placed at the origin of ethics and philosophy. The duty to care for the quality of living of those who have a disability represents the principles of charity and brotherhood which were already shared in early Christian societies and which were included in philosophic models of the state since the renaissance [125, 126]. Nevertheless, scientific research into the concept of QoL gained prominence only following Thorndike's [127] work on life in cities and environmentally based social indicators. In the 1960s, large cross-cultural studies were carried out, which aimed at investigating QoL in different countries, with a prevalent reference to socio-economic indicators, infant mortality, opportunities for access to study, work, and healthcare. A more individual-focused approach was developed in the 1980s, especially in the United States, although the first research aimed at systematically exploring the subjective perception of psychiatric patients about their living conditions was carried out already in 1969 by Fairweather and collaborators within a wider evaluation of the outcomes of a program of community intervention [128]. During this period, various models and definitions of personal QoL were produced, among which the three that have most influenced the evolution of the concept up to now: the satisfaction model, the importance-satisfaction model, and the role functionality model.

The first two models basically refer to a subjective perspective, which favors the subjective experience of persons with disabilities and/or mental health problems, the third

instead refers to an objective perspective, which favors objective indicators of QoL, such as social, environmental, and occupational conditions.

According to the satisfaction model, initially proposed by Lehman and collaborators [129], and Baker and Intagliata [130], QoL consists of the subjective perception of satisfaction with life, determined by the relationship (or gap) between current living conditions of a person and those conditions desired by the same individual. This gap has been called the Colman's gap, from the name of the scholar who elaborated it [131]. The satisfaction model includes three components: personal characteristics, objective living conditions in various areas of life, and personal satisfaction with the same areas [132]. McDowell and Newell [133] suggested that QoL "relates both to the adequacy of material circumstances and to people's feelings about these circumstances" (p. 205), while Coulter [134] defines QoL as "a sense of personal satisfaction with life that is more than just pleasure or happiness and yet something less than meaning or fulfilment" (p. 61).

The combined importance-satisfaction model incorporates both the subjective satisfaction with the various areas of life and the importance that each of these areas has for the person. Importance, which is aligned with preferences, desires, expectations, and values, significantly influences the subjective perception of QoL and allows to explain why persons living in totally different conditions can express similar levels of satisfaction or, on the other hand, why persons living in very similar contexts perceive very different levels of satisfaction [135].

The combined importance-satisfaction model is the model on which two of the main QoL evaluation tools for persons with ID and/or ASD are based, which are the one of the WHO [136, 137] and that of the Toronto University [138, 139]. The WHO defines QoL as the "individuals' perception of their position in life in the context of the culture and value systems in which they live, and in relation to their goals, expectations, standards and concerns" [140], while the researchers of the Toronto University refer to "the degree to which a person enjoys the important possibilities of his or her life" in the three categories of "being," "belonging," and "becoming," which respectively describes who one is, how one is not connected to one's environment, and whether one achieves one's personal goals, hopes, and aspirations [141]. This model of the Toronto University is linked in a straightforward way with the Hans Reinders' QoL concept and the well-known capability theory proposed by Amartya Sen [141]. According to Reinders [142], a dynamic approach to QoL "is oriented toward the goal of human flourishing. Human beings flourish to the extent that they are enabled to develop their own capabilities … when they receive sufficient opportunities to develop their own gifts and talents". In Sen's perspective, agency goals are those objectives that people set themselves, even if they do not benefit directly from them, or indeed even if they undermine other capabilities [141].

The role functionality model is based on the assumption that the social environment demands a variety of role performances from the individual. For example, in the friend role, one is expected to support, give advice, be loyal, and protect. In the work role, one must concentrate, get along with colleagues without excessive conflict, be punctual, and be productive. The individual meets society's demands through his cognitive, affective, behavioral, and perceptual abilities, and he/she receives in return opportunities to have his or her needs satisfied. As far as adequate satisfaction and performance are achieved, the individual is adjusted to his or her social context and has a high QoL [143].

In other words, in this theoretical framework, QoL consists of a state of satisfaction for the individual related to the extent to which he/she meets the performance requirements of his or her environment. At the level of the social and healthcare system for mental health, the negotiation between client's and service's outcomes is expressed in this QoL model under the heading of satisfaction and performance (functioning), which together constitute adjustment.

An alternative classification of QoL models has been proposed by Borthwick-Duffy,

based on the following four main groups: life conditions, satisfaction with these conditions, a combination of life conditions and satisfaction, and a combination of life conditions and satisfaction modulated by personal values, aspirations, and expectations [144].

Michalos carried out a thoughtful analysis of the various uses of the term "quality" in QoL, finding that at a minimum it can describe the characteristics of a population, such as gender, income, age, etc., while in a second sense, it can depict the value or worth of something. Michalos termed the former as the descriptive use and the latter as the evaluative use of quality in QoL [145].

Cummins argued that one of the characteristics of a good QoL model and measure is that it should refer to characteristics of individual life that are shared with other people (shared QoL) and enable comparison between groups of people regardless of their characteristics, including cognitive and relational difficulties [146, 147]. Other main characteristics of a good QoL model are the consideration of the person as a whole (whole-person QoL) and the exclusion of standardized goals. The QoL approach has to be interpreted in qualitative rather than quantitative terms, as it does not coincide with the achievement of a high quality of living but with an exploration of the rich intricacies of the personal perception of quality toward life and the provision of a way for healthcare professionals to view the patient-person and his/her relationship with the system of care. QoL assessment should not represent a classification of individuals, services, or systems, but it should help provide, within service systems and organizations, a value system that is consistent with those values held by people with ID/ASD [14].

In contrast to this view of whole-person QoL, a completely different interpretation has been publicized widely over the past two decades with the main focus on those domains of life that are considered to be affected by a particular illness. This branch of QoL practice and research, which is called health-related QoL (HR-QoL), typically tries to measure both the degree to which symptoms of a specific disease are present and their effects on daily functioning. Thus, although it focuses on the individual in a way that is of some benefit, they still support a theoretical perspective of QoL that has not significantly departed from the traditional medical approach [14].

In the field of ID/ASD, QoL models and measures are further distinguished on the basis of the consideration of personal QoL and family QoL. The first aspect proceeds from the fact that each individual has a changing set of personal attributes that determine the subjective experience of life, while in the second aspect, shared and personal QoL are applied to the family that includes a person with ID/ASD. Disability impacts the whole family and the determination of appropriate conceptualization of family outcomes requires an understanding of the impact of members with a disability on family QoL [148, 149].

At the current state of knowledge, it seems that the best way to carry out a QoL assessment is to follow a comprehensive approach, which integrates subjective and objective aspects, self-reports and multiple evaluations [150]. Such assessment may be used effectively with all people with NDD, independently from the severity of their functioning impairment. Individuals with higher communication and cognitive limits may express their inner states through consistent behavioral repertoires, which can be discerned by people closest to them and validated by more independent others. A good example of how this can be implemented is the Lyons' Life Satisfaction Matrix [151].

The field of QoL has developed rapidly, and QoL measures have been improved to a degree that they have several solid uses. Still, there is much to do, as currently available tools, although they have some common conceptual and evaluation characteristics, still show considerable differences in the areas to be included in "shared QoL," the dimensions used to evaluate "Individual QoL," and the role attributed to indicators of QoL.

A more detailed discussion of all the aspects of QoL mentioned in this paragraph and other aspects has been provided in the specific chapter included in this textbook (► Chap. 15).

Despite its widespread implementation, QoL measurement is still associated with numerous problems, related to the definition of the QoL concept, the way it can be effectively measured, for whom, by whom, and to what purpose. QoL measures can be grouped as referring to three main aspects: shared QoL, personal QoL, and family QoL. An optimal measure should be as much comprehensive as possible in terms of areas of shared QoL and dimensions of individual QoL. QoL assessment should not aim at classifying individuals, services or systems, but at providing, within social and health services, a value system that is consistent with those values held by persons with ID/ASD.

14.3 Inclusion

Social inclusion is increasingly recognized as a key outcome for evaluating global mental health programs and interventions [152], particularly for persons with ID [153], who in many parts of the world are still excluded from participating in social environments, have limited opportunities to develop meaningful interpersonal relationships, experience stigma, discrimination, and lack of access to basic services [154, 155].

In the 1950s, families of children with disabilities who did not want to institutionalize their children began to advocate for schooling, day programs, and community homes for their children. Up until about the 1970s or so, children with disabilities could be excluded from school programs and there were few alternatives for adults. Once programs began to start, however, many were segregated ones—for example, special schools, special classrooms just for students with disabilities, and special day programs.

As all students with disabilities started to attend school, and as more and more people with ID started to attend special day programs and live in community homes, advocacy increased for inclusive programs. Schools began to "mainstream" students. Homes started to be in regular neighborhoods, and people with ID began to work in community jobs. These shifts came about due to philosophical changes, advocacy, lawsuits, and legislation, for the right of individuals to live as normal a life as possible and to be as integrated as possible with the larger community.

The outcomes and benefits of such inclusion have been researched over the years, in several different ways. In school programs, the academic and social benefits for students with disabilities have been documented, as well as the benefits for students without disabilities. For adults, the benefits and outcomes of inclusion have been documented in residential, employment, and other community programs. There has also been some research about the benefits to other community members, with a call for additional research in this arena.

14.3.1 Overall Benefits and Current Outcomes

It is important to note that "inclusion" means not just physical presence in community settings, but also includes social inclusion, including interactions and relationships with other community members, and a sense of community membership and belonging. While many people with ID participate in community activities such as shopping and going to restaurants or movies, there is a great need to also increase their inclusive relationships. Physical inclusion has not necessarily translated to improved social inclusion for people with ID. As argued by Cummins and Lau [156], social exposure, as fostered through typical community living or employment settings, is not a suitable proxy for social inclusion in which a person with ID has rich social relationships and is also an active participant in his or her community.

Social inclusion of young adults with ID seems to be strengthened when social interactions are supported and streamlined through structured and guided social activities, which also increase the chances to foster new and stronger social bonds [157].

Relationships affect physical, mental, and emotional health. Extensive studies at the University of Chicago have found that

some health benefits of friendship include less depression, increased resiliency, and increased capacity to recover from adversity and strength to deal with stress [62, 158]. An example of effects on physical health includes a study of US nurses that found that nurses with no close friends were four times as likely to die from cancer as those with many friends [159]. Social relationships also play a vital role as a contributing factor to happiness in the general population [160, 161] and the role of social relationships in happiness has also been established among people with ID [162].

Despite the importance of relationships, many people with ID experience significant challenges in maintaining meaningful friendships and developing close relationships. Solish et al. [163] found that children with an ID participate in significantly fewer social activities with friends and have fewer reciprocal friendships than reported by fellow students who do not have disabilities. Buttimer and Tierney [164] reported that adolescents with ID spend more time without the company of close friends and more time alone than their age peers. Tint and collaborators [165] found that parents of adolescents and young adults with co-occurrence of ID and ASD identified social demands and peer relationships to represent barriers to community participation. Adults with ID consistently identify fewer friendships than those without disabilities. Also, the relationships they have are most often with other people with ID [166–168]. Bigby and Knox [169] surveyed older adults and senior citizens with ID and they reported that their closest relationships are primarily with other individuals with ID, paid support staff, or family members.

> It is important to note that inclusion means not just physical presence in community settings but also acceptance by other community members, interactions and relationships with them, and a sense of community membership and belonging.

14.3.2 Outcomes of Inclusion in School Programs

Benefits of inclusion for students with disabilities have been documented in several areas, including academic achievement, social capacities, and improved behavior [170, 171]. Students with disabilities who are supported in general education classes have been able to achieve grade-level academic standards [172]. Causton-Theoharis et al. [173] demonstrated that instruction in inclusive settings was more effective than self-contained classrooms in the acquisition, maintenance, and generalization of skills for students with disabilities. Instructional practices that are paired with tiered support improve learning for all students, those with and without disabilities [174]. Inclusive education has also been demonstrated to predict post-school success such as employment, postsecondary education, and independent living [175].

Inclusive school programs also benefit all students in the classroom, including those without disabilities. The use of differentiated instruction, which is tailored to fit individual needs, leads to increased student engagement and every student accessing the full curriculum. School inclusion creates a welcoming environment and a positive learning environment for all students [176].

14.3.3 Inclusion for Adults

Several studies have found that individuals with ID in the services system have a paucity of a variety of relationships in their social networks. For example, Verdonschot and his colleagues [177] found that there was an average of 3.1 people in the social network of individuals with ID, and that of those relationships, one of the 3.1 was a staff member. They contrasted the small size of these social networks of people with ID with studies of the general population, which have found an average of up to 125 people

in the social networks of those without disabilities [178]. Emerson and McVilly [166] found that people with ID reported that they typically had only two instances of engaging with a friend in a 4-week period, and none of those were typically with an individual without ID.

The experience of loneliness and feelings of isolation are a common experience for adults with ID. Studies in the United States and Australia have found that up to half of persons with ID experience frequent feelings of loneliness [179, 180], as opposed to reports in the general population of 15–30% experiencing loneliness [167]. Thus, although many individuals with ID are physically integrated into their community and frequently participate in community social activities, they still report few close friends and a lack of meaningful social connections in the community [181].

Many research and demonstration projects have shown that there are simple, pragmatic ways that can be used to increase relationships and a strong sense of belonging and community membership. A summary of many of these programs can be found in Amado, Stancliffe et al. [182] and include interventions in faith communities [183], local neighborhoods [184], recreational programs [185], and retirement networks [186]. Volunteering, for example, is one useful avenue in supporting adults with ID to establish meaningful, socially valued roles in their communities, particularly in the absence of paid employment [187]. Furthermore, participation in spontaneous and voluntary engagements has been reported to foster more valued accounts of social interaction and a better quality of social roles with people without disabilities, compared with the more organized social activities [157].

While interventions such as these have been demonstrated to be successful in increasing social inclusion, they are yet to be brought to a larger, community-wide, or system-wide scale.

14.3.4 Outcomes for Community Members

Mahar et al. [188] among others have noted the power of inclusion to truly impact the lives of people both with and without disabilities. There have been a few studies that have documented the benefits and outcomes of inclusion for those without disabilities. Chadsey and Beyer [162] and Irvine and Lupart [189], among others, have documented the beneficial outcomes to employers and co-workers of having employees with disabilities.

Amado and her colleagues [183, 190] studied two national programs in faith communities that were aimed at increasing the inclusion of people with disabilities. Congregation members reported increased outcomes of acceptance, knowledge, and relationship/emotional ties. Of congregations surveyed that had participated in the Accessible Congregations Campaign, some congregation members in 76% of the congregations achieved enduring relationships with their fellow congregants who had ID [191]. Respondents to surveys about the BeFrienders social ministry program reported that when the BeFrienders ministers became connected to individuals with ID, there were outcomes such as increased awareness of disability-related issues, recognizing gifts in all people, and closer relationships. In a study of membership of people with ID in community groups and associations in four US states [191], community members reported outcomes of increased tolerance/acceptance, learning experiences, recognition of talents and abilities, and friendship.

For older adults with ID, studies in Australia have demonstrated that social inclusion can be increased through recreation and volunteering as people with ID move into retirement. The transition-to-retirement program outlined by Bigby and colleagues [186] combined a gradual transition to retirement with opportunities for inclusion in community groups with other older adults without ID. Establishing mentors in these groups,

in which an older adult without ID assisted an older adult with ID become a part of the group, had positive results, both for the mentors and the mentees with ID [192].

14.3.5 The Measurement of Inclusion Outcomes

The challenge of measuring the outcomes of inclusion efforts is tied to the difficulty in measuring inclusion itself. Both physical and social inclusion have been conceptualized in a number of different ways and have faced the use of varying terminologies, including community "inclusion," "integration," "participation," and "belonging" [183]. The difference between these terms is not always clear, and the definition of each has varied.

Different authors have conceptualized the factors which compose inclusion in multiple ways, all of which would lead to differentiated approaches to measure and determine outcomes. Martin and Cobigo [193] proposed that social inclusion encompasses relationships, leisure, productive activities, accommodation, and informal supports. Hall [194], in her qualitative meta-analysis of the literature on social inclusion, concluded that six core themes defined social inclusion: (a) being accepted as an individual, (b) relationships, (c) involvement in activities, (d) appropriate living accommodations, (e) employment, and (f) supports. Simplican and colleagues [195] developed an ecological model of social inclusion and proposed that it consists of two domains: interpersonal relationships and community participation. They also posited that inclusion can be addressed at the individual, interpersonal, organizational, community, and socio-political levels.

In the absence of a unified definition of inclusion, examination of its outcomes becomes challenging. Most conceptualizations of social inclusion have included both dimensions of Simplican's [195] model: relationships (e.g., [193, 198]) and community participation (e.g., [177, 194, 196]). Community participation has been frequently studied in at least three different community environments: in neighborhoods, in recreational activities, and within faith congregations.

Different studies have utilized different measurement approaches. For example, McConkey [197] studied the degree of social inclusion for individuals with ID living in different settings in Ireland. Using the definition of social inclusion as the combination of the number of social contacts and uses of community resources, his measurements of these two factors found higher levels of social inclusion among adults who lived in semi-independent supported living arrangements, compared with those who lived on larger residential campuses. These results were also found in a similar British study [198].

Besides "inclusion," relationships and friendships are also defined in many different ways. Matheson et al. [199], for example, asked teens with developmental disabilities to identify the elements of friendship, and the teens in their study identified themes of doing activities together, companionship, sharing interests, and proximity. These are all factors that could be measured, and different outcomes evaluated.

Current measurement approaches of different elements of inclusion have varied, with some effort to determine both the quantity and the quality of inclusion. Many researchers have used the frequency of social activities to measure social inclusion, and some measurement instruments, such as the National Core Indicators [200], include questions about who else participated in the activity (e.g., staff, family, housemates, friends, etc.). Other common domains are connectedness and citizenship, with citizenship being the least covered. No single instrument measures all aspects within these most common domains [201].

While such measurements provide a solid foundation reflecting the degree of community participation, they do not effectively measure the degree of inclusion, personal satisfaction

with the social interaction, or other indicators that would indicate the level of the quality of the interaction. Qualitative studies have effectively examined some elements of the different degrees of quality in social inclusion efforts [194], but the quality has been difficult to measure precisely in quantitative studies.

Absent from many attempts to measure inclusion and its outcomes is individual preference and interpretation of one's own social involvement and interactions with others [165]. For example, it is often assumed that a wider social network including both more people and/or more community members without disabilities is preferable than only interactions with other people with ID. However, for a number of reasons, this may not be preferable to all people with ID. It is also often assumed that more social activities result in more satisfaction, but some individuals with ID may prefer a less active social life.

Ideally, the measurement of inclusion and its outcomes would account for individual preferences and self-determination. Measurement could then also gauge the extent to which an individual's vision of inclusion for themselves is fulfilled or not, as the contexts of personal experiences can either facilitate or hinder community involvement [202]. Since one size does not fit all, approaches to measurement and outcomes should include such individualization.

In this debate about how to define and measure the construct of inclusion, there is consensus that it goes beyond simple community presence or participation in activities, and that real inclusion embodies a true presence and relationships in the broader community. However, without a more unified theoretical understanding of the various elements of what entails inclusion, the measurement of the construct and its outcomes remains difficult, perhaps more than almost any other areas of supports and quality of life.

Inclusion has been conceptualized in a number of different ways and using different terminologies, such as community "inclusion," "integration," "participation," and "belonging." Core aspects of inclusion are being accepted as an individual, relationships, involvement in activities, appropriate living accommodations, employment, and support. No single instrument measures all aspects within these most common domains. Absent from many attempts to measure inclusion and its outcomes is individual preference and interpretation of one's own social involvement and interactions with others.

Tip

Future research and practice should address the actual possibilities of choice that are left to persons with ID by the members of their family, professional assistants, and the organization of services. Furthermore, there is a shortage of studies on community members' awareness of, acceptance, and respect for persons with ID.

Key Points

- There is considerable variability of outcomes definition and measurement, also according to numerous factors, such as specific intervention, clinical features, the person's own factors, and context of use;
- Outcome measures can be classified into four groups, clinical, economic, social, and person-centered;
- Outcome measures concerning objective and standardized aspects of health have the advantage of being easy to measure and highly reliable, but fail to capture the person's experience;
- Functioning is intended as the result of a dynamic interaction between individual health conditions, environmental factors, and personal factors. This conceptualization is aligned with an integration of the social and medical models of disability;
- Empowerment refers to the level of choice, influence and control that users of services can exercise over events in their lives;

- Person-centered outcomes focus on holistic and multidimensional aspects of health other than symptoms intensity or pervasiveness of a disease or a disorder;
- Subjective well-being is commonly used to describe a broad category of psychological phenomena that include emotional responses, feeling, and mood, satisfaction with different aspects and situations of life, fulfillment and positive functioning, and global judgments of life satisfaction;
- In persons with ASD, IQ, social relations, adaptive functioning, and physical problems have been reported among the factors with the highest impact on subjective well-being;
- Quality of life measures can be grouped in three main aspects: shared QoL, personal QoL, and family QoL. An optimal measure should be as much comprehensive as possible in terms of areas of shared QoL and dimensions of individual QoL;
- Inclusion means not just physical presence in community settings, but also acceptance by other community members, interactions and relationships with them, and a sense of community membership and belonging;
- Core aspects of inclusion are represented by being accepted as an individual, relationships, involvement in activities, appropriate living accommodations, employment, and supports.

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Quality of Life as an Outcome Measure

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Learning Objectives

This chapter explains what is meant by quality of life, and it explores how quality of life can be used as an outcome measure. Readers will learn that quality of life is a social construct, subject to changing meanings over time and place, and thus, its corresponding measurement is subject to change. They will come to understand that there is no absolute method to measure quality of life and that different approaches to measuring quality of life as an outcome may be taken according to the purpose of applying the measure. Whatever method is used, it is best interpreted the results through the lens of the lived experience of individuals and families.

15.1 Introduction

It has sometimes been said that the ultimate outcome of everything we do in human services, including the overlapping fields of mental health and neurodevelopmental conditions, is to enhance a person's quality of life (QoL). In the broad context of human services, we have set for ourselves quality-related goals and ideals that set the direction for us to follow in providing support, and our fields are developing more and more strategies that help us move in that direction. Ideals such as human equality, full and meaningful participation, respectful inclusion, and full access to opportunities for all dominate the thinking of those of us providing supports and services today because we have increasing evidence that moving toward such goals will result in a greater degree of quality in our lives.

Intellectual disability (ID) and other neurological disability alone can make this challenging, and it is even more challenging when such disability co-occurs with mental health and behavioral problems. For individuals and families who experience such challenges, it is particularly important for services and supports to focus on QoL. They need skills, strategies, and supports as tools to help them along the path toward quality living, and it is for this reason that innumerable ways have been developed, and are continuing to be developed, to help individuals take their steps forward.

An important aspect of stepping forward toward quality living is assessing the impact of our supports and interventions by taking outcome measures. The concept *quality of life* has increasingly gained a central and essential place in person-centered outcome measurement of supports and interventions for people with neurodevelopmental disabilities. QoL has been especially applied to individualized planning, assessing the impact of interventions, and evaluating the effectiveness of services and policies offered by professionals and professional organizations. As QoL is a developing approach to individual and family life, further development is occurring and more will occur in the future. In this chapter, we attempt to provide an overview of what has been developed and usefully employed to date, and to identify areas where further refinements, including in outcome measurement, are needed.

15.2 Understanding Outcomes and Quality of Life as Outcome Measures

Outcomes are the longer-lasting results of what we accomplish in human services. QoL measures of individuals or groups of people with neurodevelopmental disability, when taken and compared at points over time, can record longer-lasting changes in people's lives or aspects of people's lives. As such, they can be used as valid indicators of the effectiveness of the practices, structures, and environments we have in place, and of the actions we have taken – or not taken – to support them.

Outcomes are ongoing and lasting changes that occur as a result of something. In human services, we strive to bring about ongoing and lasting *positive* changes in people's lives. We then assume that such changes improve their quality of life.

15.3 The Importance of Quality of Life as an Outcome Measure

People with neurodevelopmental disabilities are among those for whom assessment of life quality is of particular concern. Compared to those without disability, or with other types of disability, people with neurodevelopmental disabilities have lower rates of employment, tend to be socially isolated, and are generally afforded fewer opportunities for meaningful community participation. To deal with the many challenges of their varied conditions and the barriers to inclusion that exist in our society, they often need specific, and sometimes unique, skills, strategies, and supports to use as tools in moving forward. In the past, we have not always recognized the need for all people with neurodevelopmental disabilities to experience quality in their lives, and thus the goal of achieving QoL is a relatively new one and an especially important one.

Services and supports for people with neurodevelopmental disability have fostered numerous types of helpful policies and organizational structures (the physical buildings, the many types of professional supports, skill development through training and education, etc.). They have, at the same time, recognized that such people, as well as their families and supporters, sometimes have personal needs and characteristics that need to be taken into account and addressed when providing support. The uniqueness of individuals is also an important guide for us in developing QoL strategies, because a great deal of what adds quality to any one person's life involves aspects that are of particular value and importance to each individual. Thus, enhancing quality of life for people with neurodevelopmental disabilities can and should be addressed at the policy, organizational, and personal levels, and quality of life outcomes are probably much better if all three levels are aligned in intent as well as in practice [1–3]. The position taken in this chapter is that professional duty to enhance QoL outcomes for people with neurodevelopmental disability is best achieved when a comprehensive approach – involving an amalgam of practices at the policy, organizational, and individual and family levels – is taken, and when all work together toward the same purpose.

> People with neurodevelopmental disabilities are an important focus for quality of life, because our services and supports to date have not always resulted in them experiencing the "goodness" of life. Quality of life outcome measures can help us identify strategies to help remedy this.

15.4 Two Approaches to Quality of Life Measurement in Health

The concept QoL is, by necessity, a general construct that is widely understood to describe the "goodness" of life. When applied to individuals, the concept describes a balance of the degree of "goodness" in the various aspects of a person's life and in its various circumstances. Several definitions and descriptions of QoL are available that try to capture this balance. One well-known such definition is provided by the World Health Organization (WHO) [4]: "an individual's perception of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards and concerns" (p. 3).

The concept QoL overlaps somewhat with closely related concepts. *Standard of living* is sometimes used as a substitute for QoL, although high or low standard of living does not necessarily mean good or poor QoL, especially for individuals. *Happiness* is another term that is sometimes assumed to be synonymous with QoL, though, again, the two concepts differ because human beings characteristically create quality that relates to their own abilities, interests, and circumstances, and they often create happiness in circumstances that by other standards would not seem to be of high quality. Similarly, *life satisfaction* is not necessarily QoL, as humans often become satisfied in situations of low quality, especially when other needs are being met or when alternatives are not available.

When understanding QoL as a measure, then it is important to base measurement on the "goodness" of life as inherent to the concept, rather than on related concepts that may not fully capture quality.

Within the broad health field, two distinct ways of thinking about measurement using the QoL concept have emerged: generic or "whole-person" QoL and health-related QoL (HR-QoL). Generic QoL evaluates the perceptions of quality by people with respect to the full range of areas that have shown a qualitative value in human life – areas of life and values that are common to the human condition. The focus here is on the quality of a person's whole life as a social human being, regardless of the presence of particular pathologies, disabilities, or existential conditions. To date, the whole life of individuals has been measured by identifying life domains that are important across the human spectrum (e.g., positive social interaction), by identifying the most important indicators of those domains (e.g., friendships are an important indicator of social interaction), and determining specific ways to measure those indicators (e.g., number of close friends, time spent with friends, and ratings of satisfaction with friends might all measure friendships). Although this method of measuring QoL captures a great deal of the quality of most people's lives, it is not a full assessment. For example, time spent with friends may not be "quality" time or even highly enjoyable, and time spent on other activities that are not captured by the indicators of the domains may be much more important to the individual. Similarly, a person may be satisfied with her friends, but times spent with those friends may not be among the most enjoyable aspects of the person's life. On the whole, though, the generic measures that have been developed correlate highly with people's overall assessment of the quality of their lives [5].

HR-QoL is a more specific focus on QoL, relating to patients' or others' perceptions of their health status, and, more specifically, on physical and mental aspects of well-being and functioning (see the Quality of Life Instrument Database of the Mapi Research Trust for a list of available instruments; [6]). HR-QoL measures focus on either overall health and functioning or health and functioning as it relates to a specific disease or condition (e.g., cancer, arthritis, schizophrenia, depression, and many others). HR-QoL instruments typically try to measure both the degree to which symptoms are present and their effects on daily functioning. Thus, although these measures may not capture the broader range of life that generic QoL measures try to capture, they do expand upon the traditional medical approach (treating an identified problem) by considering the effects of identified diseases or conditions and treating them with various interventions [7]. As the use of HR-QoL measures is only a few decades old and is still developing, there is an international agreement that some caution should be exercised in interpreting their scores [8].

One aspect of both these approaches to QoL measurement that merits special attention is that the perception of individuals about their own lives may differ – sometimes quite considerably – from the perception of others [9]. The reason for this is that people with disabilities, like all humans, appear to have personal criteria for understanding the quality in their lives, as well as a tendency to cope with their particular life circumstances and find their "own" degree of life quality.

Over the past 30 years, these two approaches to QoL measurement have led to the development and application of numerous valid tools to measure the QoL of many groups of people, and there are ample QoL outcome measures available within the field of intellectual disabilities [10]. These have developed rapidly over the past few decades, and their use has resulted in a widely held view within the field of neurodevelopmental disabilities that QoL is a key outcome measure for the effectiveness of interventions and services.

Because individuals with disabilities include a wide range of persons, the nature of self-assessment or perception often requires diverse means of assessment. For example, if an individual is severely or profoundly disabled cognitively, or has major deficits in specific areas such as vision or hearing, specific and innovative assessment may be required.

For example, it may be beneficial to assess the individual in their most familiar environment, acclimatizing an individual to a new environment, or developing specific assessment tools to circumvent the disability such as painting and drawing as an interplay between the person with disability and the professional involved in assessment [11].

Tip

If we are most interested in the effects of a specific disability condition or a specific disease, we may choose to use one or more health-related quality of life tools to assess quality of life. However, if we are most interested in understanding people function in general, or in relation to their families or communities, we may choose to use a whole-person approach to assessing quality of life.

15.5 When to Use Shared and Unique Aspects of Human Life as Outcome Measures

Available QoL outcome measurement instruments for groups of people make use of measures that are assumed to be shared by all people and thus to indicate the QoL of all people. If the purpose of QoL outcome measurement is general in nature, such as evaluating the effectiveness of a larger service or policy or providing a "snapshot" of the QoL of a larger group of people, the shared aspects of life may provide sufficient valid information. Typically, these use perceptual measurement methods that combine subjective and objective approaches [12].However, if the purpose of QoL measurement is to provide a basis for individual planning or for evaluating individual interventions, a full assessment should include both shared and unique aspects of the individual's life. This section describes what unique and shared aspects of human life are, and it addresses their utility as outcome measures. The section that follows stresses the importance of relating both types of outcome measures to the lived experience of individuals and families.

15.5.1 Measuring Uniqueness that Enhances Quality of Life

Although all individuals share characteristics that are central to the human condition, they also have varied characteristics and interests that are very meaningful in their own lives, and thus add to their life quality but may mean little or nothing to other people. Personal uniqueness emerges from such factors as genetic makeup, personality, abilities, life experiences, culture, environmental conditions, and even chance (some of life's "quality" moments arise spontaneously and unexpectedly). These factors determine not only how a person's life is experienced, but also how it is interpreted across time and different conditions. Moreover, personal uniqueness is dynamic, changing over time with changing interests, age, and life conditions. Reinders explained that "...the concept of QoL is necessarily open- ended. There is on any account of the matter a dimension of incompleteness in assessments of QoL in the sense that we may discover things about ourselves that in due time will change our judgments. Precisely as a comprehensive concept, QoL must entail an element of the unknown future of our existence" ([13], p. 210). Thus, all of us, as human beings, develop and use over our lifetimes sets of unique interests, values, and capabilities that add to and intersect with those we share with others.

Sometimes, personal uniqueness takes on particularly strong positive meaning in our lives such that they become what Brown, Raphael, and Renwick [5] referred to as dominant and driving forces. When this is the case, they can add tremendously to an individual's QoL. There are innumerable examples, but some commonly recognized ones are enjoying a particular sport, developing a particular talent such as drawing, or spending time with a favored pet. For a very few people, such things can even take up a very large part of

their lives and, if experienced in a highly positive way, form a very large part of the quality within their lives. Some examples include the musician who spends hours every day playing her instrument, an athlete who single-mindedly hones his physical skills, the spiritual follower whose life activities are strongly directed by a set of beliefs and values, a parent who is highly dedicated to the welfare of her children, the family that closely follows the customs of its cultural heritage, or the social activist who strives untiringly for social justice. Whatever their form, dominant and driving forces can add enormously to an individual's QoL in ways that are different from most other people.

Some aspects of QoL can also be negative for individuals. Such things as abuse, poverty, neglect, unsafe conditions, lack of access to food and water, war, and many others can become dominant and driving forces in an individual's life. When this occurs, either temporarily or in an ongoing way, the individual's QoL can be compromised, sometimes severely so.

The methods developed to date in QoL measurement enable us to capture personal uniqueness to some degree, but not fully. For example, an indicator of productivity may be variously interpreted by respondents as work in a paid job, a volunteer position, child-minding, or helping family with chores. What they do not do, except to the extent that there is an opportunity to provide comments or to identify additional factors that add to QoL, is ask specifically about unique personal interests and activities that provide (or detract from) QoL, especially if these are thought of as dominant and driving forces in the person's life. For this reason, when QoL as an outcome is applied to individuals, it is important to gather individually based information about unique interests, values, capabilities, and satisfaction with different areas of life and to assess their importance and value to each person's QoL. As no specific method has been set out in the QoL literature for doing this to date, standard professional assessment and counseling methods should be useful for this purpose.

> Individuals differ widely in the degree to which specific things affect their quality of life. But whether or not these factors are dominant or minor influences in their lives, they need to be taken into account. We do not have standardized methods for accomplishing this, so we have to rely on qualitative assessment methods to estimate outcomes.

15.5.2 Measuring the Quality of Shared Life Domains

Shared QoL refers to characteristics of individual life that are common to, or shared with, other people, such as sociability, productivity, or spirituality. In the field of neurodevelopmental disabilities, the measurement tools that have been validated are all based on these shared characteristics of human life. That is, they identify areas of life (referred to as domains) that groups of people identify as being important to the human condition and to almost every person. Although the various available measurement tools do not all feature the same life domains (see Table 15.1 for a comparison of four tools), there is considerable overlap among them (see [18] for a comprehensive list of available tools) and overall scores correlate quite highly (e.g., [19]). For individuals, well-researched and validated domains include physical, psychological, and spiritual well-being, personal development, social and community inclusion, and opportunities to achieve and be productive (e.g., [20]). Civic involvement and rights are also sometimes part of a measurement system for individuals and families, although it might be argued that these are determinants, rather than outcomes, of QoL. For families, well-researched QOL domains are family interaction, parenting, emotional well-being, personal development, physical well-being, financial well-being, community involvement, and disability-related supports [21, 22].

Shared QoL is relatively easy to assess, even in persons with severe cognitive impairment. The domains are most often not measured directly, but rather indirectly through

Table 15.1 Comparison of domains in four QoL tools

	WHOQOL-BREF and disabilities module [14]	Personal well-being Index – intellectual disabilities [15, 16])	Quality of life questionnaire [17]	Quality of life instrument package [5]
Number of QoL areas/ domains	4 + disability module	7	8	9
Total number of items	39	8	40	54
List of QoL areas/domains	Physical Psychological Social relationships Personal environment + Disability	Standard of living Health Life achievement Personal relationships Personal safety Community-connectedness Future security Spirituality-religion (optional)	Physical Well-being Emotional Well-being Interpersonal relationships Material Well-being Personal development Self-determination Social inclusion Rights	Physical being Psychological being Spiritual being Physical belonging Social belonging Community belonging Practical becoming Leisure becoming Growth becoming

specially developed and validated indicators. These are typically measured on a 3–6 point Likert-type scale [23] for each of the tool's measurement indicators, sometimes for more than one measurement concept such as importance, satisfaction, opportunities, choice, attainment, initiative, and stability [24].

In scoring these dimensions, self-report by people with disabilities should be central, and any means should be mobilized to enable such people to express their own views. In all cases, information on the background of the person, descriptions of the person's current context and living/working environments, and the perceptions of close caregivers constitute additional sources of information that are potentially valuable to add to self-perceptions. Such assessment may even be used effectively with people with severe cognitive disabilities, as they express their inner states through consistent behavioral repertoires [25].

QoL scores are derived from measuring the indicators of life domains specified in the QoL tool. The resultant raw scores can be transformed into standard scores, then analyzed and reported as QOL area/domain scores, or aggregated into a total QOL Score [26]. Since these domains have been validated as aspects of human life that are important to all people with some leeway for cultural influence [27], scores from these tools are considered to be credible measures of shared QoL values.

Several tools that feature shared domains have been developed to assess individual QoL in persons with ID and low functioning-autism spectrum disorder (LF-ASD). Four well-known scales are described briefly below, and their main features are compared in Table 15.1.

A commonly used QoL measure for people with disabilities is the *WHOQOL-DIS*, developed by the World Health Organisation (WHO). The *WHOQOL-100* is a QoL measure of 100 items for the general population, and it has a brief form of 26 items (*WHOQOL-BREF*). The *WHOQOL-DIS* is the brief form with an additional 13-item section specifically addressing disability. The *WHOQOL-100* was designed with a hierarchical structure that includes overall QoL, six life domains, and then 24 facets within the domains with four specific items to measure

each facet. The *WHOQOL-BREF* has a similar hierarchical organization, except that each facet is represented by a single item. The widespread international use of the *WHOQOL* provides for cross-cultural validity for QoL assessment across the adult lifespan and a range of physical, mental, and neurodevelopmental disabilities [28].

The *Personal Wellbeing Index* (*PWI*), developed by Robert Cummins and emerging from his previous *Comprehensive Quality of Life Scale* [16, 29] at Deakin University in Australia, has a useful ID version (*PWI-ID*). The seven domains and one optional domain, each represented by the same single item on the *PWI* (adult version) and the *PWI-ID,* are shown in ◘ Table 15.1. This scale was carefully constructed by a large cohort of international researchers, and takes a short amount of time to administer. Special features of the *PWI-ID* include a detailed set of pre-tests to help ensure the validity of responses, simplified wording of the *PWI* items, and, to promote comprehension, the possibility to use 2-, 3-, or 5-point rating scales in place of the 0–10 scale used in the *PWI* or to use a set of happy and sad faces [9]. The limited number of items on the *PWI-ID* suggests that its principal utility might be as a survey and program evaluation tool rather than a tool for individual assessment and support planning, but its robust psychometric properties ensure its overall credibility. Another consideration in using the *PWI-ID* is that it measures subjective wellbeing, a person's own perceptions of his or her life. The other tools in ◘ Table 15.1, as well as Deiner's well-regarded *Satisfaction with Life Scale* [30] and Heal's *Life Satisfaction Scale* [31] also use self-perception as an information source for measurement, although perhaps not quite as explicitly. But it might be kept in mind that these tools do not use objective observation of others as primary information sources for measurement.

One of the most internationally used QoL tools for ID populations evolved from the Schalock and Keith [32] *Quality of Life Questionnaire* (now known as *QOL-Q*), developed in the United States. The *QOL-Q*'s eight domains, as listed in ◘ Table 15.1, were expanded from the original four domains (satisfaction, competence/productivity, empowerment/independence, and social belonging) but, like the original, contain 40 items. The eight domains, listed in parentheses here, group within three main factors: (1) Independence (personal development and self-determination), (2) social participation (interpersonal relationships, social inclusion, and rights), and (3) well-being (emotional well-being, physical well-being, and material well-being). Like the original questionnaire, the *QOL-Q* has been extensively tested in a series of population and cross-cultural studies. Core indicators operationally define each QoL domain, and indicator items represent specific items that are used to measure the person's perception ("self-report") or an objective perspective based on the person's life experiences and circumstances ("direct observation"). The *QOL-Q* has been widely applied for a variety of purposes, especially in Schalock's far-reaching work in policy and organizational evaluation, and as such it is useful as a focus point for comparisons of QoL of populations across time and place.

A fourth QoL assessment tool is the *Quality of Life Instrument Package* (QoL-IP; I. [5]), developed in Canada. The QoL-IP features nine domains, organized into three macro-areas: being, belonging, and becoming (as listed in ◘ Table 15.1), each of which features three domains. QOL domain indicators are rated from three perspectives: the person being assessed, others who know that person well, and a trained assessor. Each indicator is assessed in four dimensions: (1) importance, (2) satisfaction, (3) opportunities that the person had/has to develop importance and to perceive satisfaction; and (4) choices that the person could/can make in the same areas. Importance and satisfaction interweave by mathematical formula to produce a basic QoL score, with satisfaction being weighted by importance. This idea had been put forward by a number of scholars in the 1990s, including Becker et al. [33], but was described by Bertelli et al. [34] this way: a thing that is highly important and gives high satisfaction has a considerable positive impact on a person's QoL, while a thing that does not interest a person or is not valued will never add

satisfaction or quality to the person's life. Decision-making from choice and the availability of opportunities from which decisions can be made act as moderators to basic QoL scores.

Uses of QoL Scores Based on Shared Life Domains QoL scores based on shared life domains can be appropriately applied to evaluation for groups of people, for services, and for supports and policies that address the needs of large numbers of individuals. They can be used to keep track of progress made by policies and programs over time, where QoL is considered an excellent indicator of progress. In this way, they can be used to evaluate the effectiveness of an organization or system's services and supports over time [35, 36], and thus to assess organizational or system quality improvement [37, 38]. Table 15.2 provides an integrated approach to quality improvement based on the work of Baker et al. [39], Gomez et al. [40], Lee [41], Reinders and Schalock [42], Schalock and Verdugo [35], Schippers et al. [3], and van Loon et al. [43].

For individuals and families, QOL outcome measures of shared life domains can and should be used as one good source of data for individual and family assessment and developing person-centered and family-centered plans. An area of life that adds quality is one in which a domain is valued in the person's life, and the domain QOL score is high. These areas should be actively maintained, as they contribute quality to the person's life. An area of life that detracts from quality is one in which something is valued in the person's life, but it is scored low on achieving it or satisfaction with it. These areas require attention to eliminate the causes of the problems or to develop strategies to improve them. Individual indicators, too, can be used in a similar way.

Table 15.2 Quality improvement strategies

Focus	Examples
Individual	Determine QOL-related personal goals Assess individual support needs across QOL domains Use an outcomes focused planning format that aligns personal goals, support needs, and QOL outcome categories Use an ISP format that is user friendly and one developed and implemented by a support team composed of the service recipient, a family member or advocate, direct support staff, relevant professionals, and a supports coordinator Assess QOL domain indicators
Organization	Provide improved access to technology that allows for real time planning/reporting Provide training programs focusing on QOL and its multidimensionality Adjust job descriptions and hiring practices to reflect the QOL framework Evaluate staff as to whether necessary support is given (rather than activities were completed) Change to person-centered planning process Develop policies that promote the use of natural /community supports Base strategic planning efforts on QOL as the aligning construct
System	Develop and launch a web portal for service providers to share ideas that promote QOL for individuals served Train systems-level staff on the importance of QOL and its difference from goal attainment paradigms Provide workshops and conferences that promote a deeper understanding of the QOL concept and its application Conduct strategic planning with QOL as the aligning construct and the strategic driver for organization and system transformation Move toward using QOL outcomes as key performance indicators Fund pilot projects that focus on the key role of the QOL concept in agency change Modify contractual arrangements to reflect QOL assessment and continuous quality improvement

For example, if spending time with friends is a specific indicator of social well-being and this is important to the person, a low QOL rating for this indicator can strongly suggest action should be taken to improve the quantity and quality of time spent with friends.

QoL scores for individuals or families do need to be used judiciously. Often, other factors need to be considered alongside such scores for their use to be effective. For example, at the systems level, a relatively high score on QoL indicators may suggest that policy changes such as increases to income assistance and sustainable funding for communities are unnecessary when in fact such changes might significantly improve the "goodness" of the lives of individuals. At the personal level, a low outcome score might suggest a needed intervention, but that intervention may not be desired by the individual or helpful if it is implemented.

Continuing Work Needed in QoL Measurement Based on Shared Aspects of Life There is still work to be done in QoL measurement, and some examples are provided here:

- Most instruments set out QoL indicators under a number of logical domains, but it is not clear whether the domains identified to date accurately capture QoL as a whole, and the extent to which these domains make unique contributions to overall QoL. For example, the factor scores on importance did not fit clearly into the core domains proposed by current available tools to describe the set of cross-cultural factors that can impact personal well-being [34].
- We are not certain that QoL tools actually measure the whole concept, or whether they simply measure their domains [13].
- Our current tools do not show exactly how people experience QoL. We might assume, however, that people do not typically experience their QoL as divided among separate domains, but rather evaluate how their experiences within these various domains interact with one another and to their whole lives.
- The relationship between QoL measures and those of other closely related concepts, such as self-determination, personal development, and the possibility of choice, needs further exploration [44].
- The distinction between indicators of QoL and factors that cause QoL is not clear in the available literature [2].
- Work might be carried out to organize QoL indicator variables for life as a whole in a hierarchical way, a process that would help us to better define a minimum set of domains with equal variance [2].
- Work needs to be done around including people with disability more meaningfully in designing projects related to QoL measurement and application.
- In numerous countries of the world, there is a superficial understanding of the nature and causes of neurodevelopmental disabilities, autism spectrum disorder, and other disability conditions. Such limited understanding may influence the treatment of people with disabilities and, in turn, influence perceptions of individual and family QoL [45, 46].

Tip

Quality of life measures based on shared aspects of life may be more reliably assessed when more than one measurement tool is used.

Tip

Scores from quality of life indicators that are based on shared aspects of life provide a solid information base for assessing outcomes – the ongoing and lasting effects of a program or an intervention. This should be amended by considering unique aspects of life and the lived experience of individuals and families.

15.6 Relating QOL Measurement to the Lived Experience of Individuals and Families

The *lived experience* of individuals and families is a term used for the reality of people's lives, and understanding lived experience is considered to provide the most important understanding of people with neurodevelopmental disabilities and family life that includes disability [47]. It follows that, for individuals or families with disability, QOL outcome measures based on both unique and shared aspects of life need to be applied within the context of their lived experience.

Lived experience sometimes modifies, or even reverses, the strength of specific QoL outcome measures. For example, some aspects of life may score low on quality, but the person is not unhappy about that or does not necessarily want change. They may also be addressing some positive function that is "working" in the larger context of the person's lived experience. Conversely, some aspects of life may score high on quality, but they are not of particular interest or value to the person, change may be desired for other reasons, or they may contribute little to positive functions. Such considerations stress the importance of the lived experience context, and they suggest that it may not be fruitful to spend time and resources on some QoL improvement interventions suggested by the QoL scores alone.

But real life experience complicates the interpretation of QoL scores even more than this. Individuals and families have a variety of coping strategies that they may or may not use, to tolerate life adversities. Some stressful aspects of life may be tolerated even without such strategies when they are balanced by others that are enjoyable. Some unpleasant aspects of life are temporary and will go away on their own, so it is often a matter of coping temporarily then waiting them out. Some people enjoy a little challenge or chaos in their lives and others do not. There are many other factors relating to lived experience that may affect how QoL outcome measures can be applied effectively. The "art" of helping a person or a family enhance the quality of life involves exploring the various factors that need to be addressed within the context of their lived experience and to determine the most effective ways to address them.

> Since quality of life is experienced by people in the context of their lived experience, quality of life outcome measures need to be interpreted through this lens.

15.6.1 Individual Lived Experience

The unique and shared QoL measures that we might assume affect an individual's QoL are not always reflected in how an individual perceives his or her own life or lived experience. The early research on individual QoL, reported in the 1990s, strongly hinted that this might be the case – QoL scores from individuals with ID and those from trained assessors, support workers, and family members did not correlate well, suggesting a difference in perspective related to differences in lived experience (see [5], for example). The questions that emerged were: how do people with ID perceive their own lives and why do they perceive their lives in those ways?

The work of Cummins and colleagues in Australia provides an intriguing hypothesis. Based on extensive research and analysis, Cummins noted that almost all adults, including those with ID, no matter what their life circumstances may be, rate their own lives as positive but not perfect, typically between 70 and 80 on a scale of 0–100. From this base, Cummins developed his *theory of subjective well-being homeostasis*, meaning that human beings have a genetically generated tendency to look for happiness, satisfaction, and meaning in their lives despite the circumstances, and that their levels of happiness are quite stable across time and across geographic areas [48, 49]. Life satisfaction can go down in difficult times or it can go up in good times – and it fluctuates with temporary emotional highs and lows – but the tendency is to move back toward a stable point. A few years later, Cummins coined the term *set-point of happiness,* which is the point on the scale that

ratings of life satisfaction tend to fluctuate around and to which mood returns under normal circumstances [50, 51]. What this suggests for QoL outcome measures is that people with neurodevelopmental disabilities, like all people, are highly likely to be subject to the homeostatic effect and rate their own lives around their set-points of happiness as their life circumstances change. Through extensive research from his *Australian Unity Wellbeing Index,* Cummins and colleagues also reported that three factors particularly influenced people's sense of well-being, which he called the "golden triangle of happiness": satisfaction with income, relationships, and life purpose [52]. These three factors, Cummins maintained, are especially strong in protecting us from ongoing negative mood and moving us homeostatically back to our set-points of happiness.

This hypothesis suggests an amendment to people perceiving their own lives within the context of their own lived experience. It suggests that people both with and without neurodevelopmental disabilities may have a genetically based tendency to return from both high and low emotional levels to their set-points of happiness, using both inherent and learned strategies, and, moreover, that this is a preferred process for humans. What this might mean for QoL is that a measure of quality or the "goodness" of life might best be described as the person's lived experience that has some enjoyable high emotional states, a few low emotional states, and very effective sets of skills both to recognize the effects of fluctuating emotional experiences and to return to the person's set-point in a timely way [53]. Those whose lives cannot be described this way might be thought to reflect lower QoL, especially where there is incapacity to return easily from negative experience to the set-point or incapacity to return at all and thus be mired in negative emotions and experiences.

Another interesting way to look at individual lived experience and QoL is by examining the "disability paradox" coined by Albrecht and Devlieger [54]. This refers to the fact that, when recording subjective well-being, it is often noted that QoL scores of people with intellectual or other disabilities do not differ significantly from those of the general population. This so-called paradox has been explained variously as a psychological mechanism of adaptation, coping, resilience, research methodological bias, or even poor conceptualization of persons with ID. But another explanation is that people with ID perceive their own lives through the lens of their own lived experience since this is the only lens they have, and their perception of their lives is of a higher quality than the perception of others. Simply put, life is experienced by all individuals through their unique sets of perceptions, and because of this subjective well-being may be an essential pathway to understanding "true" personal QoL.

An additional view of lived experience for people with ID is that personal perceptions may differ from other people's perceptions because those with neurodevelopmental disabilities view situations differently. As mentioned briefly in ▶ Sect. 15.3 above, this often results in non-significant correlations between ratings by people with ID and by proxies. For example, people with ID generally have clear views on what their choices are and what they want to do, while parents appear to respond to the barriers that they believe will inhibit the individual from attaining their wishes. It is therefore important to be aware of the different perspectives [55]. It is appropriate at this point to recognize that what are often referred to as subjective responses are in fact objective in the sense that they are heard and recorded. It is a fact that the statements were made by the two parties. The subjective component is the interpretations that are made of such data [1].

These perspectives on lived experience and QoL shed new light on scores derived from QoL tools. High or low scores (overall scores, domain scores, or indicator scores) may indicate seemingly clear courses of action that may sometimes need to be amended or abandoned when considered in the context of live experience. A challenge for disability professionals is that people with intellectual and other disabilities may not see themselves as needing support as much as others do, and they may not want

the types of "improvements" that others assume will benefit them as a result of such support. Another issue that emerged from this is that QoL scores for individuals and families with disability would not be expected to be atypically low, and when they are, as Cummins [48] has pointed out, there is real reason for concern and assistance might well be needed.

> Individuals with disabilities very often rate the quality of their own lives differently than others rate their lives. They are probably looking at their own lives through the lens of their abilities and their own lived experience. Moreover, people with disabilities, like all people, may have a natural tendency to "make good" of their particular life situations.

15.6.2 Family Lived Experience

The overriding issue in considering the lived experience of families is that, when parents and siblings find it difficult to deal with the challenges they face, it is not only the child with a disability who is further restricted, but also every member of the family. Families differ, sometimes quite markedly, in their willingness and ability to deal with disability, seek and implement solutions, work with those who can give support, manage their families, and cope with the ever-changing nature of family life. Thus, there is no "typical" or "normal" lived experience for families that have a member with a disability. Yet, like individuals, they have things in common, including set-points of happiness to which they have a tendency to return. Still, it is clear that certain disabilities and mental health problems, especially when difficult behavior is involved, have noticeable impact on the QoL of families (e.g., [56]).

Since the year 2000, family quality of life (FQOL) has been assessed in many countries of the world and in many languages using two main FQOL instruments: the *Family Quality of Life Survey, 2006* [57], and the *Beach Center Family Quality of Life Scale* [58]. These instruments produce FQOL outcome measures similar to those produced from individual QoL measurement. FQOL outcome measures are derived from indicators in identified family life domains such as parenting, family relationships, finances, support from services, support from others, leisure time, and others [7, 28].

Although the more complex family lived experience needs to be understood and taken into account when supporting families, the results from the many studies in FQOL around the world provide a basis for understanding where to begin. In a comparative analysis of 8 and 19 research studies, respectively, I. Brown [59, 60] noted that study respondents in every country rated satisfaction with supports and services as quite low, and as not nearly as important to their QoL as family relationships and values. Other analyses have reported that FQOL scores are lower when family members with ID have behavior disturbances [44], that health and financial challenges compromise QoL for many families, and that support from friends, relatives, and neighbors is low in every country studied [60]. Lack of respite means that many families are unable to take vacations or parents are unable to spend time together. A parent – usually the mother – frequently has to interrupt or stop a career to provide care for a child with a disability. Such results and the substantial general literature about families suggest that families face some major challenges, and that many of these challenges appear to cause exclusion for family members.

In many ways, this is sobering information for professionals supporting families that include a son or daughter with a neurodevelopmental disability. It tells us that much of the real quality in families' lived experience may be derived from things other than the things that those of us in the service industry are addressing. This poses a serious challenge to our services, because a great many families who have children with disabilities do need support, solving practical family and social issues as well as finding adequate intervention for mental health challenges. When additional challenges arise in the broader society (such as the COVID-19 pandemic that began in

2020), there is a considerable increase in family stresses that often result in mental health challenges. At times, FQOL measures may capture this, such as lower ratings of financial, social, or community involvement indicators, but their impact may not be fully reflected by outcome measures alone.

What steps, then, can we take to understand better families' actual lived experience, and to support the aspects of that experience that add to their measured QoL outcomes? Four key steps in this journey are briefly described below.

- *Base our efforts on practical principles embedded in the FQOL approach*. These principles have been described in detail by several authors and are expanded in R. Brown et al. [61]. General principles include, among others: dignity of disability, ethically based policy and practice, personal and professional values, duty of care, risk and safety, normalization, and exclusion/inclusion. Individual and group principles include: resilience, perception, self-image, empowerment, personal control, and intra- and inter-personal variability. Other principles are: holism, lifespan issues, opportunities and choices, and imaging the future [62]. Critical to all these principles is the perception of the individuals involved.
- *Focus on family functioning.* Family QoL research clearly shows that family relationships are very important to families in all countries yet, at times, having a member with an ID is associated with challenges in family functioning. Suggestions for professional services include developing education and support for families around effective parenting styles and family unity strategies. This education and support seems to be particularly effective when families are given opportunities to share information and expertise with each other in an organized peer support model [44]. The need to have opportunities to simply talk about issues that bother a person appears in the qualitative data from virtually all FQOL studies.

Understanding and supporting the role of siblings in a family that includes a child with a disability may also be important in supporting the on-going QoL of the entire family [63]. These supports might include sibling support groups, and the meaningful inclusion of siblings in decision making and planning. Beyond siblings, it is important to understand the function of all family members, no matter how minor that might be.

- *Design family-centered supports.* As mentioned, a lack of satisfaction with service support appears as a challenge in all countries where FQOL research has been carried out [44]. Moreover, families have indicated high levels of stress related to interactions with formal disability-related staff and professionals [64]. Some common concerns that have been voiced are a lack of information about available services, difficulties in trust and communication with support providers, and a shortage of provider focus on family functioning and family relationships. An important practical suggestion for disability support workers is to develop supports based on family-centered approaches with the family controlling and contributing to their own support, rather than having plans and programs generated by providers. This approach emphasizes the rights and capabilities of families to direct and participate in their own social welfare experience and may result in more positive FQOL outcomes [65]. In addition, professionals who support families may see benefit from working in partnership with families on an equal basis [66]. Suggestions for providers wishing to build trustworthy, responsive family services include [67]:
  1. Focusing upon improved communication with families, such as sharing information and listening to feedback from families. To have available personnel to undertake such activity in the early stages of increased stress may have important implications for reducing complete breakdown later in the family's life.

2. Providing high levels of professional competence and ensuring disability professionals who are in contact with families are well-trained experts with relevant knowledge and experience. Professionals working with families should be trained in FQOL principles, be able to provide information about disability service and funding options, and be able to assist families in navigating the service landscape.
3. Providing an environment of respect, commitment, equality, and trust. Disability professionals may find a focus on building strong relationships with families beneficial for all involved.

- *Pursue inclusion of families in physical and social communities.* It has been noted that there is often a lack of support from informal sources such as friends, extended family, and neighbors [68, 69]. One way that providers might assist families in making and maintaining social connections and building social support is through family support groups. R. Brown et al. [70] recognized the need for such groups but noted that they need to include trustworthy, safe support for the child with disability and siblings so that family members can relax and have sufficient time to network with other family members. Faragher et al. [71] suggested that formal services may assist families in family support groups by suggesting ways to access the supports they need. For their parts, family members indicated that sharing meetings with others is highly supportive across the lifespan. The initial meetings need to be led by a skilled mediator, though family members will often run later meetings themselves.

Research has also suggested that service providers might become community connectors rather than simply caregivers, empowering families to make the social connections needed to find formal and informal supports [72]. These connections in the community might assist families in finding pathways to information and collective action for service and policy improvement. The enhancement of social connections can also enable the growth of relationships leading to the emotional and practical support that are so important for ensuring the health of the family ([69]; see [73] for further practical approaches).

Tip

Families that include a member with disability often have difficult lived experience. Quality of life outcome measures, especially for people with disabilities living with their families, need to include family quality of life outcome assessment – which needs to be interpreted through the families' lived experience.

15.7 Putting It All Together

Valid QoL outcome measures have been developed and reported for both individual and family QoL. These measures provide evaluation and assessment data for helping individuals and families move toward enhanced QoL, for improving the quality of disability-related services, and for policy and practice accountability. They also provide a strong basis upon which new or amended policy and practice models can be developed. A robust body of research on the QoL of individuals and of families is currently available in the published literature, a body of work that has established the utility of applying QOL outcome measures that are common to individuals and families. The work of Schalock and his numerous colleagues have steadily moved the application of QOL outcome measures to policy and practice forward over the past several years, and this is proving to be very helpful in shaping policies and services that focus on quality outputs and outcomes.

Although future work will offer advances, the considerable knowledge that has been generated to date on QoL as an outcome measure allows us to understand that there are various options for use with people who have neurodevelopmental disabilities or autism spectrum

Table 15.3 Options in using QoL as an outcome measure for people with ID or ASD

Issue	Options
Purpose of using QoL outcome measure	Individual planning Evaluating individual interventions Program planning Program evaluation Policy evaluation
Focus of QoL measurement	Shared QoL Personal QoL Family QoL
Content of QoL measurement tools	Domains/areas (objective assessment) Dimensions (personal appraisal) Indicators (individual objective assessment)
Person whose QoL is being measured	Person with ID (mild to profound) Person with ASD (any degree of support need)
Information source	Individual with ID/ASD Proxies (persons who know the individual well) External assessors
Understanding QoL data and scores	Overall, domain, and indicator scores Subjective and objective data Amendments based on lived experience of individuals and families

Expanded from Bertelli et al. [34]

disorder. Table 15.3 lists the options that have been described in this chapter as they pertain to: the purpose of using QoL as an outcome measure, the focus of the QoL measurement, the content of the measurement tools, the person whose QoL is being measured, the information source, and understanding the QoL data and scores. Together, these options illustrate that a variety of QoL outcome measures may be used in a variety of ways for a variety of purposes.

15.8 Moving Ahead

The development of QoL, both from a conceptual and from a measurement point of view, is not complete. The term *quality of life* itself is a social construct and, as such, its meaning changes over time. QoL conceptualization and outcome measurement will both have to adapt to such changes in meaning. Moreover, we cannot assume that we fully understand the complexity of QoL at the present time, despite its simple surface meaning illustrated by the question, "How good is your life for you?" [5]. But the various combinations of factors that combine to make up a "good" life or a "quality" life at any one time, for any one person or family, is a complexity that we have perhaps only begun to describe. Finally, the task of enhancing QoL for individuals and families by drawing together the outcomes of policy, practice, and people's lived experience – so that they work together toward a common goal – is one that is somewhat daunting but nevertheless an exciting challenge ahead.

We have come a long way since QoL first became a widely recognized concept in the late 1980s. It is now understood to be the overall goal of almost everything we do in the field of neurodevelopmental disability, and it is a commonly stated policy and practice goal in almost every country of the world. As the concept of QoL continues to evolve in the future, its outcome measures will also evolve and, as they do, they show real promise of being useful tools for moving toward our goals.

Key Points

- Quality of life is a social construct that is used to encompass the general notion of the "goodness" of life.
- Outcome measures are measures of ongoing and lasting changes. Quality of life as outcome measures use the scores derived from quality of life indicators

to assess ongoing and lasting changes to the "goodness" of people's lives.

- Two general approaches to attaining scores from quality of life indicators in the human services are: (1) a focus on specific conditions or diseases (health-related quality of life) and (2) a focus on the person's whole life in relation to the environment (whole-person quality of life).
- Numerous tools are available to measure health-related quality of life, and several valid tools have been developed to measure whole-person quality of life for people with intellectual and other neurodevelopmental disabilities.
- The available whole-person tools provide quality of life scores for domains of life that are deemed to be common to, and important to, almost all humans. These quality of life scores can be used as the primary assessment information in determining outcomes (ongoing and lasting changes).
- To make a final determination of outcomes, the primary assessment information obtained from the scores of quality of life tools needs to be amended by: (1) considering life factors that are unique and important to the individual and (2) interpreting the information through the lens of the lived experience of individuals with disabilities and families that have a member with disability.

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Autism Spectrum Disorder

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Learning Objectives

The primary objective of this chapter is to provide a broad overview of ASD in terms of its worldwide prevalence and etiopathogenesis including genetic, epigenetic, and environmental influences. The chapter also provides a review of the definition leading to the current conceptualization of ASD, including the evolving changes in diagnostic criteria, its clinical features, and diagnostic assessment measures. The question of comorbidity and treatment considerations, including both pharmaceutical and nonpharmaceutical approaches, is also discussed. The emphasis on person-centered, evidence-based, multidisciplinary, and collaborative inclusionary framework on ASD across health, education, and social sectors is underscored.

16.1 Introduction

Autism spectrum disorder (ASD) represents a group of lifelong neurodevelopmental disorders emerging during early childhood and interfering with a person's ability to socially relate to and interact with others. Over the past 30 years, there has been an unprecedented increase in the prevalence of ASD worldwide with impressive strides made in increasing awareness of the condition and developing innovative ways to improve the lives of children, young people and adults with ASD. In particular, since 2007, with the passage of the United Nations General Assembly resolution [1] creating the World Awareness Day, April 2 every year is now recognized by member states to not only raise responsiveness but also report on tangible outcomes of interventions as part of the implementation of the UN Sustainable Development Goals (SDGs) [2].

There are still large gaps in the dissemination of the evidence base on ASD, and in particular, in terms of implementation of effective treatment across the lifespan. There is a need to provide persons with ASD and their families with adequate supports, and especially to ensure that children and adolescents with ASD have access to public education and services. There is also a need to recognize persons with ASD as participants in the social fabric of countries worldwide. Although this chapter emphasizes the cutting-edge science in terms of etiopathogenesis, psychiatric comorbidity, assessments, and treatment, it is important to acknowledge at the outset that ASD cannot be effectively addressed by the health sector alone and national policies require collaboration across health, education, and social sectors [3]. To accomplish the multidisciplinary goals, policymakers worldwide must:

- Encourage early detection and diagnosis of ASD, allowing children, adolescents, as well as adults to access services needed as early as possible
- Ensure that children, adolescents, as well as adults with ASD have access to evidence-based therapies by providing additional training to parents, teachers, and clinicians around the most effective interventions
- Provide those with ASD and related neurodevelopmental conditions access to public education, vocational training, and assisted employment with the goal of inclusion
- Encourage participation in high-quality surveillance and research

16.2 Prevalence

The prevalence of autism doubled from around 0.5/1000 during 1960–1970s to 1/1000 by 1980s when DSM-III definition was introduced, doubling again to 2/1000 by early twenty-first century [4]. The current prevalence of ASD is accepted to be 1% worldwide [5–7]. The prevalence has been noted to be as high as 1/38 among South Korean children in a population-based study [8], a figure that may be an overestimate [9]. The Center for Disease Control and Prevention (CDC) Autism and Developmental Disabilities Monitoring (ADDM) Network in the United States has estimated ASD in 8-year-old children to be 1/54 in 2020's analysis [10] and 1/44 in 2021's analysis [11]. This surveillance figure is an average with variation by race/ethnicity as well as state of residence of the children with notable disparities in the prevalence

depending on ascertainment from health and educational sources [12].

ASD is now universally regarded as present in all geographical regions of the world, among people of all ethnic backgrounds in both low- and middle-income country settings. A Swedish population-based study has shown that risk of offspring with autism is consistent across all parental socioeconomic strata [13]. ASD is reported to be 4 times more common in males than in females [14, 15]. Although the terms high-functioning autism (HFA) and Asperger disorder are not formally used, the M:F ratio among children and adolescents without intellectual disabilities (intellectual developmental disorders) ranges from 6:1 [16] to 10:1 [17]. Some studies have emphasized that lower representation of prevalence among females may reflect lower probability of the condition being identified in females altogether [18], or misclassified as having affective, anxiety, personality, or eating disorders [19]. A recent systematic review approached this issue by analyzing 54 studies including 13,784,284 participants, with findings pointing to a M: F ratio closer to 3: 1 [20]. It has also been noted that a "female protective effect", that is, females requiring greater familial loading to manifest the ASD phenotype in multiplex families, cannot explain the sex imbalance in ASD prevalence and there is no supportive evidence that a single common locus on chromosome X might mediate such a female protective effect [21]. Women are also reported to have much higher efficacy in compensation mechanisms of social difficulties than men, aptly called "camouflaging" [22]. This higher discrepancy between externally observable behavior and inner experience of socialisation has been found to be independent from the intellectual quotient [23], implying that it may exist even in persons with borderline intellectual functioning and mild intellectual disability.

ASD is now universally regarded as present in all geographical regions of the world, among people of all ethnic and racial backgrounds living in both low- and middle-income country settings.

16.3 Etiopathogenesis

16.3.1 Genetics

Genetics provides major findings underlying ASD as well as endorsing great interindividual variability [24, 25]. The opportunities offered by new technologies, including chromosomal microarray (CGH) to Next Generation Sequencing (NGS), have overcome the limitations of linkage and candidate gene association studies, expanding our knowledge of its genetics. ASD is now viewed as the leading neurodevelopmental disorder with the highest genetic loading, initially based on the results of twin studies in the UK and Scandinavian countries that provided heritability estimates greater than 0.90, with significant differences in concordance rates between monozygotic and dizygotic twins (73–95% vs 0–10%, respectively). In addition, the mean prevalence of ASD in the general population of 0.01 is significantly lower than the recurrence risk recorded in families with an ASD sibling with the prevalence of second ASD sibling ranging from 0.15 to 0.25 in males and 0.05 to 0.15 in females [26]. In addition, the presence of "autism spectrum" traits among first-degree relatives of individuals with ASD underscores important genetic contributions and points toward its complex nonlinearity in most cases [27]. Recent twin studies have provided lower heritability estimates, ranging from 0.37 to 0.60 [28, 29], reflecting differences in methodological issues [30], differences depending on the presence of de novo mutations and copy-number variants (CNVs), with estimates occurring in 15–25% of ASD [31]. There are putative epigenetic influences triggered by environmental factors on the fetus and/or on parental gametes (see Par. 16.3.2). Nonetheless, genetic contributions remain substantial [30] that represent a valuable path to reconstruct the pathophysiology of ASD. Based on the underlying genetics, therefore, ASD can be broadly distinguished in a variety of forms, namely syndromic vs idiopathic, de novo vs inherited, monogenic vs oligogenic vs polygenic [32].

1. *Syndromic Autisms* – Genetic syndromes significantly associated with ASD are listed in ◘ Table 16.1 [24, 34]. Syndromic patients typically display facial and physical dysmorphisms, malformations, intellectual disability of variable degree, and epilepsy of varying severity. Neurological signs and symptoms are more frequent than in idiopathic forms. The M: F ratio is collectively close to 1. Anomalies of body and cranial growth yield microsomy/microcephaly, and less frequently, macrosomy/macrocephaly. Altogether, syndromic forms explain approximately 10% of ASD and can be diagnosed through clinical genetic testing, either targeted (e.g., fragile-X screening, Sanger sequencing of single genes, FISH) or unbiased (e.g., karyotype, array-CGH, NGS for gene panel or exome) sequencing. The medical management of these patients often requires surveillance over multiple organs beyond the Central Nervous System (CNS), given their multisystemic nature [34].
2. "*Mitochondrial" Autisms* – This term here designates all forms of ASD due to deficits in oxidative phosphorylation (OXPHOS) and mitochondrial malfunctioning [24]. OXPHOS requires over 80 proteins, the vast majority encoded by genomic DNA (gDNA) while only 13 are encoded by mitochondrial DNA (mtDNA). Therefore, these forms can stem from mutations or rearrangements affecting the mtDNA, or more often, the gDNA [35, 36]. It is important that the reader bears in mind that the terminology "mitochondrial autisms" has been used by some authors to designate rare forms of ASD carrying specifically mtDNA mutations or rearrangements. These patients will vary in severity and clinical presentation, based on the degree of heteroplasmy present in different tissues and especially in the CNS (i.e., the percentage of mitochondria carrying the mutation). However, these are indeed rare conditions, whereas OXPHOS parameters are often abnormal in ASD probably due to excess of oxidative stress [35, 37]. Examples of more common gDNA rearrangements yielding ASD with these characteristics include the following:
   (a) ch. 15q11–q13 deletions involving the COX5A gene, encoding the 5A subunit of cytochrome C oxidase.
   (b) cr. 13q13–q14.1 deletions involving MRPS31, which encodes the mitochondrial ribosomal protein 31.
   (c) cr. 4q32–q34.68 deletions affecting ETFDH, encoding the flavoprotein dehydrogenase which transfers electrons.
   (d) cr. 2q37.3 deletions involving the NDUFA10 gene, which encodes for the NADH dehydrogenase (ubiquinone) 1 alfa subcomplex subunit 10 [36].

Clinically, mitochondrial forms are associated with rather unusual neurological signs and symptoms, including ptosis, oculomotor anomalies, dysarthria, sensorineural hearing loss or deafness, prominent muscle hypotonia, or at times, hypertonia and movement disorders [32, 35]. Such children with ASD may regress with fever, whereas many idiopathic children with ASD improve in social cognition due to the transient abatement of hyperactivity. Except in the presence of mitochondrial depletion, family history is often positive on the maternal side, as the fetus receives mitochondria essentially from the oocyte. Microcephaly and microsomy are present in approximately 20% of cases, whereas it is macrocephaly and macrosomy that are prevalent in idiopathic forms of ASD [38, 39]. Several biochemical parameters associated with abnormal OXPHOS may be positive; muscle biopsy is usually negative in such children, whereas ragged red fibers are often seen in adults with "mitochondrial" autism, with brain MRI showing a variety of neuroanatomical abnormalities more often in these children than in those with idiopathic forms of ASD [32, 35].

Table 16.1 Main genetic syndromes associated with autism

	Gene/ch. region	Prevalence	Incidence of the syndrome in ASD	Incidence of ASD in the syndrome	Main signs and symptoms
Fragile X syndrome	FMR1	1/3500–1/9000	2.1%	25–33%	Facial dysmorphisms, macroorchidism; poor eye contact, social anxiety, language disorders, stereotyped/repetitive behaviors, hyperactivity, sensory hypersensitivity
Tuberous sclerosis	TSC1, TSC2	1–1.7/10,000	1–4% (8–14% if seizures present)	16–65%	Hamartomas of the skin, brain, kidneys, heart, lungs, retina, intellectual disability, learning disability, seizures
Neurofibromatosis type 1	NF1	1/3000–1/4000	<1.4%	4%	Cafe-au-lait spots, neurofibromas, axillary or inguinal freckling, optic glioma, bone dysplasias
Untreated phenylketonuria	PAH	1/10,000–1/15,000	–	5.7%	Microcephaly, muscle hypertonia, intellectual disability, language impairment, aggressiveness, psychomotor agitation, seizures
Adenylosuccinate lyase deficiency	ADSL	Not known	<1%	80–100%	Intellectual disability, severe autism, seizures, psychomotor regression
Smith–Lemli–Opitz syndrome	DHCR7	1/10,000–1/60,000	<1%	46–53%	Microcephaly, facial dysmorphisms, malformations (sometimes lethal, mostly cardiac, cleft palate, hypospadia), short stature, intellectual disability, sensory hypersensitivity, language impairment, self-injurious behavior, sleep disorders, opistokinesis, and other stereotyped behaviors
Cohen syndrome	COH-1, unknown	1/105,000	<1%	48%	Microcephaly, facial dysmorphisms, truncal obesity developing in mid childhood, hematologic and ophthalmic abnormalities, intellectual disability, motor clumsiness, language disorders
Cornelia de Lange syndrome	NIPBL SMC1A SMC3 unknown	1/10,000	<1%	35–50%	Facial dysmorphisms, short stature, malformations (cardiovascular, gastrointestinal, and musculoskeletal), developmental delay, intellectual disability, difficulty with swallowing, other- and self-directed aggressiveness, ADHD, obsessive-compulsive disorders (OCD), depression
Sotos syndrome	NSD1	1/10,000 – 1/50,000	<1%	Not known	Macrocephaly, excessive pre- and postnatal body growth, facial dysmorphisms, development delay

Cole–Hughes macrocephaly	Not known	Not known	<1%	Not known	Macrocephaly, intellectual disability, ADHD, developmental delay and bone maturation, language disorders, obesity, facial dysmorphism
Lujan–Fryns syndrome	UPF3B MED12	Not known	<1%	80%	Mild to moderate intellectual disability linked to the X chr. with marfanoid habitus, muscle hypotonia, ascending aortic aneurysm, aggressiveness, hyperactivity, emotional liability
San Filippo syndromes:		0.3–1.6/100,000	<1%	Not known	Behavioral regression or developmental delay, motor and verbal stereotypes, hyperactivity, aggressive behavior, sleep disorders, inappropriate affect, malformations (visceromegaly, facial, skeletal, etc.). Age at onset generally (but not always) after 3 years
A	SGSH				
B	NAGLU				
C	HGSNAT				
D	GNS				
ARX syndrome	ARX	Not known	<1%	Not known	Intellectual disability linked to the X chr. with or without autism; X-linked infantile spasms due to mutations or insertions; X-linked lissencephaly with agenesis of the corpus callosum and ambiguous genitalia in the presence of stop mutations, which lead to death due to developmental delay and drug-resistant epilepsy
Ch 2q37 deletion syndrome	2q37.3	Not known	<1%	Not known	Brachymesophalangy, intellectual disability
Williams–Beuren syndrome	7q11.23 del	1/7500–1/25,000	<1%	7%	Elfin facies, supravalvular aortic stenosis and in other large arteries, short stature, hypodontia, microdontia, hypercalcemia, friendly and gregarious personality or more rarely autism, ADHD, anxiety, visuocognitive dysfunction
Williams–Beuren region duplication syndrome	7q11.23 dup	1/12,500–1/20,000	<1%	35.7%	Growth retardation, facial and dental dysmorphisms, intellectual disability, developmental delay, expressive language disorder, seizures
Ch 13 deletion syndrome	13q	?	≤1%	?	Intellectual disability, language disorder, retinoblastoma, growth retardation, malformations (cardiovascular, craniofacial, gastrointestinal, renal, skeletal limbs, and fingers)
Ch 15q syndromes: Angelman syndrome	Del/mutation in maternal UBE3A	1/10,000–1/12,000	≤1%	42%	"Happy puppet" syndrome, facial dysmorphisms, developmental delay, language disorder, stereotyped behaviors, intellectual disability, ataxia, ADHD, psychomotor agitation, microcephaly, and seizures

(continued)

Table 16.1 (continued)

	Gene/ch. region	Prevalence	Incidence of the syndrome in ASD	Incidence of ASD in the syndrome	Main signs and symptoms
Prader–Willi syndrome	Del in paternal 15q11–q13	1/10,000–1/15,000	?	25.3%	Developmental delay, short stature, intellectual disability, hyperphagia, obesity, hypotonia, hypogonadism, OCD
Isodicentric cr. 15q	Dup 15q11–q13, GABRB3	1/30,000 (?)	≤1%	70%	Short stature, diabetes, anal and jejunal atresia, acanthosis nigricans, severe autism, developmental delay, intellectual disability, hypotonia, seizures
Hypomelanosis of Ito	Mosaic dup/del 15q11–q13	1/10,000	≤1%	10%	Macular hypopigmented whorls, streaks, and patches, neurological deficits, intellectual disability, seizures, multiple malformations (cerebral, ocular, and musculoskeletal)
Smith–Magenis syndrome	Del 17p11.2	1/25,000	≤1%	93%	Intellectual disability, developmental delay, self-injurious behavior, facial dysmorphisms, hearing loss; skeletal, renal, cardiac, and ocular abnormalities
Potocki–Lupsky syndrome	Dup 17p11.2	?	≤1%	?	ADHD, intellectual disability, developmental delay, short stature, hypotonia, facial dysmorphisms, cardiac and dental abnormalities
Down syndrome	Trisomy cr. 21	1/1000	≤2.5%	1–42%	Facial dysmorphisms, cardiac and intestinal malformations, intellectual disability of variable degree
Velocardiofacial/ Di George syndrome	Del 22q11.2	1/4000	≤1%	20–31%	Facial dysmorphisms, cleft palate, cardiac malformations, hypoplasia or aplasia of the thymus gland, hypoparathyroidism, autism, intellectual disability, developmental delay, ADHD, psychosis, seizures
Ch 22q11 duplication syndrome	22q11.2 dup	Not known	<1%	Not known	Facial dysmorphisms, velopharyngeal insufficiency, intellectual disability, developmental delay
Phelan–McDermid syndrome	22q13.3 del	Not known	<1%	50–70%	Minor dysmorphisms, severe muscle hypotonia, intellectual disability, developmental delay, language impairment

Del deletion; *Dup* duplication; *Ch* chromosome. Modified from Persico and Napolioni [24] and Persico [33]

3. *Autisms due to* de novo *or inherited rare genetic variants* – Genetic variants are defined "rare" when their minimum allele frequency (MAF) is <1%, as applied to the term "mutation," whereas they are defined "polymorphisms" when their MAF is ≥1%. In this class of autisms, a *single genetic variant* is entirely responsible for the onset of ASD [24, 25]. Rare pathogenic variants can be distinguished as follows: (a) large chromosomal aberrations, detectable by classical G-banding karyotype (many yield syndromic forms, as listed in ◘ Table 16.1); (b) CNVs, microdeletions, or microduplications ranging from 1Kb to several Mb in size and detectable only by array-CGH; (c) small insertions/deletions (indels), up to 1Kb in size, including frameshift mutations due to codon insertions resulting in additional amino acids being inserted into proteins; or (d) mutations, that is, single-nucleotide variants (SNVs) typically generating stop codons, yielding functionally inactive truncated proteins, or missense mutations causing amino acid changes resulting in "loss of function" or "gain of function." Both indels and mutations require Sanger sequencing or NGS for detection. Additional pathogenic mechanisms can include triplet repeat expansions, uniparental isodisomy, or CNVs which impact promoters, long noncoding RNAs, miRNAs or imprinting centers, affecting gene expression through epigenetic dysregulation. In reference to their *allele frequency*, pathogenic variants are often "very rare" or even "private" (i.e., found in a single patient or in very few patients worldwide, as can be seen on the Decipher database [40]). There are, however, some recurrent forms, due to pathogenic CNVs of identical or similar size which, albeit rare, are found in several patients (e.g., dup/dels in ch 1q21, 15q13, and 16p11.2). In terms of *inheritance pattern,* pathogenic gene variants can be dominant, recessive, X-linked or de novo. Clearly, pathogenic variants inherited from a parent either completely healthy or displaying only minimal ASD traits with variable penetrance require additional genetic variants or epigenetic contributions to express full pathogenicity. De novo mutations are estimated to occur in 15–25% of cases [31].

To date, there are well over 200 genes that if mutated, deleted, or duplicated, yield ASD [41]. Updated lists of these genes can be found in several databases like that of the Simons Foundation [42] and the "autismKB" database of the University of Beijing [43]. In general, ASD due to rare genetic variants is not clinically identifiable in any specific way, as carriers often do not display dysmorphisms or systemic signs/symptoms, especially when CNVs affect only genes expressed in the CNS. Nonetheless, mutation or indels in specific autism-associated genes can produce peculiar systemic signs, such as macrocephaly or oculomotor deficits for PTEN and HOXA1, respectively [44, 45], albeit with sizable interindividual phenotypic variation. Finally, it is important to consider that CNVs and SNVs are typically nonspecific in their neurodevelopmental outcome, due to the pleiotropy of many genes in psychiatric disorders [46]. It is thus not unusual that mutations affecting the same gene or at times even the same mutation may yield clinically different neurodevelopmental disorders, ranging from ASD, intellectual disability, global developmental delay, language development disorder, learning disability, as well as schizophrenia spectrum (see also Chap. 10).

4. *Autisms due to common genetic variants and gene–environment interactions* – Collectively, 10% of individuals with ASD are syndromic, another 8–10% carry one pathogenic CNV detectable by array-CGH [47], and the yield of exome sequencing for indels and mutations located in the coding sequence reaches 8–16% [47] with even higher positive rates reported for developmental delay [48]. Another 10–20% of ASD patients carry likely pathogenic CNVs or functionally relevant CNVs able to contribute, in conjunction with other genetic variants, to the disorder. At this time, it is not yet clear how much whole

genome sequencing (WGS) shall add in providing additional variants that may be responsible for ASD [49]. Therefore, at this time, at least half, and possibly the majority of genetic risk for ASD, appears to be carried by common variants able to negatively affect neurodevelopment by influencing gene expression or protein function [50, 51]. Single-nucleotide polymorphisms (SNPs) are SNVs with MAF >5% in the general population. Each single common variant obviously cannot be pathogenic (otherwise, it would not be "common" in the general population), and explains on average <1% of phenotypic variance, in some instances reaching 1–5% [52]. However, an unfavorable combination of common variants can additively increase vulnerability, enhancing the penetrance of rare variants and/or decreasing the sensitivity threshold for environmental factors able to disrupt neurodevelopment [51, 53]. Several genes encompass common variants with replicated and well-established functional relevance in ASD, including genes encoding the GABA-A beta3 receptor (GABRB3), the oxytocin receptor (OXTR), reelin (RELN), the serotonin transporter (SLC6A4), subunit 2B of the glutamate NMDA receptor (GRIN2B), arginin-vasopressin receptor 1A (AVPR1A), engrailed2 (EN2), integrin beta3 (ITGB3), the proto-oncogene MET, and contactin-associated protein-like 2 (CNTNAP2) [24, 25]. Many other genes are listed in the databases and gene lists mentioned above.

Over the years, the genetics of ASD has moved away from single gene analysis and has been focusing more on "gene networks," the genetic substrate of "molecular pathways" whose dysfunction would explain ASD in each individual [54, 55]. The concept of "gene network" refers to the functional role of the protein encoded by each gene, and collects into a single network all those gene-encoding proteins which interact in a single molecular pathway (examples of gene pathways can be found in the "Gene Ontology Resource" [56]). Genes located at the intersection between different pathways can be identified as "hub genes," whose dysfunction can negatively affect multiple functional pathways. These hub genes often correspond to genes which, if mutated, deleted, or duplicated, yield ASD due to rare single variants.

Gene networks more frequently involved in ASD, as identified through array-CGH, NGS, and/or RNAseq for genome-wide transcriptomics, are involved in synaptogenesis and synaptic management, neurite outgrowth, chromatin remodeling, cytoskeleton and synaptic scaffolding, cell proliferation, transcriptional regulation, and activity-dependent translation [54, 55]. Molecular pathways include Wnt (Wingless and Int-1) signaling, PI3K/mTOR, and Ca^{2+}/calmodulin [57].

16.3.2 Epigenetics

The study of rare and common genetic variants has provided converging evidence for transcriptional dysregulation as one of the pathways most able to lead to ASD. Epigenetics refers to variation in gene expression as a cause for (a) interindividual phenotypic variability, when genetic factors are held constant (e.g., phenotypic differences between monozygotic twins) and (b) nongenetic transmission of traits from one generation to the next [58, 59]. Governing the interaction between genomic DNA, histones and the transcriptional machinery, epigenetic mechanisms, primarily involving DNA methylation/demethylation with the formation of 5-methylcytosines in CpG (C-phosphate-G sequence of DNA nucleotides) islands and histone deacetylation/acetylation, respectively, control chromatin structure [58, 59]. This, in turn, determines the accessibility of coding genes to the transcriptional machinery, as well as the production rates of regulatory miRNAs and long noncoding RNAs. Epigenetics is highly organ- and tissue-specific, even brain region-specific, if we refer to the CNS. This regional specificity in epigenetic markings (i.e., cytosine methylation and histone acetylation patterns) is paralleled by regional differences in gene expression, as evidenced by genome-wide transcriptomics

[60, 61]. While functionally ideal to provide maximum adaptation to different tissues, organs and systems [58], this aspect poses a relevant experimental limitation, preventing from extending conclusions beyond the cell type, organ, or brain region directly assessed in each specific study.

At least three aspects of epigenetic contributions to ASD deserve attention:

(a) Prenatal and early postnatal exposure to environmental factors during critical periods in neurodevelopment, produce permanent changes in DNA methylation patterns able to influence gene expression throughout the lifetime of the individual. This mechanism is a primary contributor to the teratological consequences of prenatal exposure to pharmacological and illegal drugs [62], as well as to malnutrition and gestational complications, during pregnancy [63, 64].

(b) Abnormal methylation patterns have been recorded in individuals with ASD applying various technologies to different tissues and brain regions. These differences impact several genes relevant to neurodevelopment, such as OXTR, RELN, EN2, MECP2, and GAD1 [65]. Interestingly, several of these genes also contain common genetic variants contributing to ASD risk, as discussed above. Epigenome-wide changes are usually complementary to differences reported in genome-wide transcriptomic studies [65], highlighting enhanced expression of immune genes likely due to neurinflammation and microglial activation in ASD brains, as opposed to hampered expression of neural activity genes [60, 61]. Interestingly, also epigenetic mechanisms are under the influence of common genetic variants particularly enriched in the proximity of immune genes [66].

(c) DNA methylation is removed from gDNA following fertilization during preimplantation development through the process of "DNA methylation reprogramming," only to be reestablished subsequently in the blastocyst [67]. However, in real life, the process of demethylation can be hampered, as it occurs in cloned embryos which display reduced demethylation and precocious de novo methylation already at the 4- and 8-cell stages [69]. Even in normal embryos, DNA reprogramming is far from complete: some DNA methylation marks present in parental gametes do remain and foster the multigenerational transmission of phenotypic traits in the absence of direct contributions by the DNA sequence [69]. Classical studies following up the offspring of mothers pregnant during the Dutch famine of 1944 have recorded an epigenetically driven increase in incidence of obesity and metabolic disorders in the second-generation offspring of fathers exposed to the famine in utero [70]. Interestingly, hypo- or hypermethylation has been uncovered in the gDNA extracted from sperm cells of fathers of autistic children, especially in the vicinity of loci relevant to neurodevelopment, including several SNORD genes [71]. Initial evidence of possible correlations between the degree of abnormal methylation at some of these loci and clinical scores at the Autism Observation Scale for Infants (AOSI) [71] is indeed exciting but requires caution until replicated.

In conclusion, epigenetic mechanisms represent the bridge between genetic predisposition, environmental influences, and dysimmunity/neuroinflammation, the three components currently viewed as most relevant to the etiopathogenesis of multifactorial forms of ASD. Possible contributions by epigenetic abnormalities present in parental gametes significantly broaden the time window of environmental influences potentially able to confer autism risk.

16.3.3 Environmental Factors

The role of environmental factors in the etiology of ASD has been the object of intensive research, especially spurred by the dramatic increase in ASD incidence recorded during the past 20 years [53, 72]. In interpreting the results, it is critical to keep in mind that a temporal correlation between an environmental

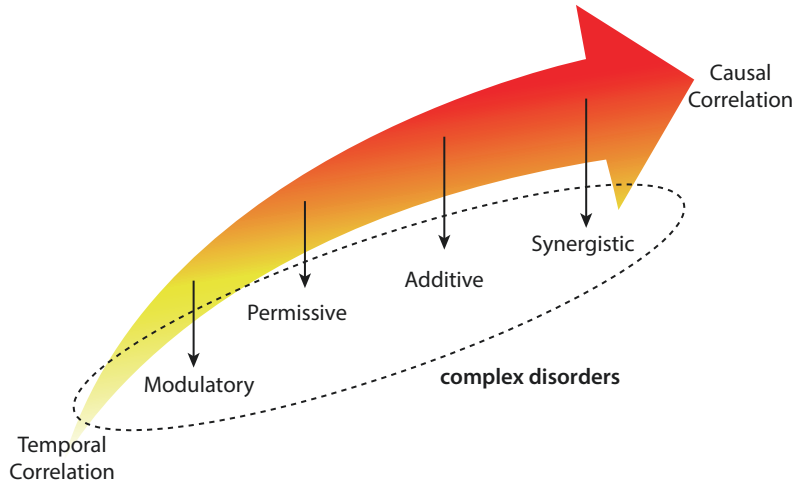

Fig. 16.1 Environmental factors in human disorders. (From Ref. [72], with permission)
A "none-to-all" upward scaling model, spanning from temporal coincidence without any pathogenic role to full causality. Environmental Factors (EF), in interaction among themselves and with Genetic Factors (GF), can exert: (a) modulatory effects: EF influence the phenotypic expression of GF only qualitatively; (b) permissive effects: one EF is necessary for another EF to exert damage, or for a GF to become penetrant and to achieve phenotypic expression; (c) additive effects: EF and GF act independently and their combined effect equals the sum of each contributing factor; (d) synergistic effects: EF and GF potentiate each other, so that the combined effect is greater than the predicted sum of each contributing factor

exposure and ASD phenotype will need to be further substantiated by means of cellular and animal models and understood at the molecular level with support through human epidemiological, neuropathological, and clinical studies. Furthermore, causal links can assume various forms (Fig. 16.1 from [72]). An environmental factor can play a variety of roles:

- *Causal* – the environmental factor alone is sufficient to cause the disorder.
- *Synergistic* – the environmental factor boosts the penetrance of a genetic predisposing factor or the damaging effects of another environmental factor, so that [1 + 1] > 2.
- *Additive* – environmental effects add up linearly with other environmental and genetic effects, so that [1 + 1] = 2.
- *Permissive* – the environmental factor does not exert direct damage on neurodevelopment, but sensitizes the CNS or produces the priming needed to allow the damaging effects of another factor (environmental or genetic), so that [0 + 1] > 1.
- *Modulatory* – the environmental factor cannot cause the disorder, which is caused by other genetic and environmental factors; however, once these are present and active, the environmental factor influences the timing of onset and the severity of the clinical phenotype.

Although not considered as sole cause of ASD, early environmental factors may play an important role (Table 16.2A) [53, 72] envisioned under prenatal, perinatal, and postanatal factors.

Prenatal Factors

The prenatal environmental factors include infections, mainly caused by neurotropic viruses, allergic or autoimmune problems, endocrinological problems, exposure to drugs or toxic substances, gestational hypertension, gestational diabetes, excess of testosterone in the amniotic fluid, threat of abortion and predelivery hemorrhage [73–78].

Fetal anticonvulsant syndrome – caused by prenatal exposure to valproic acid or, less frequently, to carbamazepine [79]. Clinically it is characterized by expressive language delay, global psychomotor delay, relatively preserved cognition. Facial dysmorphisms are typically

Table 16.2 Environmental factors able to (A) cause autistic disorder or (B) exert synergistic or additive effects (from Ref. [72], modified)

A	Critical period	Mechanism
Valproic acid	E18–E30	Altered gene expression due to histone hyperacetylation, inhibition of neural progenitor proliferation vie Ras-ERK, oxidative stress, folic acid antimetabolism
Thalidomide	E20–E30	Altered gene expression, ROS-mediated DNA damage, angiogenesis inhibition
Misoprostol	E18–E42	Altered gene expression, angiogenesis inhibition
Rubella	Up to E56	Direct viral damage, maternal and fetal immune response
Cytomegalovirus	Throughout pregnancy	Direct viral damage, maternal and fetal immune response
B		
Air pollution *Smoking during pregnancy*	Throughout pregnancy & early postnatal life	Oxidative stress, CNS and systemic immune activation, cerebral vascular damage, neuronal cell death, altered gene expression through dysregulated DNA methylation
Pesticides: *Organophosphates* *Pyrethroids* *PhtalatesPhthalates*	Throughout pregnancy	CNS and systemic inflammation, oxon disruption of neuroglial proliferation & differentiation, decreased Reelin expression and enzymatic activity, decreased BDNF expression, interference with intracellular Ca^{2+} signaling, disruption of GABAergic neurotransmission, disruption of androgen activity
Polychlorinated biphenyls (PCB) *Polybrominated diphenyl ethers (PBDE)*	Throughout pregnancy	Endocrine disruption of thyroid hormone homeostasis, oxidative stress, interference with intracellular Ca^{2+} signaling, altered gene transcription through decreased DNA methylation, immune system activation
Heavy metals	Throughout pregnancy	Neurotoxicity, immune system activation/autoimmunity, altered gene transcription
Antidepressants (prolonged and high-dose)	Throughout pregnancy	Extracellular serotonin excess

Legend: *E* embryonic day postfertilization [24].

present (see hypertelorism and frontal bossing in Figs. 1 and 2 of Moore and colleagues [80]). It is primarily caused by histone deacetilase inhibition, interfering with epigenetic control of chromatin structure [72].

Exposure to other theratogenic drugs – thalidomide and misoprostol exposures can also cause autism. Thalidomide was prescribed during pregnancy as a sedative-anxiolytic instead of barbiturates, but was withdrawn from public use in 1961 due to severe teratogenicity. Misoprostol is used to produce abortion in South America. Exposure to these two drugs, in addition to producing major systemic consequences, yields ASD when exposure occurs in a very early time window in pregnancy corresponding to the closure of the neural tube, that is, embryonic day 18–30 post fertilization (Table 16.2) [81].

Congenital viral infections (rubella, cytomegalovirus) – Prenatal infection with rubella virus and cytomegalovirus have been

the most linked to ASD, typically associated with severe intellectual disability, deafness, and malformations, often affecting the eye and heart. Epilepsy and cerebral palsy are also frequent among outcomes. Brain MRI typically shows a heterogeneous variety of abnormalities, ranging from cortical malformations (polymicrogyria, pachygyria, heterotopias) to periventricular leukomalacia, due to defects in neuronal migration and myelination, respectively [82, 83]. This damage is due both to direct viral damage and indirect disruption produced by the maternal immune reaction and local neuroinflammation.

These agents have been conclusively demonstrated to cause ASD, although their effects depend on the timing of exposure during pregnancy and on the genetic and immune predisposition of the maternal-fetal pair.

Converging evidence is also supporting additive or synergistic effects for other environmental factors, with conflicting results due to confounding (Table 16.2B) [53, 72]. The association between air pollution and ASD, for example, is primarily supported by studies performed in the United States, with results influenced by confounding factors such as socioeconomic status and living close to sources of traffic and/or industrial air pollution. European studies do not support such an association with air pollution and prenatal exposure to cigarette smoking has also yielded inconclusive results [53]. Pesticides and insecticides, especially the organophosphate clorpyrifos, used in households by exterminators until a few years ago, were noted as potential candidates due to genetic predisposition in common variants of the PON1 gene being associated with ASD in North American, but not in Italian families [84]. This initial genetic evidence was subsequently followed by studies demonstrating an association between prenatal exposure to organophosphates and various neurodevelopmental outcomes, including ASD or autistic traits, ADHD, mild intellectual disability or borderline cognitive level, language development disorder, and learning disability [85–91], with some studies yielding no association [92]. Studies regarding phthalates and pyrethroids have also been suggestive, but with less consistent findings compared to studies involving organophosphates. Similarly suggestive, yet still inconclusive, has been evidence of contributions by heavy metals and persistent pollutants like polychlorinated biphenyls (PCBs) used in electric equipment [53]. Prenatal exposure to selective serotonin reuptake inhibitors (SSRIs) was initially proposed as a potential disruptor of neurodevelopment, based on minor barrel cortex anomalies observed in serotonin transporter Hz knockout mice [93]. This proposal was later confirmed by seven studies collectively demonstrating a 1.8-fold increase in ASD risk for children prenatally exposed to SSRIs [94].

Other prenatal factors, ranging from gestational complications (diabetes, preeclampsia, and prenatal birth) involving maternal folic acid, vitamins, iron, and fatty acids, have also been recently discussed [53]. For these latter set of factors, effect sizes appear inconsistent; the association with gestational complications may likely stem from shared genetic underpinnings and the demonstration of putative add-on environmental effects on ASD risk is complex.

Perinatal Factors

Perinatal factors are mainly represented by obstetric problems, such as the abnormal duration of gestation, the pharmacological induction of childbirth, low birth weight, and perinatal asphyxia. Children born at 28-week gestation have a high risk of neuropsychic problems that remains high up to the 33-week gestation particularly for ASD. The specific determinants of these risk factors have not yet been precisely identified. Research is investigating the role of alterations in the intestinal microbiome, the use of synthetic oxytocin to induce or stimulate delivery, excessive production of stress hormones, in particular CRH (corticotropin-releasing hormone), and derivatives of oxygen reduction (ROS - Reactive Oxygen Species) [95–99].

Postnatal Factors

The postnatal factors that have been evaluated are numerous and heterogeneous, includ-

ing the breakdown of the equilibrium between the production and the elimination of oxidizing substances (harmful to nerve cells), anomalies of development of the amygdala, vitamin D deficiency, heavy metals, increase of intestinal permeability by food toxicity (especially gluten and casein), and vaccines. For all these factors, there is no convincing evidence of causal value.

The recommendations of the National Institute for Health and Care Excellence (NICE) state not to consider special diets in the management of ASD symptoms. Analyses of numerous case–control and cohort studies have shown no association of vaccines with ASD risk over the last two decades [100]. The hypothesis that measles-mumps-rubella (MMR) vaccine causes ASD by damaging the intestinal lining [101] has been refuted with retraction of the Lancet paper in 2010. Further, neither the changes in vaccination policies nor the elimination of the mercury containing preservative thimerosal from vaccines has been followed by changes in ASD incidence trends in countries where these changes have been applied [100]. Thimerosal is ethylmercury, whose toxicological target organ in case of intoxication is the kidney because it crosses the blood–brain barrier only to a very limited extent; instead, it is methylmercury that readily crosses the blood–brain barrier targeting the brain and that has been responsible for the tragic incident of Minimata Bay in Japan [102]. The World Health Organization has also endorsed safety of vaccines: "The overall evidence clearly indicates no association of MMR vaccine with either inflammatory bowel disease or with developmental disorders, including autism" [103] and "There is no evidence of a link between measles-mumps-rubella (MMR) vaccine (or any other vaccine) and autism or autistic disorders" [104].

A possible increase in ASD prevalence risk is linked to some autoimmune diseases and viral infections with temporal correlation reported by some parents in the setting of developmental and behavioral regression, with potential for enhanced oxidative stress that accompanies immune reactions, but the same phenomenon can be observed in children with syndromic ASD, as well as those with idiopathic form who have regressed following immune activation due to recurrent infections, such as otitis media, or different forms of allergies, both significantly more frequent among in children with ASD compared to typically developing children [105, 106]. This phenomenon may characterize a cluster of what has been termed as "dysimmune" children with ASD with relatively specific clinical features and family history [38, 107, 108].

Anatomopathological Correlates

The application of recent neuroimaging techniques has made it possible to study the anatomical development of the brain and neural connectivity, providing in vivo information on the structure of the brain in people with autism. Structural magnetic resonance studies have shown an early and accelerated development of the brain, both of the gray and the white matter, and of the cranial circumference in about 20% of children with ASD [109], detectable in the early childhood and estimated at 5–10% of total brain volume [110]. This acceleration seems to occur starting from the first year of life (9–12 months) [111] and peaks around 2–4 years of age, and is followed by a gradual reduction, until recovery of normal dimensions at the end of adolescence [112, 113].

Abnormalities of the cortex at the dorsal-lateral, superior parietal, intraparietal sulcus, anterior cingulate, as well as insula, amygdala, and the caudate nucleus were found [114]. Further studies with magnetic resonance imaging and other imaging techniques of CNS functioning have explored the abnormalities of activity in various brain areas in relation to the neuropsychological skills frequently compromised in ASD. Alterations of the corpus callosum, cingulate gyrus, superior longitudinal fasciculus, temporoparietal junction, orbital-frontal cortex, and cerebellum have been reported. These brain areas have been repeatedly associated with complex cog-

nitive and motor functions, such as attention, working memory, muscle coordination, or inhibition [115].

- **Electrocortical Correlates**

Research on this topic has mainly used electroencephalography and magnetoencephalography. The most frequently detected alterations are a reduction in long-range connectivity in low-frequency waves [116–119] and an increase in connectivity to short range for high-frequency waves [120]. In general, alterations in wave frequencies have been reported, interpreted as correlates both of excess and defect of connections between neural areas [120, 121].

Multiple types of genetic factors influence the continuum of behavioral and developmental characteristic, with the extreme end of the continuum resulting in ASD. Insights into genomic variability and biology of ASD are increasingly being uncovered with rapid progress in establishing a correspondence between behaviorally defined clinical symptoms of ASD and genetic causation. However, finding the genetic variations and understanding their meaning are equally important, including identification of protein networks affected by many different gene mutations that lead to similar brain dysfunction. The study of rare and common genetic variants has provided converging evidence for epigenetic factors and transcriptional dysregulation as one of the pathways also able to lead to ASD. The role of environmental factors in the etiology of ASD has also been the object of intensive research. In interpreting the results, it is critical to keep in mind that a temporal correlation between an environmental exposure and ASD phenotype will need to be further substantiated by means of cellular and animal models and understood at the molecular level with support through human epidemiological, neuropathological, and clinical studies.

16.4 Diagnostic Criteria and Clinical Features

16.4.1 Definition and Diagnostic Criteria

ASD represents a group of heterogeneous conditions sharing two key features: (a) impairment of socialization and qualitative deficits of communication, for example, reluctance to make eye-contact, lack of appropriate peer relationships, lack of emotional reciprocity, delay in verbal responses, poor conversation skills, lack of pretend play; and (b) restricted and/or repetitive behaviors or interests, for example, repetitive motor movements, preoccupation with parts of objects, hyperfocused interests, and hyper- or hyporeactivity to sensory input [122–130].

The term "autism" was coined by Eugen Bleuler in 1911, to describe one of the four fundamental aspects of schizophrenia, reflecting the affected individual's 'inward turning' into his/her own world and thereby losing contact with the outer world. Bleuler's overall conceptualization of schizophrenia is focused on the loss of interpretative and relational skills, as summarized in the 4A theory: inappropriate or flattened *Affect*, cognitive, affective, and volitional *Ambivalence*, loosening of thought *Associations*, and *Autism*, which was defined as "preference for living in a fantasy world," a gap between the patient's inner fantasy world, inaccessible to reality, and the unloving environment [131, 132]. European phenomenological psychiatry in the field of schizophrenia offered other views to grasping the nature of autism: Eugene Minkowski considered it as "a loss of vital contact with reality" [133, 134], Ludwig Binswanger identified its main characteristic in the "inconsistency of natural experience" [135], and Wolfgang Blankenburg defined it as "the global crisis of common sense" [136].

The discovery of autism owes its origins to the separate works of Leo Kanner and Hans Asperger, who in 1943 [137] and 1944 [138], described a series of children with clinical

picture consistent with current descriptions. The "Autistischen Psychopathen" patients described by Asperger were different from those of Kanner in three aspects: (1) more fluent speech; (2) difficulty in carrying out gross motor movements but not fine movements; (3) different level of learning ability. Asperger called such patients "abstract thinkers" or "little professors," which, according to Kanner, have great problems with mechanical learning. Even with important common traits, two different clinical forms were determined: Kanner's Autism and Asperger's syndrome. During the period 1994–2013, Asperger's syndrome became the term applied to the individuals with ASD without deficits in intellectual functioning and significant language delay.

The term autism had been used in pediatric circles in Europe as early as 1920s to describe the social withdrawal experienced by young children [139]. In this perspective, the transition from the Bleulerian autism to current use has its roots in a literature of clinical descriptions produced between the end of the nineteenth century and the beginning of the twentieth century. Children with a marked tendency to self-isolation and poor affective contact were characterized under the rubric "hereditary insanity" or hereditary psychosis, many of whom with remarkable intelligence, as described in his textbook by Heinrich Schüle, a German physician [140]. By the 1880s, similar features associated with catatonia were noted in children with developmental delay. In 1887, the English pediatrician Langdon Down associated autistic behavior with catatonia in "developmental idiocy," although he used neither the term autism nor catatonia. In his descriptions of the children with poor social reciprocity he noted, "returns your kiss by a bite," inflexibly mute, withdrawn, and scarcely reactive, regardless of the circumstances around them, and sometimes fascinated by peculiar sensory stimulations, for example, the rhythm of music. Langdon Down also described automatic movements of the fingers and rhythmical movements of the body, now termed as stereotypies [141]. In 1913, Kraepelin described a form of childhood-onset dementia praecox, akin to the current picture of autism. He referred to these individuals by embracing the Bleulerian autism [142]. In the ensuing years, the term autism got widespread use across Europe. In 1924, Moritz Tramer described gifted children with developmental delay, idiosyncratic language, and excellent memories and drawing skills. In Russia, the pediatric neurologist Grunya Sukhareva was the first to describe infantile autism but entitled her paper published in 1926 as "schizoid psychopathy"; she applied Kretschmer's idea about autism and underlined the association with catatonia among these children [143]. The work of Sukherava was expanded in Moskow by Ewa Grebelskaja-Albatz, who in 1934 published a case series on 22 children with autism, psychosis, and/or catatonia, focusing on patients with lower functioning [144]. Due to the language, these remarkable works were not widely publicized, until the 1981, as was the case with the translation of Asperger's paper by Lorna Wing. Even in the United States, some authors had descriptions of young individuals with schizophrenia who can now be considered as having an ASD, as with description by Howard Potter, Lauretta Bender, and Juliette Louise Despert in the context of childhood schizophrenia [145] consistent with the notion of a neurodevelopmental continuum of social deficits that may be shared by these conditions [146–148].

Advances in the clinical conceptualization of ASD as a spectrum are attributed to Lorna Wing who was a mother of a son with autism and founder of the National Autistic Society in the UK. Wing distinguished three types of social interaction:

- ALOOF: individuals indifferent to other people, detached, characterized by the presence of motor stereotypes, good mechanical skills and visual-spatial abilities, and medium-severe cognitive impairments ("classic" autistic disorder)
- PASSIVE: individuals who tend to allow others to interact with them but drift back soon to isolation if left to their own routines or devices; in comparison to the aloof type, this type may be detected later in life

- ACTIVE/ODD: individuals who make spontaneous approaches to others but in a peculiar, naive, and/or nonreciprocal way; they present good cognitive abilities (high-functioning autism) [149]

During the 1950s and 1960s, autism was still considered an early manifestation of schizophrenia, which was thought of as an emotional disorder caused by pathological parent–child interaction. Even the subsequent editions of Kanner's textbook on Child Psychiatry during this period grouped autism under childhood schizophrenia. Bruno Bettelheim, an Austrian-born psychologist, used the term "refrigerator mothers" to characterize a cold and unemotional mother theory [150]. Melanie Klein published a paper on child personification in which she argued that if a child's instinctual wish fulfillment took precedence over their recognition of reality, the child was suffering from a form of 'psychosis' [151], a term Freud coined in 1894 to describe 'hallucinatory confusion' [152]. In the first two editions of the DSM, autism was classified under the terms "schizophrenic reaction, childhood type" [153] and "childhood schizophrenia" [154], respectively. During the 1970s, the psychogenic paradigm was abandoned, and the scientific community began to consider biological roots of autism, especially with the publication of Bernard Rimland's book on infantile autism in 1964, written after his son was diagnosed with the disorder, on "its implications for a neural theory of behavior" [155].

The International Classification of Diseases – Ninth Revision, ICD-9 [156] considered autism as a diagnostic subcategory of the childhood schizophrenia. The publication of DSM-III [157] was an important threshold and represented a cleavage between the international classification systems. The DSM-III [157] introduced six inclusion criteria for infantile autism, with symptoms onset early by 30 months of age, communication and language impairments, narrow interests, and fear of change. For the first time, the distinction between childhood schizophrenia and autism was emphasized with the introduction of overarching metacategory of pervasive developmental disorder (PDD) with five subcategories: infantile autism, residual infantile autism, childhood-onset pervasive developmental disorder, residual childhood-onset pervasive developmental disorder, and atypical autism. The concept of autism as a "syndrome" was also mentioned.

The DSM-III-R [158] provided a more complex definition requiring at least 8 of 16 criteria to be met in three domains of social interaction, communication/imagination, and interest/activity. The age of onset was moved to first 36 months of life. A new subcategory under the umbrella PDD term, pervasive developmental disorders-not otherwise specified (PDD-NOS) was also introduced. The PDD-NOS was defined by subthreshold symptoms, with no specification of requisite symptoms and often too mild to allow a diagnosis of true autism that could be considered if a child does not meet the diagnostic criteria for PDD-NOS. Revisions made in the DSM-III-R included changing infantile autism to autistic disorder, while childhood onset pervasive developmental disorder and residual infantile autism were dropped.

The DSM-IV [159] further refined the diagnostic criteria and increased the number of conditions included under PDD to five subtypes: autistic disorder, Asperger's disorder (introduced for the first time), Rett's disorder, childhood disintegrative disorder, and PDD-NOS. The DSM-IV also added autism to the general chapter "global alterations of psychological development" and particularly to "generalized developmental disorders."

In the DSM-IV text revision (DSM-IV-TR) [160], the diagnostic categories and criteria remained consistent with the earlier edition, with the diagnosis of autism requiring the following criteria:

A. A total of six (or more) items from (1), (2), and (3), with at least two from (1), and one each from (2) and (3):

1. Qualitative impairment in social interaction, as manifested by at least two of the following:
   (a) Marked impairments in the use of multiple nonverbal communication
   (b) Failure to develop peer relationships
   (c) A lack of spontaneous seeking to share enjoyment, interests, or achievements with other people
   (d) Lack of social or emotional reciprocity
2. Qualitative impairments in communication as manifested by at least one of the following:
   (a) Delay in development of spoken language or absence of spoken language not accompanied by an attempt to compensate through alternative modes of communication
   (b) In individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
   (c) Stereotyped and repetitive use of language or idiosyncratic language
   (d) Lack of varied, spontaneous make-believe play or social imitative play
3. Restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least two of the following:
   (a) Stereotyped and restricted patterns of interest that are abnormal either in intensity or focus
   (b) Excessive adherence to specific, non-functional routines or rituals
   (c) Stereotyped and repetitive motor mannerisms
   (d) Persistent preoccupation with parts of objects

B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years:
   1. Social interaction
   2. Language as used in social communication
   3. Symbolic or imaginative play

C. The disturbance is not better accounted for by Rett's Disorder or Childhood Disintegrative Disorder.[1]

Given the symptom variability and inconsistency across clinical and research settings in diagnoses, the DSM-5 [161] combined the four previous subtypes into ASD, as single condition with three severity levels. The DSM-5 also introduced a new symptom of hyper- or hyporeactivity to sensory input or unusual interests in sensory aspects of the environment. The DSM-IV autistic disorder, Asperger's disorder, childhood disintegrative disorder, and PDD-NOS (atypical autism) were no longer included and Rett's disorder was now considered as an independent genetic disorder [162]. According to DSM-5, the diagnosis of ASD requires two core symptoms: (a) deficits in social communication and social interaction and (b) restricted repetitive behaviors, interests, and activities [163]. The age of onset is modified from 36 months to "early childhood," with the possibility of other ages of onset in which deficits may not become fully manifest until social communication demands exceed limited capacities. In DSM-5, concurrent diagnosis of ADHD with ASD is allowed.

According to DSM-5, the ASD diagnostic criteria include the following:

A. Persistent deficits in social communication and social interaction across multiple contexts, as manifested by the following, currently or by history:
   1. Deficits in social-emotional reciprocity, ranging from abnormal social approach and conversation to reduced sharing of interests, emotions, or affect
   2. Deficits in nonverbal communicative behaviors used for social interaction, ranging from poorly integrated verbal and nonverbal communication to abnormalities or even absence of eye contact, facial expressions

1 The text of these diagnostic criteria is not the same as that of the DSM-IV-TR. For the exact wording of the criteria, please refer to the original manual.

3. Deficits in developing, maintaining, and understanding relationships, ranging from difficulties in adapting to various social contexts and making friends to absence of interest in peers

B. Restricted, repetitive patterns of behavior, interests, or activities, as manifested by at least two of the following, currently or by history:
   1. Stereotyped or repetitive motor movements, use of objects, or speech
   2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior
   3. Highly restricted, fixated interests that are abnormal in intensity or focus
   4. Hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment

C. Symptoms must be present in the early developmental period (but may not become fully manifest until social demands exceed limited capacities or may be masked by learned strategies in later life)[2]

The DSM-5 defines three levels of severity based on a person's limitations in adaptive functioning and management of daily life:

- Level 1: Requiring support
- Level 2: Requiring substantial support
- Level 3: Requiring very substantial support

Level 1 ASD is the mildest, or the most "high-functioning" form of autism. Persons diagnosed with this level are usually able to speak in full sentences and communicate but have problems with engaging in back-and-forth conversation with others, initiating interactions, and making friends as well as with flexibility, organization, planning, and switching between activities.

Persons with level 2 have more problems with verbal and nonverbal communication and social skills. Likewise, they find it harder to change activities or focus and present markedly restricted and repetitive behaviors.

Level 3 corresponds to the highest impairment of functioning. Persons in this level show severe impairment of verbal and nonverbal communication and social interaction. They also face extreme difficulty in changing their daily activities or routine and experience a high level of distress if a situation requires them to alter their focus or task.

In the ICD-11 [164], ASD is defined by a) persistent deficits in the ability to initiate and to sustain reciprocal social interaction and social communication, and by b) a range of restricted, repetitive, and inflexible patterns of behaviour, interests or activities that are clearly atypical or excessive for the individual's age and sociocultural context.

- Understanding of, interest in, or inappropriate responses to the verbal or non-verbal social communications of others.
- Integration of spoken language with typical complimentary non-verbal cues such as eye contact, gestures, facial expressions, and body language. These non-verbal behaviors may also be reduced in frequency or intensity.
- Understanding and use of language in social contexts and ability to initiate and sustain reciprocal social conversations.
- Social awareness, leading to behavior that is not appropriately modulated according to the social context.
- Ability to imagine and respond to the feelings, emotional states, and attitudes of others.
- Mutual sharing of interests.
- Ability to make and sustain typical peer relationships.

Manifestations of alterations of patterns of behavior, interests, or activities include the following:

- Lack of adaptability to new experiences and circumstances, with associated distress, that can be evoked by trivial changes to a familiar environment or in response to unanticipated events.
- Inflexible adherence to particular routines; for example, these may be geographic such as following familiar routes or may require precise timing such as mealtimes or transport.

2 The text of these diagnostic criteria is not the same as that of the DSM-5. For the exact wording of the criteria, please refer to the original manual.

- Excessive adherence to rules (e.g., when playing games).
- Excessive and persistent ritualized patterns of behavior (e.g., preoccupation with lining up or sorting objects in a particular way) that serve no apparent external purpose.
- Repetitive and stereotyped motor movements such as whole body movements (e.g., rocking), atypical gait (e.g., walking on tiptoes), unusual hand or finger movements, and posturing.
- Persistent preoccupation with one or more special interests, parts of objects, or specific types of stimuli (including media) or an unusually strong attachment to particular objects (excluding typical comforters).
- Lifelong excessive and persistent hypersensitivity or hyposensitivity to sensory stimuli or unusual interest in a sensory stimulus, which may include actual or anticipated sounds, light, textures (especially clothing and food), odors and tastes, heat, cold, or pain.

As for the DSM-5, the onset of the disorder is referred to the developmental period, typically to early childhood, although symptoms may not become fully manifest until later, when social demands exceed limited capacities. Deficits are requested to be sufficiently severe to cause impairment in personal, family, social, educational, occupational or other important areas of functioning and are usually a pervasive feature of the individual's functioning observable in all settings, although they may vary according to social, educational, or other context. Individuals along the spectrum exhibit a full range of intellectual functioning and language abilities. The ICD-11 [164] divides ASD into 5 subcategories based on whether or not a disturbance of intellectual development is present, as well as the degree of functional language impairment.

The core symptoms that characterize ASD include qualitative impairments in (i) communication and social interaction and (ii) restricted and repetitive patterns of behavior, activities, and interests, with an important feature now included in the latter criteria set in the DSM-5 and ICD-10 involving the alteration of sensory modulation.

16.4.2 Onset and Presentation

Complementary sources of information have contributed to the notion that early signs of ASD are detectable during the first year of life. The observations of Leo Kanner in 1943 [137] of infantile autism as an innate disorder of emotional contact are reminiscent of this view that include the following:

1. Studies noting parents' memories of the early characteristics of their child's development collected through parents' questionnaires or interviews
2. Retrospective analysis of family videos recorded by parents before diagnosis was made and through which it is possible to have a direct view of the child in the first 2 years of life
3. Population-based early screening and observational diagnostic tools
4. Prospective studies of early development of high-risk siblings of children with ASD.

Research on the early presentation of ASD historically coincides with the continuous growth of knowledge on the social development of the young child. The research emphasis on early detection of ASD has progressively shifted from identifying positive symptoms to defining difficulties in social behaviors. Nonetheless, even if the socio-communicative difficulties are the most important indices for early identification of ASD, data also indicate early presence of repetitive movements [165]. The onset of ASD symptomatology may range from emergence by age 12 months to more commonly between age 18 and 24 months. According to the epigenetic perspective, the growth of the social brain, typically immature at birth, depends on the environment. This perspective has increasingly led to emphasis on early intervention and efforts for alleviation of communicative and social impairments. Early screening and identification efforts and timely implementation of therapeutic interventions have therefore become one of the most priority research topics as they can significantly mitigate the developmental impact of the disorder. Research on the early signs of autism has shown that during the first 3 years of life,

the children with ASD show fluctuation in the organization of their intersubjectivity, particularly in terms of their facial, vocal, and gestural synchrony with reduced frequency, coordination, and dependence on external stimulation. In addition, they present with hyporeactivity, poor eye contact, communicative babbling, pointing, and gesturing [166].

Early identification of ASD and timely implementation of therapeutic interventions are the foremost priority as they can significantly mitigate the developmental impact of the disorder.

16.4.3 Communication and Social Interaction

The first set of core symptomatology that characterizes ASD is the qualitative impairment of communication and social interaction. The delay or plateau in the progression of the development of children's social communication skills, especially in the first and second years of life, remains as the first alarm signal perceived by parents that is also the foremost reason why they seek specialist attention. Communication difficulties are also enhanced by language developmental anomalies. The latter can manifest themselves in various ways along a continuum from total absence to the presence of a well-developed language, with vocabulary that often lacks contextual flexibility and reciprocity. These communication problems emerge both in relation to the comprehension and language production skills and are an obstacle in the overall development of social competence. Problems in understanding are often associated with the literal interpretation of language, in turn linked to the deficit in imaginative and/or abstraction skills and to scarce attention paid to communicative stimuli. The absence of verbal language characterizes about 20–50% of children with ASD, with children acquiring a certain number of words (five or more) between 12 and 18 months of age, with some children undergoing a regression with loss of language as well as other acquired developmental skills.

Parallel to the developmental anomalies of language, difficulties in nonverbal communication are also characteristic in ASD communication deficits. Whereas children with simple delays in development of language show sufficient compensatory communication at a non-verbal level, children with ASD often have reduced or atypical use of nonverbal and pragmatic communication. For this reason, it is fundamental to pay attention to the idiosyncratic ways that each child with ASD uses nonverbal communication. Echolalia, or literal repetition of a word or a group of words heard from an external source, may also affect about 25% of children with ASD. The echolalia can be immediate, if the repetition takes place immediately after listening, or delayed occurring some time from listening. The use of echolalia by the child can be understood in some children with ASD as an expression of a communicative intent, in others as a form of self-stimulation with a reassurance function. The proportion of children with ASD who are "minimally verbal" is estimated to range from 25% to 50% [167, 168]. However, there is no universal agreement on what constitutes minimally verbal communication (i.e. no words, few words, the extent to wich words are used for communication), and different definitions and assessment instruments result in different groupings of children and adults [167, 169].

Children typically learn within their everyday interactions; they understand that their actions can have an effect on others and they learn the meaning of words and function of objects. From the first moments of life, the experiences of the newborn are guided by the actions of adults. The presence of ASD prevents this natural learning process by reducing the child's sensitivity to the most salient social and affective stimuli, such as the baby talk, characterized by spikes and changes in intonation that their parents use when they turn to the newborn during the first months of life. It has been shown that among children with ASD, the ones who are more attune to baby talk develop better language skills than those who do not. A crucial skill in the acquisition of language is joint attention (JA), as characterized by their ability to direct attention toward a common focus, such as activities

looking at an object with carer. JA is acquired around 18 months and is a precursor of language and indication of intentional nonverbal form of communication. Frequently, children with ASD show difficulties in JA that are strictly correlated with a delay in the development of language and symbolic activities. At the base of the development of social competences, there is the ability, already present in the newborn, to preferentially orient itself toward social stimuli. Children with ASD, on the other hand, preferentially orient their attention toward the stimuli of physical–mechanical world characterized by perfect contingencies and an aversion toward unpredictable stimuli. Such unpredictability in paying attention to people, and in particular to faces, is not only the basis for learning and understanding the behavior of others, but is fundamental for the development of emotions, relationships, and attachments. Persons with ASD and/or other neurodevelopmental problems are more likely than the general population to have transgender identity, non-heterosexual sexual orientation, and other gender non-conformities. Clinicians and family members may mistake gender dysphoria for an autistic feature, an uncommon or too concentrated interest. In contrast, if teenage social issues are attributed to gender dysphoria, ASD may go unnoticed.

16.4.4 Interest and Behavior

A second set of core symptomatology that characterizes ASD is the presence of restricted and repetitive patterns of behavior, activities, and interests. Children with ASD appear to be attracted by the details of objects, a particular sound or specific body sensations. They often put into practice stereotyped behaviors, such as flapping their arms repeatedly, rocking their body, moving their fingers in front of their eyes, and walking on their toes. Their propensity to obsessively repeat the same routine of actions, such as putting objects in a row and stacking the cubes, is attributable to a deficit of imagination and abstraction, or to a particular propensity to focus on detail rather than on the globality of the stimulus. Moreover, these behaviors make the child resistant to changes in his environment.

16.4.5 The Anomalies of Reactivity to Stimuli

An important feature of ASD now included in the core symptoms of restricted and repetitive behaviors in DSM-5 is the alteration of sensory modulation. The sensory profile of children with ASD is different from that of children with neurotypical development. These sensory abnormalities are not due to deficits in the visual, auditory or sensory apparatus that are often normal in tests of visual or auditory acuity. Sensoperceptive anomalies represent a possible basis of the central characteristics of the disorder. The majority of children with ASD show significant impairment of the integration and subsequent organization of sensory information with which they come into contact.

The impossibility of developing a direct line with the outside world, and often also with the internal world, negatively affects their general functioning, altering their mode of response to stimuli. The anomalies of sensory responsiveness can be divided into two broad categories: hyper-responsiveness and hypo-responsiveness. The same child can be hypersensitive to certain stimuli and hyposensitive to others. It has in fact been established that these two patterns of sensory response coexist; despite this, hypo-responsiveness appears to be prevalent and specific to autism in early childhood.

The investigations on hypo-responsiveness to social and nonsocial stimuli have shown that the increase in mental age, even more than the chronological age, corresponds to an increase in responsiveness, both in the social and nonsocial context: this may be due to maturation of general processing mechanisms and attention skills, learning skills and/or development of compensatory strategies. The presence of sensory hyper-responsiveness has been associated with a greater likelihood of increasing stress in parents, restrictions in family life, as well as behavioral problems and anxiety in children. These findings underscore the need to assess the practical and psychological impact that sensory symptoms have on

children and their families, to develop even more effective intervention plans.

16.4.6 Mental Functioning

Over the years, various psychological theories have been developed in an attempt to better clarify the mechanisms of mental functioning of ASD. The main models are shown below.

■ Deficit of the Theory of Mind

Baron-Cohen, Leslie, and Frith [170] identified a deficit of the theory of mind (ToM) underlying the difficulty of people with ASD to interact with others and with the outside world, stemming from the inability to attribute mental states to other people. Having a theory of mind is interpreted as a prerequisite for understanding the full range of own and others' mental states (i.e., beliefs, desires, intentions, imagination, and emotions) that cause actions and behaviors based on the reading of these states. The concept of theory of mind is closely related to the more general concept of metacognition, in the sense that, it would be a specific aspect of larger metacognitive capacities acquired during the ontogenetic development.

■ Primary Deficit in Interpersonal Relationships

All children, from birth, are involved in mutual exchanges with others and emotional expressions of their carers form the basis for understanding the mental states of others and for using facial expressions. Hobson [171] has argued that children with ASD are not able to perceive the expressions of emotions of those who care for them. The author believes that these children fail in the competence acquired through the experience of relationships with others and therefore do not learn to recognize and express mental and emotional states by observing adults and sharing such mental states with them. Similarly, Trevarthen [172–174] has postulated that children with ASD display deficits in intersubjectivity during communicative acts. According to Trevarthen, this ability is innate and does not require abstract cognitive abilities nor does it depend on cultural learning. Intersubjectivity is therefore postulated to manifest itself as an immediate natural awareness of the presence of the other, made possible by the production and recognition of body movements, especially of the face, mouth, and hands, adapted to this communicative function in the course of evolution as social beings. The process of intersubjectivity cannot be completely articulated in children with ASD. The first phase of the process, called primary intersubjectivity, is generally experienced by all children with ASD. The phase of secondary intersubjectivity or collaboration, which requires shared attention, is the most compromised and altered. The hypothesis of the primary deficit in the interpersonal relationship has received renewed attention, supported both by the discovery of the neurophysiological systems of empathy and the mirror neuron system and by the observational studies on the primary relationships of children at high risk of developing ASD [175].

■ Deficit of Planning Executive Functions

The term executive functions (EF) refers to the neuropsychological activities that regulate the processes of planning, control, and coordination of cognitive and motor activities. Examples of the main EF are the organization of actions in hierarchical sequences of objectives, the focus of attention on the most useful information for the task to be performed or the inhibition of responses not adequate to external stimuli. The use of executive functions is indispensable in all types of problem solving, not only in the most complicated and abstract ones, such as the solution of mathematical problems, but also in the acquisition of social skills. Despite numerous studies conducted, the impairment of EF in ASD has not yet been clarified, with a persistent finding of high variability by clinical subgroups or even interindividual [176]. The most frequent anomalies concern inhibition, working memory, flexibility, attention, planning, behavioral monitoring with respect to its planning, preparatory processing, fluency (ease or difficulty in performing a task), and concept formation [176, 177]. Executive dysfunctions also seem to depend on gender: males appear to have less marked alterations in working memory, while females in flexibility [178].

Central Coherence Deficit

Persons with typical development show the ability to organize the various stimuli present in the various contexts in a coherent situation, giving meaning to the experience. The hypothesis of a deficit of central coherence, developed for the first time by Frith in 1989 [179] and subsequently taken up by Happé in 2001 [180], is based on the idea that in ASD there is difficulty in the operations of synthesis and integration of cognitive components and affective information. These difficulties would be associated with a marked propensity to focus on detail rather than on the global nature of the stimulus. This problem could explain the social anomalies found in people with ASD, which would be unable to integrate social skills and competences into everyday life. However, this mode of perceptive processing would make people with ASD more skilled in perceptual discrimination by favoring the development of islands of abilities and talents that sometimes characterize them.

Deficit of Mental Simulation and of the Functioning of Mirror Neurons

Difficulties of joint attention, for example, not alternating the orientation of the gaze from the object fixed by an adult to the adult himself, and simulation, for example, not anticipating or inducing the reactions of an adult, are frequently found in children with autism. These functions have been judged in close relationship with each other, with the activity of the so-called mirror-neurons [181], and with that of the other brain areas appointed to corporatized cognition (embodied cognition), or to the integration of higher cognitive functions with the sensorimotor system.

The first mirror neurons identified were those belonging to the motor system and took their name from the ability to activate not only when the individual to whom they belonged performed an action but also when he saw it performed by others. The results of recent research suggest that this function, technically called "mirror mechanism," is managed by complex networks of neurons rather than single cells and that it suffers from numerous contextual variables, such as the familiarity with the observed action, the position in which it is executed, the perspective from which it is observed or the value attributed to it [182–185]. In animals, mirror mechanisms have been identified in many different cortical areas and brain centers [186–188]. The repeated finding in people with ASD of alterations in the functioning of neural networks with mirror mechanisms [189–195] seems to support the hypothesis that the disorder is caused by a defect in the motor representation necessary to simulate the mental state of others.

Irrespective of many theories that have been postulated to clarify the mental functioning of children with ASD, the delay or plateau in the progression of the development of their social communication skills, especially in the first and second years of life, remains as the first alarm signal perceived by parents that is also the foremost reason why they seek specialist attention.

16.5 Psychiatric Comorbidity in ASD

The psychiatric comorbidity in children, adolescents, as well as adults with ASD, is a subject of critical scientific importance, affecting their quality of life, prognosis, and functional outcomes. The prevalence of psychiatric conditions in persons with ASD vary considerably according to factors such as setting, sample characteristics and diagnostic methods used. Many studies reference clinical or treatment samples, those in treatment in tertiary care or rehabilitation centers, as well as subjects enrolled in clinical trials and cross-sectional studies. Such samples carry a potential for overestimation of both the frequency and severity of psychiatric comorbidity. Nevertheless, a better understanding of the relation between diagnostic constructs, prognostic insight, and preventive as well as treatment approaches in ASD can be gained in the assessment of population-based psychiatric comorbidity. Furthermore, diagnosis of co-occurring psychiatric disorders is also challenging due to several difficulties including the impact of ASD symptoms and behaviors on the assessment procedures, communica-

tion deficits, atypical presentation of psychiatric symptoms, and scarcity of standardized diagnostic tools [196]. Moreover, the clinical manifestations of ASD often overlap with the symptoms of other disorders; thus, it can be difficult to distinguish between them [197–199]. The prevalence estimates of co-occurring psychiatric conditions therefore vary widely, and improved estimates and identification of moderators are needed to improve care [200].

16.5.1 Prevalence of Psychiatric Disorders

Co-occurring psychiatric conditions are more frequent in the ASD population compared to the general population. The prevalence reported in most studies varies from 70% to 72% [201–203] and about 40% of persons with ASD may have two or more comorbid psychiatric disorders [204]. Individuals with ASD are likely to experience a higher prevalence of common mental disorders compared to the general population [205]. A recent comprehensive systematic review and meta-analysis conducted by Lai et al. [200] identified eight co-occurring psychiatric diagnoses most reported in the literature among people with ASD, and indicated that the overall pooled point prevalence figures varied between 4% and 28% [200]. Anxiety and attention-deficit/hyperactivity disorder (ADHD) are the most prevalent psychiatric disorder in children as well as adults with ASD. Recent finding showed that older age was associated with lower prevalence of ADHD and higher prevalence of depressive, bipolar, and schizophrenia spectrum disorders than younger age [200]. Studies with a high proportion of female participants report higher prevalence of depressive disorders. Studies with a high proportion of people with intellectual disability report higher prevalence of schizophrenia spectrum disorders than those with a low proportion of people with intellectual disability did.

In contrast, report from the Simons Simplex Collection (SSC), a deeply phenotyped sample of more than 2800 individuals with ASD that includes only simplex cases, that is, lack of another affected family member closer than first cousins, have shown higher rate of family history of psychiatric disorders (including bipolar disorder and schizophrenia) among higher functioning cases of ASD with higher IQ scores [206].

▪ Anxiety Disorders

The most recent studies on adults with ASD estimate that rates of co-occurring anxiety range from 17% to 27% [200, 202, 207, 208]. In addition, the prevalence of anxiety disorder was highest among adults with autism who did not have a comorbid intellectual disability, although this figure may be skewed due to the inadequacy of the diagnostic criteria utilized [208]. Clinically significant anxiety is common in individuals with ASD and is related to increased psychosocial and familial impairment [209]. People with ASD and comorbid anxiety may show increased ritualistic behaviors and PBs [210]. However, anxiety disorders can be difficult to diagnose in this group, due to difficulties in expressing worry and fear. Study found that individuals with ASD, especially high functioning, are likely to suffer from social anxiety, which may contribute to the avoidance of social interaction typically seen in this population. An important difference between social anxiety disorder and ASD is the presence in this latter group of social awkwardness [211].

▪ Mood Disorders

Studies on prevalence of psychiatric disorders in adults with ASD have identified high rates of mood disorders [212–214]. A recent systematic review reported a pooled prevalence of mood disorders at about 19% [202].

Depression spectrum disorders were the most frequent mood disorders described throughout the studies when considering the specific categories, with prevalence ranging from 11% to 23% [200, 207, 212]. The difficulty in detecting depression in this population is due to problems in communicating and expressing feelings of sadness, hopelessness, low self-esteem, guilt, or suicidal ideation [215]. Furthermore, individuals with ASD often show flat or constricted affect, so that changes associated with the onset of depression may not be identified [196]. Depression in people with

ASD is characterized by a variety of symptoms, including decreased interest in favorite activities, worsening in self-care capabilities, and decline in personal hygiene [216, 217]. In other cases, stereotyped and repetitive behaviors may intensify [218], and there may be worsening of problem behaviors (PBs) such as agitation, aggression, or self-injury [217, 219]. Also, neurovegetative symptoms as changes in sleeping patterns, appetite, and weight are common, especially in those with lower verbal abilities [196].

Bipolar disorder presentation in ASD may give unique challenges depending on several factors, such as age, the subtype of bipolar disorder, the subtype of ASD, the presence of intellectual disability, and the presence of concurrent psychiatric and medical comorbidity. In persons with low-functioning ASD, aggressive behavior toward others, irritability, and hyperactivity may be more common during elevated to hypomanic mood phases, whereas loss of appetite, associated weight loss, sleep disturbance, and decreased communication may be more common during the depressed phase [220, 221]. In addition, during phases of elevated moods, pressured speech or phonations, laughter, and intrusiveness are notable including sexualized impulsive behaviors. Bipolar disorder has been more frequently reported among adults with ASD, although prevalence rates are not consistent across studies especially towards persons living in the community. Some clinic-based studies have suggested high rates of up to 21% [202, 222], with Lai and colleagues reporting a rate around 5% [200]. In prototypical bipolar disorder, the symptoms related to mood dysfunction are severe in its expressive range, with euphoria, irritability, and sadness. With disruptive mood dysregulation states, the reactivity to events and provocations vary, as well as levels of physical energy levels, sleep, and appetite disturbance.

■ Obsessive-Compulsive Disorder

The prevalence estimates for obsessive-compulsive disorders (OCD) co-occurring in ASD range from 9% to 22% [200, 207, 223]. Rigid thinking and stereotypical behaviors are commonly observed in ASD and overlap with OCD in the symptomatic profile [224]. So it can be difficult to distinguish symptoms that are related to a comorbid OCD, particularly in people with lower functioning levels. However, studies have indicated that rigid and repetitive thoughts and behaviors in ASD differ from those of the OCD [225, 226]. In OCD, typical obsessive themes include worries about germs, harm coming to self or others, distinguishing right from wrong, and guilt about morals, or religiosity. Therefore, typical compulsions include ritualized washing, checking, ordering, apologizing, or mental rituals such as counting or praying, whereas people with ASD show a pervasive need of sameness and perform repetitive or ritualistic behaviors such as arranging, counting, or touching/tapping [227]. However, people with ASD are not likely to think about their rituals and may present with limited insight into the reason behind their ritualized behaviors. People with ASD are also less likely to be using their ritualized behaviors to neutralize fear or anxiety as would be true for OCD. Rather, ritualized behaviors and repetitive thoughts may satisfy other needs, such as modifying sensory input, knowing what is going to happen next in their daily routine, controlling and gaining reinforcement from the environment, or preserving routine and identicalness in their daily lives.

■ Psychotic Disorders

Recent meta-analyses reported prevalence rates of psychotic disorders ranging from 4% to 12% among adults with ASD. When considering the specific categories, Schizophrenia was the most frequent reported throughout all the studies [200, 202]. Transition into adulthood and exposure to stressful events are related to the onset of brief psychotic episodes in adults with ASD. Furthermore, psychotic symptoms are often sign of an underlying mood disorder or a schizoaffective disorder [199].

■ Eating Disorders

Of the 5381 adolescents with autism traits in the Avon Longitudinal Study of Parents and Children (ALSPAC) answering reporting on their eating habits at age 14, nearly 8% had engaged in some form of disordered eating monthly, and almost 3% did so weekly, with female adolescents three times more likely to

report such behaviors as males. Furthermore, adolescents with disordered eating habits had more autism traits at ages 7, 11, and 14, and greater the loading of autism traits, the more frequent was the prevalence of eating disorders [228]. Furthermore, adolescents' eating disorders predict psychiatric high-risk behaviors in young adulthood [229].

The prevalence of eating disorders among adults with ASD ranged from 1.4% to 7.9% across recent reviews [202, 230]. The specific category more prevalent is anorexia nervosa [230]. Eating disorders are often reported among adults with ASD, particularly pica, food refusal, and food selection for color, type, or texture. Selective food refusal is also associated with food spitting, obsessions, and rituals in ASD [231, 232]. Persons with ASD and eating disorders, especially anorexia nervosa, have some common features concerning cognitive style and behaviors [233], such as the tendency to focus on details and the insistence on sameness [234].

■ Posttraumatic Stress Disorder

Reported prevalence estimates for posttraumatic stress disorder (PTSD) ranges from 3.6% to 40% [200, 235]. Although negative life events, physical and sexual abuse are quite common in people with ASD, studies are lacking. Furthermore, the range of life experiences that are perceived as traumatic appears to be significantly greater than in the general population [235, 236]. Traumatic events and clinical presentation of PTSD are difficult to detect in people with ASD, especially in view of difficulty in defining their inner psychic state [237]. Diagnosis of PTSD requires an accurate assessment to understand cognitive interpretation, PBs, emotional disorders, and recognize signs of increased physiological arousal and sensory processing of the traumatic events, which might otherwise be misdiagnosed as exacerbation of ASD symptoms.

■ Attention-Deficit Hyperactivity Disorder

Attention-deficit/hyperactivity disorder (ADHD) is the most frequent psychiatric diagnosis found in adults with ASD. The pooled prevalence derived from the most recent meta-analyses ranges from 25% to 28% [200, 202]. Although ASD and ADHD have distinct clinical symptoms, they show overlapping features. Inattentive, hyperactive, and impulsive symptoms are frequently reported in persons with ASD and on the other hand, impairments in social communication, as well as repetitive and stereotyped behavior, have been described in person with ADHD. In addition, there is behavioral, biological, and neuropsychological overlap between the two disorders. Below-threshold cross-disorder symptoms are also common, that is, having symptoms of the other disorder despite not having the diagnosis [238]. Missing a diagnosis of ADHD not only leads to inadequate care but can also result in inadequate treatment and poor outcomes [239]. While there is a very high rate of comorbidity of ASD and ADHD, caution is needed in prescribing stimulants to children with ASD that may lead to increase in irritability, and careful documentation of adverse effects is paramount.

> The prevalence estimates of co-occurring psychiatric conditions vary widely across studies and improved estimates and identification of moderators are needed. Anxiety and ADHD are the most prevalent psychiatric disorders in children as well as adults with ASD. Recent finding showed that older age was associated with lower prevalence of ADHD and higher prevalence of depressive, bipolar, and schizophrenia spectrum disorders than younger age subjects with ADHD.

16.6 Specific Assessment

ASD is highly stable over the lifetime and is often associated with a poor overall outcome and high mortality rates [240, 241]. Diagnostic clarification of ASD is crucial in order to align targeted treatment and support and to avoid overmedication due to diagnostic misclassification. In accordance with the United Nations Convention on the Rights of Persons with Disabilities and the Charter for Persons with Autism adopted by the European Parliament, a right to diagnostic clarification and treatment assistance applies across the entire autistic spectrum, that is, including people with additional intellectual disability. Due to the lack of

distinct biological markers, the diagnosis of ASD is based on the course of development and the individual's current symptomatology [242]. Diagnostic criteria are defined in the current diagnostic manuals of the American Psychiatric Association (DSM-5, [161]) and the World Health Organization (ICD-10, [243], from 2018 ICD-11 [164]). In the ICD-11, ASD (6A02) is incorporated in the chapter on neurodevelopment disorders [164].

The WHO has long advocated for complementing the clinical diagnosis (ICD) with a functioning profile as attainable through the WHO International Classification of Functioning Disorders (ICF, [244]). The ICF marks a paradigm shift in the way health and disease are understood and recorded. A comprehensive set of different aspects of functioning, including biological, psychological, social, and environmental aspects, offers the possibility to precisely describe the support needed for the various activities of daily living (see Chap. 1). Current ASD research aims to outline clusters of characteristics in "core sets" incorporating strengths and potentials [245]. This refines the level and type of support and might also impact future diagnostic assignments.

16.6.1 Diagnostic Challenges

Neither ASD nor intellectual disability is a discrete disorder, but rather presents as syndromes with a multitude of symptom compositions [246]. As a result, diagnosing ASD in adults with intellectual disability is challenging, and the availability of evidence-based standardized measures is limited. Several issues impede the assignment including tendency for diagnostic overshadowing, attribution of the symptoms to intellectual disability, or ASD rather than comorbidity, and in more recent years, diagnostic substitution of intellectual disability by ASD alone [247]. For many adults with limited contact with their relatives, obtaining a complete medical history is unlikely. Physical conditions, such as hearing or vision impairments, may further complicate the differential diagnoses [248]. According to the ICD-11, ASD may be diagnosed in individuals with intellectual disability "if deficits in initiating and sustaining social communication and reciprocal social interactions are greater than would be expected based on the individual's level of intellectual functioning and if the other diagnostic requirements for ASD are also met. In these circumstances, both ASD and the Disorder of Intellectual Development should be assigned and the 'with Disorder of Intellectual Development' qualifier should be applied with the ASD diagnosis. Because ASD inherently involves social deficits, assessment of adaptive behaviour as a part of the diagnosis of a co-occurring Disorder of Intellectual Development should place greater emphasis on intellectual functioning and the conceptual and practical domains of adaptive functioning rather than on social skills. The diagnosis of ASD in individuals with Severe and Profound Disorders of Intellectual Development is particularly difficult, and requires in-depth and longitudinal assessments. However, the diagnosis may be assigned if skills in social reciprocity and communication are significantly impaired relative to the individual's general level of intellectual ability" [2]. Further information on the differential or co-occurrent diagnosis of ASD in persons with intellectual disability are provided later on in the chapter.

Another relevant diagnostic challenge is related to cultural variation in norms of social communication, reciprocal social interactions, as well as interests and activities. The ICD-11 states that "signs of impairment in functioning may differ depending on cultural context. For example, in some societies it may be normative for children to avoid direct eye contact out of deference, which should not be misinterpreted as impairment in social interaction" [2].

16.6.2 Diagnostic Assessment

Early developmental history is crucial for correct diagnostic classification. In the first 6 months, developmental indicators include absence of social smile or joyful expressions directed to people, limited or no eye contact, and inappropriate eye gaze. Within 9 months, additional signs are absence of vocalizations or other nonverbal communication, while within

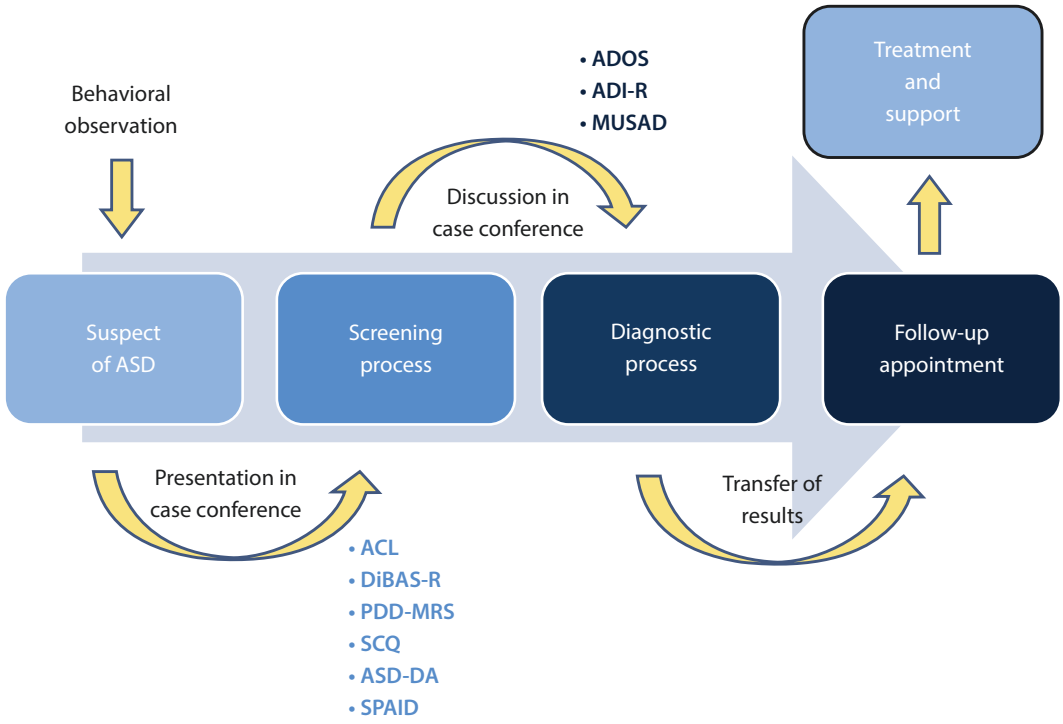

Fig. 16.2 Three-step diagnostic process for ASD

12 months, absence of babbling and communicative gestures. For example, reaching, pointing, and response to name are relevant. In the second year of life, main symptoms refer to lack of verbal communication, with absence of words within 16 months and no meaningful or two-word phrases within 24. Loss of any previously acquired speech, social skills, babbling, or deficit in social interactions at 2–3 years are clear indicators [249–251].

Also peculiarities in frequency and other structural parameters of the cry, as well as repetitive games, repetitive movement with objects, repetitive posturing of body, arms, hands, or fingers, repetitive language, selective feeding, lack of sharing interest or enjoyment, unusual prosody (little variation in pitch, odd intonation, irregular rhythm, or unusual voice quality) may indicate the presence of an ASD in early childhood [250, 252].

Beyond the information listed above, further aspects to investigate are the presence of neuropsychiatric disorders in parents or relatives, pregnancy course, birth, age of language development, age and way of walking, age of sphincter control, educational ability and need of educational support, sensory issues, self-harm, other medical and neurological disorders in childhood [253].

In adulthood, the NICE guidelines recommend a stepwise process for suspected ASD [254]. Initial assessment may be supported by the application of screening instruments such as the Autism-Spectrum Quotient (AQ) questionnaire [255], the Social Communication Questionnaire (SCQ) [256]or the Scale of Pervasive Developmental Disorder in Mentally Retarded Persons (PDD-MRS: [257]). If suspicion of ASD is affirmed, a more comprehensive assessment should be conducted by trained professionals. The assessment should examine the core symptoms of ASD, early developmental history, behavioral problems and adaptive functions, past and current physical and mental disorders, other neurodevelopmental conditions and sensory peculiarities. The results of the diagnostic process ought to be provided to the affected person, family members, and care team and support needs addressed. Figure 16.2 shows the three-step diagnostic process for ASD.

Tip

"Although diagnosis is an important aspect of most assessments, the focus of assessment should not only be on diagnosis but should also consider the person's physical, psychological and social functioning, and any risks that they might face. Crucial to the effective delivery of any assessment is the competence of the staff who is delivering it, including the ability to conduct an assessment, interpret the findings of the assessment and use these findings to support the development of appropriate care plans and, where necessary, risk management plans" ([258], p.118).

No single diagnostic procedure can solely define the diagnostic decision. The combination of all relevant present and past information provided by several team members and carers and the results of structured tools, interviews, and behavioral analyses by trained experts provide a basis for the diagnostic decision. Due to the complexity of the ASD, a range of professions and skills have to be part of the diagnostic process in order to use a wide range of resources to obtain diagnostically relevant information [258] supported by interdisciplinary conferencing and approaches to increase knowledge and capacity of the care team [259].

16.6.3 Diagnostic Instruments

The following screening scales have been validated for assessment of ASD in persons with intellectual disability:

- The *Autism Spectrum Disorder – Diagnosis Scale for Intellectually Disabled Adults* (ASD-DA: [260]) is a structured interview that is used to screen for ASD in adults with intellectual disability. It is administered to direct care staff who have known the person for at least 6 months. The staff member have to compare the person to similar persons living in the community.
- The *Social Communication Questionnaire* (SCQ) [256] – current or lifetime versions validated for adults with intellectual disability [261]. The *Social Communication Questionnaire for Adults with Intellectual Disability* (SCQ-AID) [262] is specifically tailored for adults with intellectual disability and consists of a core set of valid and adult-appropriate items.
- The *Autism Checklist* (ACL) [263] is a screening tool based on the ICD-10 research criteria. It facilitates a structured medical history and assessment in medical visits.
- The *Diagnostic Behavioral Assessment for ASD-Revised* (DiBAS-R) [264] is an ICD-10/DSM-5-based caregiver-report screening tool that consists of 19 Likert-scaled items validated in an independent sample [265].
- The Systematic Psychopathological Assessment for persons with Intellectual and Developmental Disabilities (SPAIDD) is an Italian tool package for diagnostic assessment in adults with intellectual disability. It consists of a general form and specific checklists for certain disorders, including ASD [266, 267].
- The *Scale of Pervasive Developmental Disorder in Mentally Retarded Persons* (PDD-MRS) [268] is a structured interview to be conducted by a qualified and experienced rater with a professional caregiver evaluating ASD-related problem behaviors in situations of daily life. It consists of 12 categorical items [257].

The following validated psychometric measures can be applied in the second step to diagnose persons with suspected ASD:

- The *Autism Diagnostic Interview - Revised* (ADI-R: [269]) is a semi-structured parent interview assessing social reciprocity, communication, and restrictive, repetitive behaviors between the ages of 4 and 5 years. It consists of 93 Likert-scaled items, and a selection of these is adopted by the final algorithm, resulting in a classification of 'autism' or 'no autism'. While showing good psychometric characteristics, the ADI-R is only applicable in about one-third of adults with intellectual disability [270].
- The *Autism Diagnostic Observation Schedule* (ADOS: [271]) is a semi-structured observation tool to assess social communication in persons suspected of having

ASD. Depending on their verbal abilities, one of four modules can be applied. It has been indicated to be applicable for some adults with intellectual disability, and for better discrimination, revised algorithms were proposed [270, 272]. In 2015, an update of the instrument has been produced, namely ADOS-2 [273].

- The *Music-based Scale for Autism Diagnostics* (MUSAD) [274, 275] was specifically developed for people with intellectual disability. In standardized musical settings, the quality and level of social interaction are prompted and assessed by the examiner. Due to its nonverbal interactive quality, it is particularly suitable for nonverbal individuals [276].
- The *Gilliam Autism Rating Scale* (GARS; [277]) is an interview used to assess ASD comorbidity with moderate–severe intellectual disability, when the cognitive impairment does not allow the patient to collaborate during the clinical evaluation. An updated GARS-3 has been published in 2013 [278].
- The *Social Responsiveness Scale - Second Edition* (SRS-2) [279] is a functional evaluation that can be used from childhood to adulthood. It identifies the presence and severity of social impairment and differentiates it from that occurring in other disorders.

In childhood, most common screening test are the following:

- The *Checklist for Autism in Toddlers* (CHAT) [280] assesses 18-month children and can be used also in the periodic health reports in pediatric settings. It includes 9 questions to parents and the direct observation of 5 target behaviors: eye contact, joint attention, play of fiction, and indication and ability to build a tower with cubes.
- The *Modified-Checklist for Autism in Toddlers* (M-CHAT) [281] can be used with children from 18 to 24 months, and it is comprised of 23 items. The 2009's version includes a revision with follow-up (M-CHAT-R/F) [282] which can be used between 16 and 30 months.
- The *Quantitative Checklist for Autism in Toddlers* (Q-CHAT) [283] consists of a 25-item questionnaire for parents, in which they are asked to quantify how many times their child has a particular behavior on a 5-point scale (0-4). CHAT, M-CHAT, and Q-CHAT are used to assess siblings of children with ASD or children born preterm [284].
- The *Childhood Autism Rating Scale* (CARS) [285, 286] is a 15-item measurement scale which evaluates, through direct observation and interview, social interactions, verbal communication, and behaviors starting from 24 months. The scores obtained also provide a severity level of the symptoms and allow an assessment of change over time. The standard version (CARS2-ST) is used for children up to 6 years or older with IQ scores 80 or below or children with established communication difficulties; the high-functioning version (CARS2-HF) is used for children 6 years or older with IQ scores above 80. The CARS 2 also contains recommendations on how to orientate treatment in each area.
- The *Checklist for Autism Spectrum Disorder* (CASD) [287] is composed of 30 items, based on the DSM-5 diagnostic criteria. It can be used in children and adolescents, regardless of the cognitive level and severity of symptoms.
- The *Gilliam Autism Rating Scale* (GARS) [277] is a checklist for parents, teachers and professionals based on DSM-IV diagnostic criteria. It allows both the identification and the estimate of the severity of the symptoms in subjects from 3 to 22 years. It is also useful to focus on the objectives of the enabling and educational interventions, as well as to document their efficacy. The third DSM-5 edition has also been updated [278].
- The *Gilliam Asperger's Disorder Scale* (GADS) [288] is specific for children with high-functioning ASD.
- The *Krug Asperger's Disorder Index* (KADI) [289] is a screening instrument for Asperger Syndrome, also useful for developing goals for intervention. There are two forms, covering 6–21 years.
- The *Childhood Asperger Syndrome Test* (CAST) [290] is a 37-item parent questionnaire designed to screen for high-functioning ASD that focuses on social behaviors and communication.

In addition to these tools, behavioral analysis of video recordings from everyday situations can provide insights into social interactions or stereotypical behaviors in a familiar environment with familiar persons.

The NICE guidelines do not consider specific instruments for routine use in the assessment of adults with intellectual disability and suspected ASD. However, the ADOS and the ADI-R may be of value in augmenting diagnostic assessment in some adults with intellectual disability, mostly those with milder cognitive impairment.

A useful complement to the diagnostic measures is the assessment of functioning and adaptive skills, which is commonly through the following tools:

- The Psychoeducational Profile- Third Edition (PEP-3) [291]
- The Psychoeducational Profile Revised (PEP-R) [292]
- The Vineland-II Adaptive Behavior Scales (VABS-II) [293]
- The Verbal Behavior Milestones Assessment and Placement Program (VB-MAPP) [294]
- The TEACCH Transition Assessment Profile (TTAP) [295]
- The Essential for Living scale [296].

16.6.4 Differential and Co-occurrent Diagnoses

The diagnostic process is further challenged by the fact that other neurodevelopmental disorders as well as physical and mental ill-health are frequent in persons with ASD. Although biological or genetic tests, as well as neuroimaging, should not be considered routine in the diagnosis of ASD [242, 297], they can be used to rule out other suspected conditions.

Intellectual Disability

Social communication deficits as well as stereotyped behaviors not caused by ASD are frequently observed in persons with intellectual disability. The deficit in social communication has to exceed the level of impairment which can be expected for a certain severity of intellectual disability to allow for additional diagnosis of ASD. According to Sappok, Matson and collaborators [270, 298], social reciprocity and non-verbal communication are especially valuable to distinguish between ASD and intellectual disability. Symptom presentations may change over the lifespan which can further increase the challenges of diagnosing ASD. Moreover, susceptibility to stress may lead to regressive behaviors further increasing the diagnostic difficulties. In those cases in which the differential diagnosis or the co-occurrence of intellectual disability and ASD has to be defined, a comprehensive cognitive assessment is indicated, using Wechsler Preschool and Primary Scale of Intelligence-III (WPPSI-III, [299]), Wechsler Intelligence Scale for Children-IV (WISC-IV, [300]), Wechsler Intelligence Scale–IV (WAIS-IV, [301]), Leiter International Performance Scale – Revised (Leiter-R, [302]), Raven Progressive Matrices (RPM, [303]), or Coloured Progressive Matrices (CPM, [304]), according to the different ages and individual characteristics.

Behavioral Phenotypes in Genetic Syndromes

Genetic syndromes may be associated with ASD. In tuberous sclerosis, especially type 2, interaction and communication as well as attention and impulse control are impaired, while ritualized and stereotypic behaviors can be observed less frequently. Fragile X syndrome is the most common monogenetic cause of intellectual disability. Many of its sufferers, who are predominantly male, show hyperactivity, deficits in attention and self-regulation and repetitive behaviors. Moreover, social anxiety and shyness in eye-contact may resemble ASD at first glance, while interest in social relations is unaffected.

Rett syndrome affects mainly girls. While development initially proceeds in a typical way, skills are progressively lost and head growth decelerates after the first year of life. Although presenting with pronounced stereotypical hand movements, loss of expressive language and rigid, poorly modulated eye contact, affected persons continue to show an interest in social interaction and seek attention. Therefore, knowledge about the underlying genetic syndrome and the associ-

ated behavioral phenotypes are supportive to prevent psychiatric misdiagnoses [305].

In Down syndrome, a higher rate of ASD is observed rather than in general population. In persons with this syndrome a complete neuropsychological assessment is recommended in adulthood to screen for dementia. Several genetic syndromes could lead to ASD and psychosis (as Del22q11 or Di George syndrome) or to different clinical phenotype in the same family (psychotic or ASD or personality disorder or mood disorders) [306, 307]. Further information on the characteristics of main genetic syndromes associated with ASD are provided in the paragraph 3.1.

Other Mental Disorders

ASD ought not to be misclassified with schizophrenia [308, 309]. Developmental history and careful appraisal of symptom phenomenology are necessary [310]. Abnormalities in attention, impulsivity, and physical hyperactivity that are frequently encountered in persons with ASD, especially in the most severe part of the spectrum, are shared with ADHD. However, as specified in the ICD-11, "individuals with ADHD do not exhibit the persistent deficits in initiating and sustaining social communication and reciprocal social interactions or the persistent restricted, repetitive, and inflexible patterns of behaviour, interests, or activities that are the defining features of ASD. However, ASD and ADHD can co-occur and both diagnoses may be assigned if diagnostic requirements are met for each" [2]. Disorders such as social phobia, depressive disorder, or avoidant personality disorder may show symptoms which may resemble ASD, as these disorders are often associated with social withdrawal, but a different age of symptom onset can be observed. Moreover, catatonia and tic disorders, which may be associated with psychomotor abnormalities may also need to be considered.

Sensory Impairments

Serious sensory impairments such as blindness or severe hearing impairment can also evoke autism-like symptoms. However, in these cases, social behavior is appropriate considering the existing sensory deficits.

Medical Assessment

Various physical and genetic disorders can co-occur in person with ASD with intellectual disability, especially endocrine, metabolic, and immunologic issues, which necessitate haematology tests including organic acid, plasma amino acids, ammonia, ferritin, copper, and porphyria. Moreover, epilepsy is a common comorbidity [311]. An immune dysregulation may be involved in tic symptoms in ASD, as in PANDAS syndrome [312, 313]. In persons treated with atypical neuroleptics, it is important to monitor blood glucose, cholesterol, triglycerides, lipase, liver function tests, thyroid function tests, and prolactin level. Among patients with limited nutritional intake, diet should be augmented with folate, B12, iron, ferritin, and D vitamin.

Deprivation and Hospitalism

ASD and intellectual disability have been associated with deprivation and hospitalism, especially in early childhood. Persons with ASD and intellectual disability are also exposed to emotional or sexual abuse and may have reactive attachment disorder (RAD) and PTSD that may further complicate social withdrawal, anxiety, stereotypies, as well as somatic preoccupations.

16.7 Treatment

16.7.1 Pharmacological

Although there is no evidence for the effectiveness of any particular drug on the core symptoms of ASD, pharmacotherapy is an important intervention within a person-centered treatment plan, to address associated problem behaviors as well as co-occurring psychiatric disorders [314]. It is reported that around 45% of people with ASD are on psychotropic medications [315, 316], the rate increasing with advancing age [317]. Polypharmacy is frequent [318] and often not prescribed appropriate to psychiatric diagnoses. Primary target behaviors include irritability, aggression, self-injury, temper tantrums, anxiety, and associated attention-deficit hyperactivity disorder (ADHD) symptoms.

Pharmacotherapy for people with ASD alone or with co-occurring intellectual disability should be part of an integrated person-centered multidisciplinary planning/intervention [258, 319–321]. It is also suggested that pharmacological treatments should be considered for severe challenging behavior after other nonpharmacological interventions have failed. Drugs should be used at the lowest possible dose and for the minimum duration, with nonmedication-based management strategies and the withdrawal of medication being considered at regular intervals [320]. Most commonly prescribed medications, in order of frequency, are antipsychotics, antidepressants, antiepileptics (AEs), and psychostimulants, many of them without definitive studies guiding their usage.

16.7.1.1 Antipsychotics

There is strong research evidence to suggest that some antipsychotics (APs) may be beneficial for the treatment of aggression, irritability, hyperactivity, self-injury, and repetitive behavior in people with ASD [322, 323]. The current role of first-generation antipsychotics (FGAs) is limited due to the high risk of severe side effects, which include cognitive deterioration, extrapyramidal symptoms, acute dystonia, withdrawal-emergent dyskinesia, and tardive dyskinesia. Because of this, second-generation antipsychotics (SGAs) are more commonly used now in people with ASD and/or other neurodevelopmental disorders. Risperidone and aripiprazole have undergone many studies, while there is little high-quality research available on most of the other SGAs.

In general, the use of APs needs some specific cautions in addition to the usual "go slow and stay low" rule valid for any psychotropic drug in people with ASD and/or intellectual disability. People with ASD, particularly with comorbid intellectual disability, are inclined to develop catatonic symptoms which can be precipitated by APs. Epilepsy can affect up to 22% of people with ASD. APs can lower the seizure threshold, alter EEG, induce, unmask, or aggravate seizures. Therefore, while prescribing APs for a person with a high risk of seizures, a concomitant use of AEs may need to be considered [324].

Some AEs (i.e., phenytoin, carbamazepine, and barbiturates) induce the metabolism of some APs such as quetiapine by the enzyme CYP 3A4, which reduces APs' blood levels. Valproic acid may be an inhibitor and/or inducer of clozapine and olanzapine, an inhibitor of paliperidone, and a weak inducer of aripiprazole. Also, for other AEs which do not seem to have a major impact on APs metabolism, individual differences of effect have to be carefully considered, especially for drugs which have a complex metabolism by multiple enzymes [325, 326]. APs do not seem to have major influence on AEs metabolism and do not seem to affect their blood levels [327].

APs and AEs may also reciprocally potentiate their side effects. Additive sedation with APs seems to be relevant for many AEs, while weight gain is particularly evident for olanzapine in combination with valproate, pregabalin, gabapentin, and carbamazepine. Carbamazepine increases the risk of agranulocytosis of clozapine and valproate can lead to leukopenia when taken with clozapine or olanzapine.

Many people with ASD may also be more vulnerable than the general population to brain volume changes induced by APs. In fact, ASD has been associated with complex alterations in lifetime trajectories of several brain regions and gray matter volumes which underpin social-cognitive and motor functions [328, 329]. Also, studies on the general population receiving long-term treatment with APs suggest that these drugs may determine regional brain volume changes over time, with FGAs in particular being related to neurotoxic effect and global gray matter volume reduction [330, 331]. In elderly adults of the general population and in patients with dementia, the use of APs, especially FGAs, has also been reported to increase the risk of cerebrovascular incidents [332]. These side effects deserve special consideration in persons with co-presence of intellectual disability or genetic syndromes including premature brain aging and early-onset dementia, such as Down syndrome. Unlike FGAs, SGAs have been reported to exert measurable neuroprotective effects, ranging from preventative to restorative, mediated via multiple molecular mechanisms, which are often dose-dependent [333].

Persons with ASD are also at a higher risk of weight gain and associated metabolic disturbances attributed to a number of factors, including selectivity for unhealthy high-calorie foods, excess food intake, sedentary habits, sleep problems, genetics, gut microbiome, endocrine issues, and developmental risk factors. This vulnerability has to be carefully considered when prescribing and using APs, especially SGAs, which are known to increase the risk of hyperglycemia, peripheral resistance to insulin, type II diabetes, dyslipidemia, and weight gain [334–336]. Further attention is needed in elderly people with intellectual disability, in whom the use of SGAs has been associated with an increased risk of cardiovascular morbidity and mortality [337, 338]. For all the above-mentioned safety issues, APs should only be used with specific indications, as part of a comprehensive treatment program, under specialist supervision, and where other measures prove insufficient.

First-Generation Antipsychotics (FGAs)

Among the FGAs, the most used are chlorpromazine and haloperidol. Although some efficacy has been demonstrated in the management of problem behavior (PB), especially aggression, FGAs should not be considered as the first-choice drugs due to tolerability limits [339, 340]. For the same reason, they should be used at low dosage and for the minimum possible time.

Chlorpromazine

Chlorpromazine is often chosen for its broad receptor binding profile, which includes 5-HT2A receptors, and for sedative properties, despite the lack of evidence of its efficacy [341]. Chlorpromazine and clozapine have a relatively high epileptogenic potential, higher than the other APs [342]. Thus, caution should be taken in individuals who have a history of epileptogenic conditions, such as epilepsy, brain alterations, or brain damage.

Haloperidol

Haloperidol is indicated as short-term adjunctive therapy for the management of psychomotor agitation, excitement, and impulsive and violent behavior [343] in people with intellectual disability. Few observations and small open-label studies have shown efficacy also on hyperactivity, aggression, stereotypies, emotional lability, and temper tantrums [344, 345], although a more recent study on adults with intellectual disability and aggressive behaviors showed no statistically significant difference among risperidone, haloperidol, and placebo [346]. In persons with ASD multiple studies have found haloperidol to be efficacious for improving a variety of behavioral symptoms [322]. Sedation is the most common side effect, followed by motor symptoms, which occur in up to one-third of treated patients and include acute dystonia, withdrawal dyskinesia, and tardive dyskinesia. Another serious side effect is neuroleptic malignant syndrome, which seems to have a higher prevalence in people with ASD and/or intellectual disability than in the general population [347–350].

Second-Generation Antipsychotics (SGAs)

The SGAs have a better tolerability profile and a greater number of randomized controlled trials (RCTs) to support their use [351]. They show a complex receptor binding profile, which seems to have a lower impact on the cognitive and neuromotor vulnerability of many persons with ASD and intellectual disability [352].

Risperidone

Risperidone is included in the largest number of studies in ASD and intellectual disability including RCTs, most of which are aimed at exploring its effectiveness on disruptive and aggressive behavior [353–360] (see review by [361, 362]). In people with ASD and intellectual disability, risperidone has been shown to reduce severe PB such as aggression, anger, and self-injurious behavior by around 50% in approximately half of cases [363].

Short-term RCTs showed a significant reduction in irritability compared with placebo, with positive effects on overall functioning. In persons with low-functioning ASD, effectiveness on PB and adaptive skills seems to be dose-related [364] and to increase while associated with educational-rehabilitation interventions [365]. Some data on drug discontinuation show a relative maintenance of the effect even in the long term, with an increase of time to relapse [357, 366].

Side effects are frequent and, if not managed, can be a reason for treatment discontinuation. The most common are sedation (37–72%), especially in the initial phase of treatment [358, 367, 368], weight gain (up to 10% body mass index increase after 6 months) [367], and hyperprolactinemia [369, 370], which can in turn lead to amenorrhea and hypogonadism [371]. Among SGAs, risperidone shows the greatest risk of induction of extrapyramidal symptoms, especially tardive dyskinesia and parkinsonism [346, 372, 373]. Other side effects are lowering of seizure threshold, asthenia, dizziness, sialorrhea, hyperglycemia, and hyperlipidemia [343].

Paliperidone

Paliperidone is the primary active metabolite of risperidone. It offers some advantages over risperidone, especially for orthostatic hypotension. Nevertheless, RCTs supporting their effectiveness are lacking. A few open-label studies provided promising results particularly on effectiveness on irritability [374–376].

Aripiprazole

Aripiprazole is reported to have efficacy on irritability, aggressiveness, and disruptive behavior similar to risperidone, although less consistent [377, 378]. In adults with intellectual disability, it is indicated for the treatment of bipolar disorder and schizophrenia spectrum disorders, either as monotherapy or as an add-on to mood stabilizers [343]. The support for the treatment of self-injurious behaviors is less evident, despite promising data from open-label studies [343]. The most frequent side effects are sedation, extrapyramidal symptoms, and weight gain, whose intensity is commonly low and rarely lead to drug discontinuation [379], at least in the short or medium term [380]. Hyperprolactinemia is quite infrequent, unlike most drugs of the same class. In the long term, besides weight gain, aggression and dyslipidemia are reported [381, 382]. Longitudinal data did not support the hypothesis that in persons with ASD, aripiprazole has a better metabolic tolerance profile than risperidone, unlike what has been observed in other clinical conditions [383, 384].

Olanzapine

There is only one RCT on olanzapine, which mainly refers to people with intellectual disability [385], and all other studies are retrospective or naturalistic [386–391]. The designs of these studies are very different from each other and, therefore, do not allow to draw firm conclusions. In general, olanzapine seems to be effective in reducing symptoms related to psychotic and affective disorders, both alone and as an add-on to FGAs, as well as in the management of aggression and destructive behavior, especially if not associated with stereotypies or self-injurious behavior. In adults with intellectual disability, it may be a therapeutic option also for acute mania as an add-on treatment to lithium and valproate, for major depressive episodes including psychotic symptoms, at low dosage and in association with antidepressants, and for the prophylaxis of bipolar disorder [343]. Olanzapine allows dosage reduction and treatment discontinuation over time, although considerable discontinuation effects have been reported, eminently connected to its dibenzodiazepine structure and anticholinergic effects [361, 392, 393]. Most relevant side effects are metabolic disturbances, including increased risk of hyperglycemia, peripheral resistance to insulin, type II diabetes, dyslipidemia, and weight gain, which have been reported to be higher compared with risperidone and aripiprazole. Particular attention to these side effects needs to be paid for adolescent and elderly patients [334–336].

Metabolic side effects have been explained through several hypotheses, referring to (i) blockage of histaminergic (e.g., H1) and serotonergic (e.g., 5-HT2C) receptors, which would manipulate the satiety regulator AMP-K and lead to increased appetite and caloric intake; (ii) complex metabolic hormone imbalance, which would result in an alteration of the levels and functioning of leptin, ghrelin, GLP-1, insulin, and glucose [394–396]; (iii) accelerated adipose tissue lipogenesis and elevated liver fat content in SGA-treated subjects [397]; and (iv) decreased energy expenditure due to the sedative effect of SGAs [398]. Recent evidence points to the modification of

gut microbiome towards an obesogenic shift of the bacterial profile [399]. A screen for more than 1000 marketed drugs against 40 representative gut bacterial strains reported that almost one-quarter of non-antibiotic drugs used in humans, predominantly SGAs, possess antimicrobial property with potential to imbalance the gut ecosystem [400]. Other studies have indicated an association between SGA-induced weight gain and alteration of the microbiome composition [401, 402] as well as a potential impact of some gut bacteria on the systemic bioavailability of orally administered olanzapine or other SGAs [403].

Further research is needed to inform the clinical management of weight gain associated with the use of these drugs in ASD. This might include identifying patients who are more susceptible to this side effect as well as determining the best approach to management (e.g., diet and/or exercise, switching drugs, or pharmacological treatments).

▪ Quetiapine

Although insufficiently studied (no RCT and only few open-label studies), quetiapine does not seem to be particularly effective for the treatment of irritability and PBs, while the response rates for self-injurious behavior vary between 22% and 60% [404–408]. It is frequently used at low doses also for sleep disorders, and for the reduction of anxiety, especially in people with intellectual disability [343, 406, 409]. Like other SGAs, quetiapine can be used in the maintenance therapy of bipolar disorder and in the treatment of bipolar depression, while it seems to be less effective for psychotic disorders. The dosage varies according to the target symptom, with the most common range between 25 mgs and 300 mgs per day. In people with intellectual disability, the daily dose should not exceed 800 mgs. Most relevant side effects are weight gain, increased risk of diabetes, and orthostatic hypotension. In comparison with other SGAs, the risk of hyperprolactinemia and extrapyramidal side effects is low [343, 410].

▪ Clozapine

There are no RCT on the use of clozapine in people with ASD, with or without intellectual disability, but only case-reports, case-series, and retrospective studies. As for the general population, clozapine has to be considered a second- or third-choice drug for the treatment of psychoses refractory to other antipsychotics and psychosis complicated by the onset of chronic extrapyramidal side effects related to the use of other antipsychotics [411]. In schizophrenic patients, clozapine has shown specific anti-suicidal properties [412–414], as well as anti-impulsive and anti-aggressive effects [415, 416]. In persons with ASD, it has been widely used to manage severe PBs refractory to other treatments, including aggression toward others, self-injurious behaviors, hyperactivity, motor restlessness, negativism, tantrums, with indirect positive effects on social interaction [417–422], also in the long-term [421]. Findings supporting efficacy are stronger and more replicated for physical aggression toward others and objects than for self-injurious behaviors.

Clozapine cannot be used as a first-choice treatment because of the relevant safety issues, some of which are considered medical emergencies and potentially life threatening. These include hematological (neutropenia and agranulocytosis), CNS (seizures), cardiovascular (tachycardia, postural hypotension, myocarditis, and cardiomyopathy), metabolic (diabetes, metabolic syndrome), gastrointestinal (constipation, ileus), and neuroautonomic (sialorrhea, fever, urinary incontinence, and sweating) [343, 423]. Metabolic risk is reported to be one of the highest among all antipsychotics, as for olanzapine [424, 425]. Furthermore, monitoring requirements, that is, frequent blood tests and frequent clinic visits, may be difficult to carry out in persons with low-functioning ASD. Nevertheless, a full awareness of these side effects and competence to prevent and manage them can maximize the clear clinical benefit of this drug and minimize the known risks.

▪ Asenapine

Asenapine is a new SGA, available as a sublingual tablet, approved in Europe for the treatment of moderate-to-severe manic episodes in adults, and in the US for manic or mixed episodes of bipolar I disorder in adults and

adolescents. Despite its potential pro-cognitive effect and good metabolic tolerability, asenapine has not yet been considered for any RCT in people with ASD. A case series relating to people with low-functioning ASD or intellectual disability with co-occurrent bipolar disorder or schizoaffective disorder reported a significant improvement between second week and fourth week of treatment, especially on PBs. The dosage varied between 5 mgs and 20 mgs/day and side effects were limited, mainly represented by sedation and emotional flattening [426].

In the general population, few studies highlighted efficacy in borderline personality disorder, where asenapine was shown to be superior to olanzapine in reducing affective instability [427]. Together with clozapine, asenapine is more effective on aggression, particularly physical aggression, than other antipsychotics, which has been associated with the high affinity for D4 receptor (D4/D2 > 1) [428, 429]. Asenapine showed significant and clinically meaningful improvement also in posttraumatic stress disorder patients who had not responded to selective serotonin reuptake inhibitors or mirtazapine [430].

Until recently, asenapine was formulated only as a sublingual tablet and was found to be difficult to administer by this route in some patients with ASD or intellectual disability. At present, transdermal patches are also available, to be applied once daily to the hip, abdomen, upper arm, or upper back area.

Long-term flexible dose administration of sublingual asenapine is generally well tolerated. Somnolence, sedation, and headache are the adverse effects most commonly reported. There is also a considerable reporting of transient oral hypoesthesia with dysgeusia, which result from the local anesthetic properties of the sublingual formulation. Other less common side effects are weight gain, extrapyramidal symptoms, and akathisia.

▪ Ziprasidone

Ziprasidone is indicated for the treatment of psychotic disorders and acute mania, also in people with intellectual disability. Since it was originally designed as a cholesterol-lowering drug, ziprasidone has a favorable metabolic effect, with a low impact on body weight and even some benefits on lipid profile. In a small open-label nonreplicated study on patients with intellectual disability and severe maladaptive behaviors, ziprasidone showed response rates comparable to those of other SGAs and reversed metabolic side effects caused by previous treatments [431]. Other promising results, even if still limited to small open-label studies and case reports, concern its use in the treatment of severe disruptive and other PBs in persons with ASD [432–437]. The response rates that emerged were moderate for irritability, hyperactivity, and self-harm, while it was ineffective on repetitive behaviors [361].

Ziprasidone should be titrated from an initial dosage of 20 mgs/day to a maximum dosage of 80–160 mgs/day, in two daily administrations preferably after a meal of at least 500 kcal. Dosage above 200 mgs/day is not recommended. Adverse effect like insomnia is common. Other commonly reported side effects are drowsiness, dizziness, headache, nausea, respiratory problems, extrapyramidal symptoms, and orthostatic hypotension.

▪ Lurasidone

Lurasidone is a benzothiazol derivative, whose chemical structure resembles ziprasidone and risperidone. Several trials reported positive outcomes in adults with schizophrenia [438–442] and depression in bipolar disorder [443, 444]. The use in people with ASD has been evaluated only in children with and without intellectual disability through one RCT and one case report, which showed higher effectiveness than placebo only for irritability [445, 446]. Another multicenter, open-label, single and multiple ascending-dose study, which evaluated the pharmacokinetic and tolerability profiles of lurasidone in children and adolescents with psychiatric disorders, included a child with ASD [447]. Lurasidone has low risk of inducing weight gain, metabolic, or cardiac abnormalities, but its risk of akathisia may exceed that of other SGAs. Other common side effects are nausea, insomnia, and sedation.

▪ Cariprazine

Cariprazine is a partial agonist with high binding affinity at central D2 and 5-HT1A receptors, and antagonist with high-to-

moderate affinity for 5-HT2A receptors [448, 449]. It is approved for the treatment of schizophrenia and acute manic or mixed episodes associated with bipolar-I disorder in adults. The literature search on its use in ASD identified only one case series, in which all patients received other atypical antipsychotics and/or other psychoactive compounds; 87.5% showed improvement in aggression, impulsivity, and/or self-injurious behavior. No patients reported serious adverse effects [450]. Cariprazine is generally well tolerated, with most common side effects being insomnia, extrapyramidal symptoms, sedation, akathisia, nausea, dizziness, vomiting, and constipation [451, 452]. Due to its long half-life, patients need to be monitored for clinical response and adverse events for several weeks after starting and after each dosage change.

Pharmacotherapy for people with ASD alone, or with co-occurring intellectual disability, ought to be part of an integrated person-centered multidisciplinary planning/intervention. Second-generation antipsychotics (SGAs), such as risperidone and aripiprazole, are currently the most commonly used agent targeting irritability, aggressiveness, and disruptive behavior. They should be used at the lowest possible dose and for the minimum duration, always in combination with non-medication-based management strategies, with withdrawal of medication being considered at regular intervals. Close monitoring of side effects including metabolic parameters is paramount.

16.7.1.2 Antidepressants

Tricyclic antidepressants (TCAs) had been widely used in people with ASD, particularly those with low-functioning ASD, both for the treatment of symptoms linked to the ASD itself and for the associated psychiatric and behavioral disorders. Clomipramine had been the most studied drug of this class and some evidence of efficacy in reducing inadequate eye contact, inappropriate language, and repetitive behaviors is reported, whether the latter are attributable to stereotypies or obsessive-compulsive symptoms [453]. It was reported to be more effective than haloperidol in reducing irritability, temper tantrum, aggression, and hyperactivity [454], but these findings have not been replicated in all studies [455]. In some cases, in fact, TCAs have been associated with onset or increase of aggression, irritability, and hyperactivity [456]. Other frequent side effects are sedation, QTc prolongation, tachycardia, constipation, dry mouth, fatigue, lethargy, and seizures [454, 456–458]. Nortriptyline has shown equal efficacy in reducing hyperactivity, aggressiveness, and ritualized behavior [459], while imipramine has been shown to be poorly tolerated in children with ASD [460].

Selective serotonin reuptake inhibitors (SSRIs), such as fluoxetine, fluvoxamine, sertraline, citalopram, escitalopram, and paroxetine, are the most prescribed drugs due to their greater safety profile than the TCAs, although the results of studies on their efficacy are equivocal. The systematic review of the literature showed some effectiveness in adults, but not in children and adolescents, who reported many side effects [461]. Based on the current evidence, SSRIs cannot be recommended for treatment of children or adults with ASD. The decision on the use of SSRIs for preestablished clinical indications that may occur concurrently with ASD, such as obsessive-compulsive disorder and depression, should be taken on a case-by-case basis.

Fluvoxamine showed low effectiveness and poor tolerability in children and adolescents with ASD, but had positive effects on the management of repetitive behaviors as well as on aggression, social skills, and language use in adults with ASD [462].

Fluoxetine has not shown efficacy in treating repetitive behaviors in children. It is found to be more effective on obsessive-compulsive symptoms in adolescents and adults, although reliability of study findings is poor [463–465].

Sertraline was moderately effective and relatively well tolerated in the treatment of aggression and repetitive behaviors in adults. Some open-label studies suggest some efficacy even in children and adolescents, with reduction of anxiety symptoms at low dosage, but a higher dose may precipitate PBs [466–468].

Citalopram has been shown to have limited efficacy in the treatment of repetitive behaviors in children and adolescents and to be more associated with side effects, in particular, hyperactivity, stereotypy, diarrhea, insomnia, decreased concentration and impulsiveness [469–471]. Studies on adults are lacking.

Escitalopram has been shown to have some beneficial effects in children and adolescents, although its use is limited because of little evidence and some side effects [472]. Studies on adults are lacking as well.

Venlafaxine, a dual selective serotonin and norepinephrine reuptake inhibitor, has shown some beneficial effect in the treatment of hyperactivity, self-harm, and repetitive behavior in children, adolescents, and adults. Safety issues have also been reported. However, at present, studies are very limited [473–475].

A few studies on duloxetine, another dual serotonin and norepinephrine reuptake inhibitor with a higher norepinephrine selectivity than venlafaxine, have shown some efficacy on neurovegetative dystonias, irritability, and somatic anxiety [476].

Trazodone, a heterocyclic antidepressant, appears to be effective in reducing aggression and self-injurious behavior, although the quality of evidence is very poor and refers only to case reports [477].

Mirtazapine, a tetracyclic antidepressant, has been shown according to some reports to be effective in the management of PBs, in particular, inappropriate sexual behavior [478, 479].

Agomelatine, a melatonin receptor agonist (MT1 and MT2) and serotonin receptor antagonist (5-HT2C and 5-HT2B), has been rarely used and studied, with some indications of efficacy on repetitive behaviors, sleep alterations, neurovegetative dystonias, irritability, and somatic anxiety [476, 480].

Vortioxetine has a receptor-binding profile particularly interesting for persons with ASD and cognitive issues. A dysfunction of the glutamatergic system and 5-HT7 receptors has been supported by some studies addressing behavioral flexibility and repetitive behavior in animal model of ASD [481, 482]. Nevertheless, clinical data to date are lacking [483].

Overall, while SSRIs appear to be effective in improving some PBs in adults with ASD, findings on childhood ASD are controversial, with some studies indicating a risk of induction of irritability and agitation.

16.7.1.3 Anti-anxiety Medications

There is little evidence supporting the use of benzodiazepines (BDZ) in persons with ASD, although they are often prescribed to treat anxiety disorders, unspecified anxiety symptoms and PBs [484]. Effectiveness of BDZ in the management of PBs has been found to be limited, and sometimes associated with aggravations [485]. Their long-term use may increase the risk of tolerance and dependence (with withdrawal symptoms), as well as develop cognitive difficulties, the latter frequently documented in older adults [486]. An important area of use, even at high doses, is in catatonia, especially in the acute phase. In fact, catatonia is reported to occur more frequently in persons with ASD than in the general population and represents a potentially fatal psychiatric and medical emergency. In fact, the most authoritative epidemiological studies on the subject report catatonia rates to range between 6.5% and 17% in clinical populations of patients with ASD, especially in late adolescence [487, 488].

BDZ represent the first-line treatment of catatonia. The standard therapeutic algorithm includes a lorazepam challenge test, in which an intravenous bolus of 1 mg lorazepam is administered, and catatonic symptoms are evaluated after 2–5 minutes. In case of lack of response, the procedure is repeated. For patients showing a symptoms reduction of at least 50%, therapy with increasing doses of lorazepam can be set, up to 24 mgs per day. In the event of rapid resolution of catatonia, BDZ-based maintenance therapy is continued for the next 6 months and then tapered off [489].

Among the other compounds for the treatment of anxiety disorders, buspirone is one of most studied, particularly in reference to the efficacy on PBs associated with anxiety. Findings are controversial [490–493]. Beta blockers have also been evaluated for the same purpose, with more consistent indications of efficacy, especially with respect to aggression [494–496].

16.7.1.4 Mood Stabilizers

Despite the lack of good-quality evidence in people with ASD, AEs are widely used in clinical practice with dual indication, that is, epilepsy and mood disorders. In fact, some kind of epilepsy occurs in about 20% of people with ASD, and a higher percentage has a history of seizures and EEG alterations [497, 498]. Mood disorders are also quite frequent in people with ASD, with a prevalence rate of up to 70% [499].

Many AEs are currently used as mood stabilizers in the treatment of bipolar disorder for both acute phase and prophylaxis. In people with ASD, particularly those who are low functioning, AEs are also used for the management of PBs, even if these behaviors are not symptoms of a mood disorder. The relationship between epilepsy, behavioral disorders, and psychiatric comorbidity is complex and to date research findings on this issue are not conclusive [500, 501]. In some cases, a good pharmacological control of epilepsy is associated with an improvement of the individual's general functioning and with an attenuation of co-occurrent neuropsychiatric and behavioral alterations; in other cases, an effective therapy on seizures is followed by the onset or the exacerbation of PBs [501]. These differences seem to depend on a number of factors, including the mechanism of action of the drug and the appropriateness of the prescription [502].

The prescription of AEs as mood stabilizers and for the management of PBs should always be made with caution and after a thorough physical and mental state assessment of the person with ASD. Sodium valproate is indicated for the treatment of complex partial epilepsy, absence seizures, myoclonic, tonic, atonic, and tonic-clonic seizures. In addition to this, valproate is used for the treatment and prevention of manic and depressive episodes of bipolar disorders. It has shown good efficacy on irritability, hyperactivity, stereotypy, impulsivity, sleep disturbances, aggressiveness, self-injurious behavior, and cyclical behavioral alterations that can be interpreted as symptoms of bipolar spectrum disorders in people with low-functioning ASD and intellectual disability [503–505]. The dose requirement of valproate varies depending on the patient's level of metabolism. The most common side effects are weight gain, dizziness, drowsiness, and, in particularly predisposed individual, tremor, ataxia, and hypopiastrinopenia. In young and susceptible women, it may determine polycystic ovary syndrome. Valproate can also cause a transient increase in liver enzymes, while a frank hepatotoxicity is very rare. Pancreatitis and hyperammonemia are also to be considered, although rare and dependent on individual idiosyncrasy. Valproate is contraindicated in women of child bearing age.

Topiramate is indicated as a first-line AE in myoclonic epilepsy, and as an adjunctive therapy in partial seizures, absences and other forms of generalized epilepsy. Some studies have shown good efficacy of topiramate as an add-on therapy to other AEs for the treatment of resistant epilepsy in young people and adults with intellectual disability [506, 507]. However, the low tolerability makes topiramate a second-line treatment, especially in people with cognitive deficits and vulnerability to behavioral alterations. In fact, sedation, psychomotor retardation, ataxia, and exacerbation of speech disorders are common side effects. Topiramate can cause mild-to-moderate worsening of cognitive skills and overall functioning in a considerable proportion of children and adolescents with intellectual disability [508]. Research findings on the efficacy on PBs are controversial. In some cases, emotional lability, irritability, and psychotic symptoms have been reported as possible side effects [343], while other studies have shown improvement of aggressiveness, especially self-directed, and disruptive behavior [509]. In people with Prader-Willi syndrome, positive effects have been reported on food intake, mood stability, and impulse control, especially on skin scratching [510, 511]. Another area of potential utility of topiramate is alcohol abuse [243], which is reported to be an increasing issue in people with ASD [513, 514].

Levetiracetam is an AE indicated for the treatment of some forms of partial and myoclonic epilepsy. It is chemically unrelated to other AEs and has a pharmacokinetic profile

that makes it easy to start and to manage. The risk of interactions with other drugs is very low, since it is primarily excreted through kidney, with little hepatic metabolism and no interaction with the cytochrome P450 enzyme. Furthermore, it has a low plasma protein-binding property (less than 10%).

Levetiracetam appears to be effective for controlling subclinical epileptiform discharges in pediatric patients with ASD and was also associated with improved behavioral and cognitive functions [515]. Effects on mood are still to be clarified, with some studies reporting an increase in irritability, anger, agitation, nervousness, aggressive behavior, mania, and depression in the general population [314, 516–520], and others showing efficacy in reducing hyperactivity, impulsivity, mood instability, mania, and aggression in the general population and in children with ASD [521, 522]. The presence of intellectual disability, poor seizure control, and the presence of psycho-organic syndrome and impulsivity have been shown to be possible predictors of adverse behavioral effects [523].

In general, levetiracetam is well tolerated, with the most common adverse reactions being mild-to-moderate dizziness, flu-like symptoms, headache, rhinitis, somnolence or sedation, and asthenia.

Lamotrigine is considered a first-line treatment for partial and secondary generalized seizures, absences, and tonic-clonic seizures. Another area of use is the prophylaxis of depressive phases in bipolar disorder. However, studies on ASD with or without intellectual disability are missing. Case series and case reports on epileptic children indicate that lamotrigine could induce mania and, in the presence of intellectual disability, increase aggressive behavior [524, 525].

More generally, research did not produce unequivocal results that would support the use of lamotrigine in people with ASD. Most studies do not report any improvement on stereotypies, lethargy, irritability, hyperactivity, emotional reciprocity, sharing pleasures, language and communication, socialization, and daily living skills [526, 527], while others indicate some beneficial effects on aggression/agitation, anxiety, and irritability [314], especially in those with co-occurrent intellectual disability [528].

Lamotrigine is usually very well tolerated, with rare sedative effect and no impact on weight and metabolism. Common side effects include headache, vertigo, and diplopia. Skin rashes, which may evolve in generalized erythema and Stevens-Johnson syndrome, represent a rare but severe side effect. It requires drug withdrawal for extinction and drug titration (25 mg every 7–10 days) for prevention. Dosage should be halved, and the titration time doubled if valproate is used concomitantly, although the combined use with valproate should be avoided, since the high reciprocal interference of the two drugs and the consequent high risk of unpredictable negative effects.

Carbamazepine is widely used in people with ASD, especially in those with low-functioning ASD, for the treatment of epilepsy. In general, it is indicated as a first-choice drug for simple partial and generalized tonic-clonic seizures [258], although some genetic syndromes associated with epilepsy such as Dravet Syndrome are a contraindication [529]. Like valproate, carbamazepine is used as a mood stabilizer in the treatment and prophylaxis of bipolar disorder with a specific indication in case of rapid cycles and significant mood fluctuations [343, 530, 531]. Most benefited symptoms are reported to be irritability and anxiety [314]. Other uses with less evidence base are for the management of aggression and self-injurious behavior, especially in the case of co-occurrence with epilepsy or mood disorder [532, 533]. Carbamazepine is a potent inducer of hepatic cytochrome enzymes and reduces the levels of various drugs including oral contraceptives, valproic acid, etosuccimide, clonazepam, aripiprazole, haloperidol, chlorpromazine, olanzapine, risperidone, quetiapine, and paliperidone. This effect has to be considered also in case of polypharmacy with nonpsychotropic drugs. Furthermore, the association with clozapine is contraindicated due to the synergistic pharmacodynamic interaction, which determines increased risk of agranulocytosis and aplastic anemia, while the association with lithium, often clinically useful, must

include very slow titrations and careful monitoring for neurotoxic effect. Hyponatremia, edemas, and bone mineralization disorders are relevant side effects, particularly in persons with low-functioning ASD and with physical vulnerability. Other side effects such as ocular accommodation disorders, dizziness, and instability are less common and dose-dependent [343]. The treatment with carbamazepine should be started at 100–200 mg/day, and slowly increased to the minimum effective dose, taking into account that about 2–4 weeks from the start a dose increase might be needed to see its effect since carbamazepine self-induces its own metabolism.

Gabapentin is used as an AE only for adjunctive therapy of simple partial seizures. In case of small seizures and atonic, tonic, or myoclonic epilepsy, it may have even a worsening effect [534, 535]. In people with ASD, gabapentin has been shown to have some efficacy for the treatment of irritability, anxiety, and depressive symptoms of a bipolar disorder, especially in people with low-functioning ASD and in case of co-presence of intellectual disability [314, 536]. It has also demonstrated some efficacy in sleep disturbances associated with psychiatric comorbidities and in adults with restless legs syndrome [537, 538]. Other studies report a controversial effect on anxiety and depression [314].

Gabapentin can also be used for the treatment of neuropathic pain as monotherapy or with other AEs and even in patients with severe cognitive and/or motor disability [539].

Gabapentin shows good tolerability and safety. Drowsiness, fatigue, and vertigo are the most common side effects, which can be managed in most cases through simple dose reduction. Behavioral alterations, such as hyperactivity, irritability, anger, or temper tantrum are less common and often require drug withdrawal [343].

Pregabalin is an antiepileptic medication, mostly effective as adjunctive therapy for refractory partial-onset epilepsy. In patients with intellectual disability and resistant epilepsy, it showed a response rate of 25% [540]. It is also indicated for the treatment of neuropathic pain, especially painful diabetic polyneuropathy and postherpetic neuralgia, as well as for pain with fibromyalgia. Studies also suggest mood-modulating properties and beneficial effect on sleep, generalized anxiety disorders, and anxiety symptoms, also when expressed at behavioral level as PBs [541–543]. Some authors of the present chapters experienced some efficacy also on self-injury, anxiety, and depressive symptoms in persons whose psychiatric history contraindicated the use of antidepressants. Pregabalin presents a good pharmacokinetic profile, with rapid absorption, lack of protein binding, linear kinetics, absence of enzyme induction, and lack of interference with other drugs. Most common side effects are mild-to-moderate dizziness, ataxia, somnolence, and diplopia. Weight gain was not prominent in pivotal pregabalin trials, but was more problematic in long-term postmarketing analyses in epilepsy patients. Due to the very low impact on cognitive functioning [544], it can also be used in people with intellectual disability.

Lithium salts can be considered a first-choice drug for treatment and prophylaxis of bipolar disorders, also in people with major cognitive and communication difficulties, who express part of the symptoms in the form of PBs. Nevertheless, research evidence is limited, and most study findings refer to small samples, open-label designs, and case reports, with the main focus on people with low-functioning ASD and intellectual disability [545–548]. Lithium salts seem to be particularly effective in the management of aggression in persons showing other observable and behavioral affective symptoms, with response rates between 46% and 73% [549–551]. Efficacy on other PBs is less replicated with discordant results on self-injurious behaviors, irritability, hyperactivity, and temper tantrums. Fragile X syndrome and some genotypes of Phelan-Mc Dermid syndrome seem to represent a specific indication for the use of lithium [552, 553].

The use is limited because of the narrow therapeutic window and the need for continuous blood testing for the monitoring of serum lithium levels as well as renal and thyroid function. The therapeutically effective dose of lithium has to be assessed individually based on the clinical response and emergence of side effects, especially tremor, which can also occur

when lithium does not exceed the therapeutic range. Other common side effects are polyuria and gastrointestinal disorders. Furthermore, adaptation is necessary over time, depending on the patient's age, presence of comorbidities, and other concomitant therapies, such as thiazide diuretics. Usually, children and adolescents require a high dose to reach therapeutic plasma level, while in the elderly, the low body water volume requires a drastic reduction in the dose.

The use of antidepressants, AEs as mood stabilizers, and anti-anxiety medications in subjects with ASD ought to be with caution following a careful mental state assessment and with expertise in psychopharmacology for persons with ASD. The management of PBs in ASD also ought to be with caution. The SSRIs are the most prescribed drugs in ASD due to their good safety profile, but have been shown to have limited efficacy in the treatment of repetitive behaviors in children and adolescents and to be more associated with side effects, in particular, hyperactivity, stereotypy, decreased concentration, and impulsiveness.

16.7.1.5 Stimulants and Atomoxetine

Pervasive hyperactivity, impulsivity, and inattention, which represent the central features of attention-deficit hyperactivity disorder (ADHD), are frequently present in people with ASD and/or other neurodevelopmental disorders, with rates between 6% and 80% [554–557]. If not adequately treated, they can greatly hamper the effectiveness of habilitative and psychoeducational interventions.

Psychostimulants are the first-choice drugs in children with ADHD, with a response rate of 70–80% [558]. Among these, methylphenidate is the most studied and the most used, both in the immediate release formulation and in the extended-release formulation. Currently available data mainly concern studies on children and adolescents, highlighting a considerable superiority compared to placebo in reducing hyperactivity and impulsivity at dosages ranging from 0.15 to 0.50 mg/kg/day [559]. Research findings also suggest a response rate of 49% and an effect size of 0.54 among the ASD children, lower than what is normally achieved in ADHD children in the general population [558, 560]. The impact of stimulants on ADHD symptoms appears to be even less striking in people with co-occurrence of ASD and intellectual disability [410, 561].

Methylphenidate may represent a trigger for hypersensitivity to frustration and subsequently for a series of PBs such as aggression or temper tantrum [562–564]. Emotional dysregulation may be worsened by sedative drugs and antipsychotics [565, 566]. The efficacy of methylphenidate and other stimulants is verifiable within minutes, especially in children. Therefore, it is convenient to carry out a preliminary test of the efficacy in a protected environment before starting the treatment. Another advantage is the short half-life, so, negative effects rapidly disappearing after drug discontinuation.

Treatment should be started with minimal doses, such as 5–10 mg/day, and increased gradually. The treatment plan can be tailored to the individual needs, with the possibility of combining the immediate and extended-release formulations. In children and adolescents, attempts of temporary discontinuation of the treatment can be done, especially during the weekend or in the summer period, as well as attempts of dosage reduction over the years. In adults, continuation of treatment is recommended, especially when effective also for emotional dysregulation and PBs [343].

People with ASD seem to be more vulnerable to side effects of psychostimulants than the general population, which represents a reason for treatment discontinuation in about 20% of cases [559, 567]. Irritability is the most frequently reported side effect, while seizure threshold lowering is one of the most common concerns for prescribers, especially in patients with complex neurodevelopmental disorders. Research findings based on large samples indicate that stimulants do not increase the number of seizure-related hospitalization, even in persons with intellectual disability, cerebral palsy, or congenital abnormalities of the central nervous system [568]. Other less common side effects are tics, stereotypies, agitation, loss

of appetite, and insomnia. Insomnia may be due to a medication rebound effect and, therefore, minimized through the postponement of the day's last dose or through the introduction of a prolonged release formulation. A side effect that may be of greater importance in patients with ASD than in other patients is "hyperfocusing," which can lead to an aggravation of social withdrawal [569]. Despite this, stimulants remain a valid and safe option for persons with ASD, with therapeutic effects being reached at lower doses than in the general population [567].

Atomoxetine is a selective norepinephrine and dopamine reuptake inhibitor that is not classified as a stimulant. Although it is indicated for the treatment of ADHD in patients of any age (from 6 years on), it seems to be more effective in adults than in children, probably for the reason of the different balance between dopaminergic and noradrenergic pathways in the prefrontal cortex. The drug presents a kinetic of action completely different from that of stimulants and develops its therapeutic effect after a period of 1–2 months from the start. RCT and open-label studies conducted in children with ASD and ADHD have given controversial results in respect of efficacy on ADHD core symptoms, irritability, verbal and motor stereotypies, social withdrawal, and other PBs. More concordance has emerged on side effects, which were found to be few and almost limited to the first period of treatment. The most common side effects are gastrointestinal symptoms, fatigue, palpitations, hyporexia, irritability, and sleep disturbance [570–577].

In persons with low-functioning ASD, atomoxetine seems to be a little less effective and tends to present more side effects, especially in children [578–581]. A case report describes the induction of psychotic symptoms in a teenager with intellectual disability treated with 60 mg/day [582]. Atomoxetine should be started at the dose of 0.5 mg/kg/day, increased after 1 week to 1.2 mg/kg/day, up to a maximum daily dosage of 70–80 mg. Slower titration should be observed in patients with high sensitiveness to side effects.

In general, the quality of studies is modest for all people with ASD [580, 583].

Children, adolescents as well as adults with ASD are more vulnerable to side effects of psychostimulants than the general population, and therefore co-occurring symptoms of ADHD ought to be tailored selectively, with treatment started with minimal dose and increased gradually.

16.7.1.6 Other Medications

Clonidine, an alpha2-adrenergic receptor agonist, has been shown to be effective in reducing impulsivity, inattention, and hyperactivity. It seems to be effective in reducing sleep problems, to a lesser degree in improving ADHD symptoms, mood instability, and aggressiveness in children with ASD. Its use is based on the hypothesis that some symptoms of autism could be the result of a dysregulation of the adrenergic system that leads to a state of hyperarousal. Many side effects are reported, especially on the cardiovascular system, sometimes even serious (e.g., dysrhythmias, hypotension, and conduction abnormalities). The short half-life (a few hours, determining the need for six daily transdermal administrations) and the appearance of tolerance after a few months of therapy limit its clinical utility [584–587].

Guanfacine, an alpha-adrenergic agonist, has been shown to improve hyperactivity, distractibility, impulsiveness, insomnia, and tics in children and adolescents with ASD. It has fewer side effects than clonidine and it is certainly more manageable given the greater half-life [588–591].

Naltrexone is an opiate receptor antagonist commonly used for the pharmacological treatment of alcoholism and opioid addiction. Its use in ASD starts from the hypothesis that autism is linked to a hypersecretion of opioids in the brain as a consequence of a dysfunction of the adreno-hypothalamus-pituitary axis. In several studies, it has been shown to be moderately effective in reducing hyperactivity, restlessness, and self-injurious behavior. Improvements in social withdrawal, attention skills, and speech production were also observed [592–594]. As far as side effects are concerned, naltrexone does not appear to increase the risk of serious adverse events over placebo [595].

Secretin is a 27-amino acid peptide hormone that stimulates the secretion of bile in the liver and insulin in the pancreas. With regard to the use of secretin in subjects with ASD, it should be pointed out that its widespread use and publicity are due to the disclosure made by the mass media of "dramatic" effects arising after infusion of it as a stimulus test for diagnostic purposes in three children with ASD and gastrointestinal symptoms. Rapid improvement in language and social skills were reported. Secretin has then been studied extensively in multiple RCTs, and there is clear evidence that it lacks benefit. However, some RCTs showed a potential efficacy in a specific subgroup of autistic children with chronic diarrhea compared with autistic children without gastrointestinal symptoms [596–600].

16.7.1.7 New Therapeutic Strategies

Many compounds are currently under study as a possible treatment for ASD core symptoms and the focus of drug design is progressively addressing communication and social interaction. Research has informed our understanding of ASD as a genetically and biologically based neurodevelopmental disorder. However, there still is a great unmet need for interventions that reliably address such core symptoms of ASD. The research studies carried out so far show many difficulties encompassing the genetic and phenotypic heterogeneity of ASD, which hampers the identification of common neurobiological pathways underlying the disorder. However, many pathogenetic theories are proposed and seem to be confirmed in some specific subgroups of individuals with ASD.

Cholinergic Agents

Acetylcholinesterase inhibitors have been approved by the Food and Drug Administration (FDA) for the treatment of Alzheimer's disease. Postmortem data suggest that cholinergic abnormalities may be implicated in ASD, although the precise pathogenetic mechanism has not yet been elucidated [601]. Acetylcholine (Ach) modulates attention, novelty seeking, and memory. Recent animal studies have shown that the increase in Ach following the inhibition of acetylcholinesterase is able to reduce cognitive rigidity and improve social interaction in autistic phenotypes [602]. It is interesting to note that the increase in Ach positively modulates the selective attention and the emotional processing, which are compromised in individuals with ASD [603].

Tacrine, the first drug available in this class, has shown modest efficacy in the short-term treatment of irritability and hyperactivity and in improving eye contact and language [604]. The poor safety profile, especially regarding the possible hepatotoxicity, prevented further studies.

Rivastigmine produced satisfactory results in a 12-week open-label study enrolling 32 children with ASD. Statistically significant improvements in expressive language capacity have been noted [605].

Galantamine has been shown to improve expressive language in 3 adults with ASD [606]. In subsequent studies with larger samples and one placebo-controlled, it showed improvement in irritability, distractibility, social withdrawal, and eye contact [607–609].

Donepezil has been shown to have some effect on the treatment of irritability and hyperactivity. No definitive results have been achieved regarding its effectiveness in improving language and social interactions. It seems that it could also induce a REM sleep augmentation, increasing its duration and decreasing its latency, and this could have positive effects on learning and cognition. In a recent double-blind, placebo-controlled trial, it has been shown to improve receptive language skills [610–614].

A pilot study with mecamylamine, a non-competitive antagonist of nicotinic receptors, showed no effectiveness on autism's core symptoms [615].

Glutamatergic Agents

Glutamate, the main excitatory neurotransmitter, is highly concentrated throughout the brain. It seems to have a crucial role in neuronal plasticity and the maintenance of cognitive functions. Glutamate is transformed into gamma-aminobutyric acid (GABA), an inhibitory neurotransmitter, through a

specific enzyme (glutamic acid decarboxylase). Excess glutamate has been shown to be a powerful neurotoxin that leads to neuronal cell death (proapoptotic effect) and has been believed to play a role in the pathophysiology of some neuropsychiatric disorders, such as schizophrenia, obsessive-compulsive disorder, and Alzheimer's dementia. The hyperglutamatergic hypothesis was also proposed in the genesis of the ASD. There are many hypotheses that have been formulated to explain the increase in glutamate in these individuals and equally numerous are the medications studied to modulate the glutamatergic system in order to effect ASD core symptoms. In general, those molecules that are able to promote GABAergic or modulate glutamatergic receptors could have a potential role in treating autistic symptoms.

D-cycloserine (an antibiotic approved for the treatment of tuberculosis), a partial N-Methyl-D-Aspartate (NMDA) agonist, has been shown to be effective in the treatment of the negative symptoms of schizophrenia [616] and in improving lethargy and social withdrawal in children with ASD [590]. These positive results have been confirmed in subsequent studies conducted on young adults with ASD, showing an improvement in stereotypies and social skills [617, 618]. The findings of a recent study suggest that D-cycloserine may help youth with ASD to maintain skills gained during social skills training [619].

Amantadine, an NMDA antagonist, was only evaluated in one placebo-controlled study. The study involved 39 children and adolescents with ASD, but did not achieve statistically significant results [620]. A further study, a double-blind, placebo-controlled trial on children with ASD, that aimed to investigate the effect of adding amantadine to risperidone for the treatment of behavioral problems in such population, showed it to be effective in reducing hyperactivity and irritability [621].

Memantine, an NMDA antagonist, approved by the FDA for the treatment of Alzheimer's disease, has been evaluated in several retrospective and open-label studies. It has been shown to be effective in treating hyperactivity, irritability, lethargy, social withdrawal, and distractibility in people with ASD. Its combination with risperidone was found to be superior to monotherapy of memantine in treating irritability, hyperactivity, and stereotypies in people with ASD. Several studies report an improvement in communication and memory skills and in self-stimulating behaviors [603, 604, 622–627]. Three phase 2 trials have recently been conducted to assess the efficacy and long-term safety of extended release memantine treatment in children with ASD showing considerable improvements in social responsiveness and no evident safety concerns [628]. A randomized, placebo-controlled study did not demonstrate clinical efficacy of extended release memantine in ASD, although it reassured about its tolerability and safety [629].

Acamprosate, a GABAa agonist, approved by the FDA for maintaining alcohol withdrawal in adults, has shown efficacy in improving hyperactivity, social withdrawal, and social response in youth with ASD [630–632].

Arbaclofen, a GABAb agonist, seems to improve social functioning in individuals with Fragile X syndrome [633] even if two recent phase 3 placebo-controlled trials by the same authors did not meet the primary outcome of improving social avoidance [634]. An 8-week-long open-label study enrolling 32 children and adolescents with ASD showed Arbaclofen to be effective on irritability and social withdrawal [635].

Bumetanide, a diuretic agent that reduces intracellular chloride and increases GABAergic inhibition, seems to improve interaction, social communication, and restricted interest in children with ASD. Mild hypokalemia and polyuria are reported as side effects [636–639].

Melatonin

Melatonin (N-acetyl-5-methoxytryptamine), a hormone produced by the pineal gland (or epiphysis), acts on the hypothalamus and has the function of regulating the sleep–wake cycle. Melatonin and other melatonin-agonist substances have chronobiotic effects, that is, they can regulate the rhythms of functioning of the body with respect to the alternation of day and night (circadian system).

Abnormalities of the melatonin system, in particular reduced production of melatonin during the night, seem to be associated with pathological changes in the sleep–wake rhythm that are very frequently found in persons with ASD. Sleep delay, night awakenings, and early awakening are the most frequent disturbances in ASD, with frequencies ranging from 40% to 86% [640]. These pathological changes in sleep are persistent and difficult to manage and are often associated with a worsening of social functioning, irritability, PBs, and a deterioration in quality of life. The efficacy of melatonin in people with ASD has been demonstrated by several studies, recently revised and collected in a meta-analysis [640]. Exogenous melatonin (variable dosage 1–6 mg/day) was able to reduce sleep latency and the number of awakenings per night and increase the total duration of sleep [640–643]. On the other hand, the production of melatonin appears to be closely related to the availability of serotonin, which we know to be involved in the pathogenesis of ASD core symptoms [644]. Most of the studies available have grouped small samples of subjects with ASD, often associating them with individuals with other neurodevelopmental disorders, detecting generic outcomes mostly related to the quality of sleep. Only a few studies have attempted to more specifically investigate other symptoms typically present in people with ASD. However, the results obtained seem to suggest that the exogenous administration of melatonin may improve social communication (verbal and nonverbal communication, social interaction), social withdrawal, repetitive and stereotyped behaviors, limited interests, and finally, the ability to adapt to changes [645–648]. An RCT involving 125 individuals aged 2–17 years with ASD showed a significant improvement of sleep duration and onset but also of externalizing but not internalizing behavior and caregivers' quality of life [649]. There is need for more RCTs to confirm these results.

Neuropeptides

Oxytocin is a 9 amino acids peptide hormone produced by the hypothalamic nuclei and secreted by neurohypophysis. Its main action is to stimulate the contractions of the smooth muscles of the uterus, especially in the last period of pregnancy when it plays an important role in the beginning and maintenance of labor and delivery. It seems that the genetic structure and the phenotypic basic expression of this neuropeptide are stable and phylogenetically very ancient, while the genetic regulation of its receptors would present an extreme variability. Such heterogeneity seems to underlie the differences in social behavior both among different animal species and within the same species. Animal experiments have shown the importance of this hormone in mating and offspring behavior. Many human studies have begun to explore the role of this neuropeptide in cognitive skills with relational implications. The first results confirm that the variations of the genes that code for its receptors can modify brain function and contribute to the individual characterization of social behaviors. Particularly interesting result are found in the studies that outline the role of oxytocin on empathy, relationship with others, and self-esteem.

It has been hypothesized that abnormalities in the production of oxytocin and vasopressin may contribute to the development of repetitive behaviors and social deficits found in ASD [650]. In this regard, several studies have been carried out in people (children and adults) with ASD showing significant improvement in stereotyped behaviors and a marked decrease in obsessiveness after treatment with oxytocin via infusions [651–654]. More recently, studies with nasal application of oxytocin demonstrated its effectiveness in improving social interactions and communication [655–660]. Other studies do not suggest significant clinical efficacy on core symptoms [657, 661]. In none of these studies, oxytocin produced any significant side effects [662, 663]. Research in the coming years may better clarify the role of oxytocin in the treatment of ASD core symptoms [664–667].

Vasopressin is also a neuropeptide used by neurons in the brain to communicate with one another. High levels of vasopressin are associated with anxiety, aggression, and sensory processing. Animal studies have shown that experimental alterations of the proper func-

tioning of vasopressin determine a variety of social deficits, including impaired memory for peers and a reduced interest in social interaction. Conversely, the administration of vasopressin to mice with a genetically induced form of ASD improves their social functioning [668]. Vasopressin is already approved by the Food and Drug Administration for use in humans and has proved to be successful in treating some common pediatric conditions, including bedwetting. Similar to oxytocin, it has also been shown to improve social cognition and memory in people who do not have ASD [669–672]. Given the connection between vasopressin signaling and social behavior, vasopressin receptors have been considered a therapeutic target for the treatment of ASD, and the preliminary evidence is provided through both animal and human studies [673, 674]. New researches are ongoing in persons with ASD of any age.

Balovaptan is an orally administered selective V1a receptor (Vasopressin receptor 1a) competitive antagonist, for which phase 1 and phase 2 clinical trials did not identify any safety or tolerability concerns in healthy male volunteers and in men with ASD [675]. In persons with ASD, balovaptan showed dose-dependent clinically meaningful improvements on the Vineland-II Adaptive Behavior Scales composite score (VABS-II-ABC) compared with placebo [675], which supports further study as a potential treatment for the socialization and communication deficits.

Conclusions

The elucidation of efficacious treatments for ASD is plagued with limitations in study design, statistical power, and clinical heterogeneity of the disorder. Evidence on efficacy, dosage, and safety is limited [676] and derived mostly from naturalistic studies or case reports, with a main focus on identification of side effects and discontinuation rate. Placebo-controlled or active-controlled studies are limited, with small sample sizes. No psychoactive drug has shown efficacy on ASD core symptoms. Some efficacy is reported only for antidepressants and antipsychotics, especially SGAs, on repetitive behaviors, although discordant results indicate that further research is needed. Newer compounds, including cholinergic, glutamatergic agents, and neuropeptides, have been associated with future potential utility, while studies of fenfluramine, secretin, opiates, and mood stabilizers generally found no effect.

Psychotropic medication used for comorbid psychiatric disorders shows some evidence of efficacy, albeit of poor quality, especially for mood disorders and ADHD. For anxiety, SSRIs continue to be widely prescribed, despite the lack of positive clinical trial results and the high risk of behavioral disinhibition. SGAs have become the most commonly used drug class targeting irritability, aggression, and other PBs. They show significantly reduced rates of side effects, particularly extra pyramidal symptoms, compared to FGAs. Specifically, risperidone and aripiprazole are the two approved agents in youth with ASD for the treatment of irritability marked by aggression, self-injury, and severe tantrums. More limited data are available on drug refractory aggression and self-injury, including those for clozapine. Some of the very new-generation APs show a receptor-binding profile that matches the characteristics of many people with ASD, with low metabolic effects. Antiepileptics and mood stabilizers have also shown promising results, but indications seem to be even less precise than for the other classes.

Frequent off-label prescriptions combined with potential tolerability issues render this area of psychopharmacological practice challenging, and more work is needed from the field to identify potential predictors of treatment response toward specific target symptoms.

Basing on current knowledge, psychoactive drugs should only be prescribed for the treatment of co-occurring psychiatric disorders. For the management of aggressive behavior or other severe PBs, the decision to intervene with a psychoactive drug should be preceded by a careful evaluation of all the aspects related to epiphenomena brought to the observation of the clinician, conducted with appropriate procedures and tools. It must be ascertained that PBs do not depend on pain or physical illnesses, environmental factors, or life events. The drug should be used

only in the best interests of the person, after exclusion of any possibility of nonpharmacological treatment, in accordance with scientific evidence and available guidelines, and after a careful evaluation of the cost–benefit ratio (see also Problem Behaviour chapter). Particular attention must be paid to the search for factors for which the drug could be contraindicated, ineffective or harmful (make sure that the necessary instrumental examinations and investigations have been performed), or with which it could negatively interfere.

Before choosing a treatment, it is also advisable to collect a good pharmacological history, taking note of efficacy, tolerability, and side effects of every previous drug. An appropriate consideration of the availability of services and supports for the regular intake of therapy and for the management of any unwanted effects or iatrogenic complication may be also useful in the identification of the best drug to prescribe.

Once the treatment has been started, it is very important to monitor its efficacy and safety at regular intervals, also through the appropriate laboratory and instrumental tests. When the drug has been introduced with the main purpose of managing a PB, it is indispensable to frequently reevaluate the dosage, and to use the lowest possible dose for the shortest possible time (see also the Psychopharmacology chapter).

Psychopharmacological therapies should also be shared with the multidisciplinary team, especially with respect to the rationale and objectives. All professionals involved in the care of the person with ASD should be informed at regular intervals. If no significant beneficial effects are noted after a three-month period, treatment should be discontinued.

In persons with ASD and/or other neurodevelopmental conditions, the usefulness of psychoactive treatments should be judged in terms of effectiveness rather than efficacy (on target symptoms) and safety, with the former including the capability to keep the patient on treatment for the right time and the impact on quality of life. Psychopharmacological therapies must be considered an integral part of a person-centered program of interventions and, therefore, take into account the impact on individual interest and satisfaction toward various areas of life.

Great attention must also be paid to the ethical implications of the treatment. The professional figures involved are required to document the assessment of the patient's ability to sign his/her informed consent to the proposed therapy. When this ability is lacking, consent must be reached with the involvement of the legal representative and the family.

The intervention plan must be clearly communicated to the person with ASD, even in case of co-occurrence of severe intellectual disability, resorting if necessary, to specific or innovative systems, such as use of pictures and accessible information (see ► www.ld-medication.bham.ac.uk) [677]. This must also include the explanation of a possible use of the drug outside its official indications (off label). In this case, information must be provided on the type and quality of the available evidence on effectiveness [320].

Frequent off-label prescriptions combined with potential tolerability issues render the psychopharmacological practice challenging. More research is needed to identify potential predictors of treatment response toward specific target symptoms and the impact of treatment on quality-of-life measures.

16.7.2 Nonpharmacological

The nonpharmacological interventions with scientific evidence based on developmental ages of persons with ASD include the following:

(a) Early intensive programs oriented to improve the trajectory of the children's developmental and adaptive functioning and (b) focused applied behavior analysis (ABA) that targets specific behavior that may interfere with the well-being regardless of the person's level of functioning and age.

Nonpharmacological treatments for people diagnosed with ASD are under constant development and, since their application in the 1980s, they have been proved to be effective in reducing the impact of the core symptoms

by improving communication and autonomy skills and reducing problem behavior in children and adults with ASD. Throughout the whole life span, effective treatments are derived from different applications of ABA and of contextual behavioral sciences. However, they greatly differ both for intensity and for treatment objectives from childhood to adulthood.

Besides treatments, assistive technology (AT) may be used to help persons with ASD to increase, maintain, or improve functional capabilities. It may aid in a variety of areas, including basic communication, reading, writing, and math, telling time and managing schedules, learning and applying social skills, managing sensory issues, remaining safe, and daily activities. Low-tech, mid-tech, and high-tech AT are the most common categories. Anything that does not require electricity, such as weighted vests, sensory balls, or graphic boards, is considered low-tech AT. Mid-tech AT is straightforward enough to be both affordable and simple to use. Battery-operated sensory devices, visual timers, and social skills movies are all examples. High-tech AT refers to the most complicated devices or equipment with digital or electronic components that can be computerized. It includes robots that are designed to aid in the development of social skills. One of the most significant applications of assistive technology is Augmentative and Alternative Communication (AAC), which can help persons with ASD of all ages to gain independence, communicate more effectively, and interact more socially. There are a variety of AAC devices and therapy tools available, with many new ones emerging in recent years, such as speech-generating devices [678, 679] and less high-tech methods of communicating using pictures [680].

The evidence-based non-pharmacological treatment, also referred to as psychosocial treatment or behavioral intervention, has extensively been considered in international guidelines (e.g., [254, 681, 682]). Currently, there are many intervention models based on ABA, which integrate the more structured approaches, such as the discrete trial training (DTT) used by Lovaas [683], with more naturalistic intervention models based on development stages such as ABA-Verbal Behavior (ABA-VB), Early Start Denver Model, TEACCH, and social skills training. From the mid-1990s, the research and dissemination of interventions for ASD mainly concerned a second generation of behavioral interventions. In addition, some interventions based on cognitive-behavioral therapy (CBT) have been adapted to deal with certain related psychological problems, such as anxiety and stress perceived in people with ASD [684]. A behavioral pattern of contextualistic matrix, the dialectical behavior therapy (DBT), has been shown to be effective in adults with ADHD, even though it has never been tested on people with ASD [685]. The literature on treatments based on the analysis of applied behavior (ABA) on high-functioning ASD is still limited compared to the extensive evidence demonstrated in the treatment of low-functioning disorders and early interventions. At the moment, treatment protocols that can promote health, facilitate self-management, and reduce stress in ASD are of great interest and importance to scientific research and for the improvement of the quality of life. Among modern cognitive behavioral therapies, acceptance and commitment therapy (ACT) has been used with high-functioning ASD people and has shown a reduction in stress levels, hyperactivity, and emotional distress, and an increase in prosocial behavior [686].

Despite being applied in many forms, with different durations, intensity and focus, and being declined very differently in protocols, all ABA-based interventions for ASD have the following core characteristics [687]:

1. An objective assessment and analysis of the person's condition by observing how the environment affects the person's behavior as evidenced through appropriate data collection
2. Importance given to understanding the context of the behavior and the behavior's value to the individual, the family, and the community
3. Utilization of the principles and procedures of behavior analysis in order to improve the person's health, independence, and quality of life

4. Consistent, ongoing, and objective assessment and data analysis to inform clinical decision-making

Behavioral and other psychological interventions for people with ASD may be divided into two main groups:

(a) Intensive behavioral and developmental programs aimed at improving overall functioning and altering outcome (also known as Comprehensive ABA Treatment or EIBI, Early Intensive Behavioral Intervention)
(b) Interventions which aim to address specific behavioral difficulties associated with ASD, such as sleep disturbance, decrease problem behavior or increase positive behaviors, such as initiating social contact with peers, or developing communication skills (also referred to as Focused ABA Treatment)

16.7.2.1 Intensive Behavioral and Developmental Programs

Comprehensive ABA or EIBI refers to the treatment of multiple developmental domains, such as communicative, social, cognitive, emotional, and adaptive functioning. Typically, maladaptive behaviors, such as noncompliance, tantrums, and stereotypy, are also included in the focus of treatment. Although there are different types of comprehensive treatment, in EIBI, the overarching goal is often that of closing the gap between the person's functioning level and that of typically developing peers. According to the Scottish Intercollegiate Guidelines Network (SIGN) [682], modern EIBI programs are best described as behavioral and developmental programs. These usually start when the child with ASD is 36–48 months, with some reviewed studies starting at 18 months. These intensive programs amount to around 30 hours of treatment per week (plus direct and indirect supervision and caregiver training), but more recent reviews include evidence from programs ranging from 13 to 28 hours per week [687]. Not all children diagnosed with ASD participate in intensive programs; however, a number of more specific focused ABA-based interventions are also available (e.g., PECS that focus on the communicative skills) and they do not require the same level of intensity. The program is highly individualized, taking into account the idiosyncratic and individual motivations and specific cognitive, metacognitive, and learning needs of each child. Initially, this treatment model typically involves 1:1 staffing and gradually includes small-group formats as it sees appropriate. Comprehensive treatment may also be appropriate for older individuals diagnosed with ASD, particularly if they engage in severe or dangerous behaviors across environments.

EIBI programs are manualized, intensive, and target a comprehensive range of skills for training, practice, and generalization; they also aim to engage the child with ASD in structured learning [688]. Although the programs vary broadly in the emphasis given to different skills (verbal behaviors, pivotal responses, play, joint attention, etc.), they all tend to start with very basic skills (simple communication skills, sitting, looking, listening) and over time, they work toward more complex, verbal and metacognitive skills, such as self-monitoring and perspective-taking. Initially, treatment is typically provided in structured therapy sessions, which are integrated with more naturalistic methods, as it seems more appropriate. As the children progress and meet the established criteria for participation in larger or different settings, treatment in those settings and in the larger community should be provided. Treatment components are generally drawn from areas that will influence the quality of life of the person. EIBI programs have different levels of parental involvement and international guidelines recommend that parent-mediated intervention programs should be considered for children and young people of all ages who are affected by ASD, as they may help families interact with their child, promote development and increase parental satisfaction, empowerment, and mental health [688]. NICE guidelines 170 [689] recommend including play-based strategies with parents, carers, and teachers to increase joint attention, engagement and reciprocal communication in the child or young person. Strategies should:

- Be adjusted to the child or young person's developmental level
- Aim to increase the parents', carers', teachers', or peers' understanding of, and sensitivity and responsiveness to the child or young person's patterns of communication and interaction
- Include techniques of therapist modeling and video-interaction feedback
- Include techniques to expand the child or young person's communication, interactive play, and social routines

The intervention should be delivered by a trained professional and supervised regularly. Generalization from prompted to spontaneous use of trained skills is a key element in caregiver training. NICE guidelines 170 [689] also suggest involving and training of different people based on the age and context of the person with ASD, with parent, carer, or teacher mediation for pre-school children, and peer mediation for school-aged children.

Training family members and other caregivers to manage problem behavior and to interact with the individual with ASD in a therapeutic manner is a critical component of effective treatment models. Acceptance and commitment training (ACT) processes and procedures have recently been integrated with traditional cognitive-behavioral procedures for parent training to enhance the parent-caregiver acceptance of difficult personal experiences related to the complexity of the educational and emotional interactions with their child, and the ability to act in synergy and full awareness for the management of problem behaviors, in a context of value and full acceptance of the other as a whole person [690, 691].

EIBI thus describes comprehensive teaching programs, rather than interventions aimed at reducing symptoms. A Cochrane review [692] concluded that while EIBI cannot be recommended universally, it should still be considered on a case-by-case basis. In this review, there were no clear predictors identifying which children will respond to it or not, and while intensity or total hours were moderately correlated with some outcomes, such correlations were not always significant. EIBI may not be cost effective when considered individually on the level of early-years service, but the cost effectiveness of it may change when considered on a societal level. Although interventions can be very intensive, no difference was found between EIBI and controls on measures of parental stress, parental anxiety, parental depression, or parental positive perception; however, no adverse sequel arising from EIBI has been reported.

16.7.2.2 Focused ABA Treatment

Focused ABA refers to treatment provided directly to the person for a limited number of behavioral targets. It is not restricted by age, cognitive level, or co-occurring conditions. Focused ABA treatment may involve increasing socially appropriate behavior (e.g., requests) or reducing challenging behavior (e.g., aggression) as the primary aim. It is crucial to target increases in appropriate alternative behavior even when reduction of challenging behavior is the primary goal. This is because the absence of appropriate behavior is often the precursor of serious behavior disorders. Therefore, individuals who need to acquire certain skills (e.g., communication, learning how to tolerate change in environments and activities, self-help, and social skills) are also suitable candidates for focused ABA.

Focused ABA plans are appropriate for individuals who: (i) need treatment only for a limited number of key functional skills (examples of key functional skills include, but are not limited to, establishing instruction-following, social communication skills, compliance with medical and dental procedures, sleep hygiene, self-care skills, safety skills, and independent leisure skills) or (ii) have such acute problem behavior that its treatment should be the priority. Examples of severe challenging behaviors requiring focused intervention include, but are not limited to, self-injury, aggression, threats, pica, elopement, feeding disorders, stereotypic motor or vocal behavior, property destruction, noncompliance and disruptive behavior, or dysfunctional social behavior.

Treating behavioral disorders and challenging behavior in people diagnosed with ASD pragmatically means following these five steps:

1. Operationally define the challenging behavior

To define operationally the challenging behavior, professionals and caregivers need to describe the behavior as it is manifested, reporting it in an operative and objective way. For a definition to be operational, it is necessary that this definition is understood and interpreted, unambiguously, in the same way by all the people involved (e.g., "she is aggressive" is not an operational definition, "hits the head of other people with left hand" is an example of operational definition). Elaborating an operational definition of challenging behavior is a fundamental action for several reasons: a) it allows to clearly and unambiguously identify the behavioral parameters to be observed and it is useful for both the collection of baseline data prior to treatment and data during treatment; b) it helps the clinician to keep the phase of data collection separate from the phase of interpretation of challenging behavior (definition of the hypothesis of the functional value of the behavior in the context); c) it facilitates the comparison between the care team and the caregivers in the selection of challenging behaviors of greater severity for the person with ASD and their contexts (priority decision for the intervention on the challenging behavior).

2. Measurement of challenging behavior

Data collection of the frequency, intensity, duration, and latency of the challenging behavior helps us to define the baseline as a necessary reference parameter to understand the effectiveness of ongoing treatments. The baseline and the systematic collection of data during the treatment phase will make it possible to identify ineffective procedures and, in addition, to verify the greater effectiveness of the chosen treatments. A record of daily progress can also function as a powerful reinforcement for the treatment provider, as well as for the family member or the person receiving the treatment.

Collecting the quantitative parameters on the challenging behavior means using the different observation systems which are available in a wide variety of scientific supported methods. An exhaustive analysis goes beyond the scope of this text, but it is important to remember that the purpose of this multiplicity of methods is to allow us to find a method that fits the behavior that is going to be observed and not vice versa.

To sum up, a distinction can be made between continuous and sample recording: continuous recording is the recording during the entire period of observation. Once the observation time has been chosen (which depends on the behavior), all the events of the operationally defined challenging behavior must be observed and measured for the entire duration of that period. The main alternatives to continuous recording are sampling techniques, which divide the observation period into time intervals. In sampling techniques, the observation period is divided into intervals and the observers record whether the behavior is occurring during each interval, thus leading to an estimation of the behavior. The most commonly used sampling techniques are: (a) whole interval, (b) partial interval, and (c) momentary time sampling.

3. Functional assessment

The collection of the frequency, duration, intensity, or latency of a behavior informs us about the characteristics of that behavior and is a useful parameter to evaluate the effectiveness of treatment. However, the sole "quantitative" measure of behavior does not provide accurate information about its meaning in the context. Functional analysis is useful in this very respect: it can help us understand why that specific behavioral repertoire, in a person's learning history, remains alive and active. Therefore, effective and deontologically based intervention on problematic behavior must necessarily make functional analysis and identification of the hypothesis of the functional value of behavior the center of one's line of thought and work. For this reason, there are various functional assessment methodologies aimed at providing data to answer fundamental questions,

such as: why does this specific behavior exist? What is the objective of that behavior? What is the need or desire that this behavior nurtures? What is the need or desire this behavior responds to? How can we support the same need or desire in a functional way for the person and adapt it with regard to their environment? What is the relationship between the challenging behavior and the context in which it occurs?

Understanding or identifying the hypothesis of the functional value of behavior will thus allow us to select psychoeducational procedures and, in some cases, effective pharmacological interventions, to reduce challenging behavior. However, it will, above all, show us which new behaviors should be taught and which should be promoted, or which new opportunities should be granted to the person.

This is the purpose of the functional assessment which, due to the complexity of behaviors and the life contexts in which they are potentially operated, may use different analysis tools that can be classified into two main categories: direct and indirect.

With indirect measures, information on the functional value of behavior is gathered through questionnaires, checklists, or interviews with parents, caregivers, and people who know the person with ASD well. Some of the tools used are as follows:

- The Motivation Assessment Scale (MAS, [693])
- The Functional Analysis Screening Tool [694]
- The Setting Events Checklist [695]

Direct measures, in which the behavior is directly observed by the professional or the parent, are carried out with the aim of verifying the presence of a behavior at the moment of its occurrence, that is, in real time, in a specific context.

ABC, which stands for antecedents-behavior-consequences, is the best-known form of functional behavior assessment. In order to understand ABC, it is necessary to start by focusing on B: the observable behavior. Then, A denotes the antecedent, which is an event or events that occur before the behavior begins, and C identifies the consequence that follows the behavior. As part of direct functional analysis, functional analysis for experimental or analogous conditions is particularly effective. This system of analysis is particularly useful in the analysis of hetero-aggressive and destructive behaviors. The experimental functional analysis implies the manipulation of the antecedents and/or consequences of the behavior in order to identify a functional relationship by testing the hypothesis of functional value between these contextual variables and the behavior itself.

4. The hypothesis of functional value and integrated intervention

This phase of the intervention, aside from being the most important, is also the most complex because it requires the professional to analyze the collected data and to construct hypotheses on the adaptive value of the problem behavior using their own analytical skills as regards that specific case.

Although not infinite, the functional values of a specific challenging behavior are often multiple and sometimes they can also be changing in time. These behaviors, like any other, may be related to the consequences of behavior observed in the external world, rather than having consequences on the internal world, that is, emotions, one's belief systems, values, and symbolic systems. The reason why the challenging behavior remains active can also be attributed to elements linked not only to the learning history, but also to physiological variables, for example, the pain or the variable of a reduced or qualitatively poor sleep. For this reason, in this phase of analysis of the hypothesis of the functional value of the behavior, it is important to share with the whole team, including with those figures that have specific professional skills (e.g., the medical doctor and the nurse). The joint work of the team will define more completely and correctly the different hypotheses that underpin the problem behavior and also share and participate in pharmacological and psychoeducational treatment procedures, as well as discuss jointly and constantly the data collected during the interventions.

5. Nonpharmacological intervention implementation

The consequent step after identifying the functional value of the challenging behavior is the choice of nonpharmacological interventions and the continuation of data collection to verify the effectiveness of treatments. There are many nonpharmacological interventions that can be used for the development of new behaviors and the reduction of maladaptive behaviors; in Cooper, Heron, and Heward [696], several hundred interventions are mentioned, and the selection of the most appropriate one must be carried out by paying attention to the consistency between the chosen procedure and the adaptive/disadaptive value of behaviors that are to be increased and/or decreased. In the choice of interventions, which are often multicomponent, it is equally important to remember to maintain a proportion between proactive procedures (80%) and reactive procedures (maximum 20%). Indeed, even if the opposite is often the case in clinical practice, an effective intervention program that respects the quality of life of the person and their context should favor more proactive interventions and limit the detriment of reactive ones.

In general, any nonpharmacological intervention should be chosen and implemented in accordance to tailored, dynamic, and developmentally phased life-span approaches that recognize and focus on periods of increased or decreased specific needs, as well as change monitoring over time in the individual or their family. The need of designing efficient delivery systems for complicated therapies has recently emerged as a prominent aspect of intervention research [697].

Although adults make up the majority of autistic people, because adulthood is a much longer stage of life, services and support for adults with autism are significantly fewer than for children and adolescents, and very few programs have been extensively assessed [698]. Addressing the needs of autistic people currently necessitates collaboration among local communities and stakeholders, as well as researchers and clinicians involved in the creation of relevant programs and systemic change. Adults have proved to benefit from treatments comparable to those used with adolescents, such as cognitive behavior therapy and social skills groups [699, 700]. Supported employment and job coaching programs are available in several parts of the world, and proof of their usefulness is expanding [701, 702]. Adult adaptive skill development programs and systems exist, although they are seldom recorded in research [703]. For many years, behavioural programs for adults with ASD have been documented, although few are evidence-based, and many involve people with co-occurrence of severe intellectual disability. Furthermore, some neurodiversity advocates disagree with the use of many behavioral techniques [704]. Services for adults with significant support needs appear to be the most crucial area in several high-income countries, and they require major improvement. In low- and middle-income countries requirements are even broader and encompass to the entire life span.

Nonpharmacological treatments cannot rely only on the expertise of autism specialists. In fact, non-specialized professionals are currently providing the majority of treatment for persons with ASD of all ages, even in high-income countries; most care is done in school and community settings, with or without professional input or assistance [705]. As a result, non-expert training and assistance must be included in research and system planning. Understanding what works for whom and when, as well as some of the expected requirements and differences that must be recognized while supporting persons with ASD, are critical elements [697].

16.7.2.3 Successes and New Challenges in Nonpharmacological Treatment

Treatments based on contextual sciences and applied behavioral analysis have changed the way we act toward ASD, moving from symptom containment to building abilities to improving adaptive skills and interaction. For example, many people with ASD can now develop a communication system (verbal, by images, or gestures) that allows them to improve their interaction with the social environment. The new challenges concern the diffusion of these interventions that require

years of training and high specialization of the professionals and their application not only in the early interventions: the acquisition of skills opens new scenarios, the possibility of supporting the person with ASD in the transition through adult age for an independent life and an inclusive job. In addition, interventions and professionals will be needed to allow a response to adult behavior and psychopathologies in people with ASD.

Because the bulk of behavioral treatments include numerous components, they can be complicated and articulated. As a result, it is advised that measurements be developed that enable for rigorous analyses and uniform change and progress tracking across time. Innovative methodological approaches, such as the Multiphase Optimization Strategy and Sequential Multiple Assessment Trial, have emerged to assess treatment effectiveness, define delivery strategies within complex interventions, and investigate the effects of each treatment on persons with ASD.

The Multiphase Optimization Strategy is a methodological approach for systematically and efficiently optimizing behavioral interventions by determining which components of a targeted intervention are active and potentially contributing to positive outcomes, and which dosage of each component produces the best results for a specific individual.

Sequential Multiple Assessment Trial is a randomized experimental design for time-varying adaptive interventions planning. This method may be used to determine the best intervention component sequencing and how frequently active components within the intervention should be reviewed. The treatment sequence for each individual is designed based on observed outcomes and is continually examined and recalibrated over time depending on the individual's reaction to therapy. Individual characteristics such as intensity of symptoms, preferences, and peculiarities are utilized to tailor the intervention to a particular person, and then individual outcomes (e.g., response to treatment, adherence) are used to dynamically adjust the intervention to maximize its efficacy. Furthermore, if the individual is not responding to a focused treatment, it is vital to gradually increase the intensity and kind of intervention in order to make progress or decrease new difficulties.

Lord and colleagues [697] proposed an innovative, adapted, health-integrated stepped care model for ASD intervention, with the goal of providing effective clinical practice approaches and rigorous clinically oriented research strategies that can have a positive short- and long-term impact on autistic people and their families while avoiding under- or over-treatment. The stepped care and personalized health model emphasizes autism's heterogeneity by recognizing that each autistic individual's and family's specific and unique profile of strengths, needs, and interests should guide intervention and support priorities, but that these can change over time as development and interventions produce effects. Furthermore, because ASD is so mutable, what works now for one person may not work later, and vice versa. By optimizing resource allocation, this phased and customized treatment model can be gradually calibrated as demands and needs emerge, change, and evolve over time, progressing from broader interventions to more specific and increasingly intense treatments tailored to specific personal needs and goals.

Multidisciplinary treatment needs to coordinate, share, and participate in both the nonpharmacological and pharmacological treatment approaches, as well as discuss jointly and constantly the data collected during these interventions.

Tip

Despite advances in increased awareness and research progress in early identification and diagnosis of ASD, harmonizing early interventions and services across the lifespan has remained a paramount challenge in the light of clinical heterogeneity of ASD and its variable treatment response, beyond the important global public health questions of within- and between- country disparities in terms of proximity to services.

Key Points

- ASD represents a highly heterogeneous group of lifelong neurodevelopmental disorders.
- There have been staggering strides made in increasing awareness of ASD worldwide.
- Understanding of factors associated with individual differences in response to early intervention is an important priority.
- Large gaps and disparities also remain in the dissemination of the evidence base on ASD, and in particular, in terms of implementation of effective treatment across the lifespan both within and across countries.
- The high rate of associated medical and psychiatric conditions represents a major burden on affected individuals and their families.
- Both pharmacological and nonpharmacological treatments can help reduce the impact of associated comorbidities.
- Person-centered, and multidisciplinary care services, as well as promotion of inclusionary social environment, need to be a universal standard worldwide.
- However, ASD cannot be effectively addressed by the health sector alone and national policies require collaboration across health, education, and social sectors.
- Access to public education, vocational training, and assisted employment with the goal of social inclusion is of paramount importance.

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Attention Deficit Hyperactivity Disorder

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Contents

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Learning Objectives

- To learn about the prevalence of ADHD in people with ID and ASD.
- To understand difficulties diagnosing ADHD in people with ID and low-functioning ASD, including distinguishing comorbidity from the differential diagnosis.
- To learn about specific presentation of ADHD in people with ID, including those with more severe cognitive and communication impairments.
- To understand the complex aetiology of ADHD in people with ID, including multiple genes, non-inherited factors, and their interplay.
- To learn about the current status of research and its impact on the advancement of treatment in this patient group.
- To be knowledgeable about available pharmacological and non-pharmacological treatment of ADHD in people with ID.

17.1 Introduction

There is a common misconception that attention deficit hyperactivity disorder (ADHD) is a relatively new diagnosis in modern society. However, there are well-documented cases of ADHD over 200 years. There are records describing abnormalities in attention as early as in the eighteenth century [1, 2]. Dr. George Still [3] was one of the first physicians to describe an 'ADHD-like' syndrome. He described a 'defect of moral control' in a series of lectures to the Royal College of Physicians in 1902. In 1937, Benzedrine, an amphetamine compound, was found to be effective in children with behavioural disorders. This is one of the earliest recorded medical treatments for the behaviour associated with ADHD [4]. In 1972, a case report was published of a 22-year-old man who was diagnosed with the hyperkinetic syndrome [5]. He was reported to have responded to an amphetamine which was described by the authors as a 'pathognomonic paradoxical calming of the hyperkinetic by amphetamine'. Over the next few decades, there have been significant advances in both the diagnosis and treatment of ADHD [6, 7].

Before its current diagnostic nomenclature, ADHD attracted several names including 'minimal brain damage' and hyperkinetic disorder (ICD-10) [8]. The current term used by both the Diagnostic and Statistical Manual of Mental Disorders-fifth Edition (DSM-5) [9] and the International Classification of Diseases-11th version (ICD-11) [10] is 'attention deficit hyperactivity disorder' (ADHD). It is categorised as a 'neurodevelopmental disorder (NDD)', relating to the onset of symptom manifestation occurring during the developmental period. According to the DSM-5 [9] criterion, the onset has to be before age 12 years. However, in the previous version of the DSM [11], the stipulated age of onset used to be before age 7 years. Other NDDs include intellectual disability (ID), autism spectrum disorder (ASD), dyslexia, dyspraxia, and Tics/Tourette's syndrome. NDDs frequently co-occur, hence the importance of recognising ADHD in people with ID as well as ASD [12]. In the previous version of DSM [11], there was no provision for making a diagnosis of ADHD in the presence of ASD. However, in the DSM-5 [9], this criterion has changed, and a diagnosis of ADHD can now be made in the presence of ASD.

ADHD is a phenomenological diagnosis defined by *impaired attention, hyperactivity, and impulsivity*. For a diagnosis of ADHD, symptoms must interfere with the day-to-day functioning of the individual. Symptoms of inattention, hyperactivity, and impulsivity must be experienced to a degree that is more than what is expected for the individuals' developmental age or intelligence. This can make it difficult to establish ADHD in people with ID as low intellect is associated with some of the symptoms of ADHD including inattention. As a result, ADHD can often be missed in people with ID as symptoms can be attributed to the person's low developmental age rather than considering the possibility of ADHD. The challenge for clinicians is to make a considered judgement about whether an individual's ADHD symptoms are greater

than what is expected for the person's level of ID. Despite all these challenges, ADHD is a diagnosis that needs to be part of the differential diagnosis given the evidence base for effective therapeutic strategies [13, 14] and avoidance of other inappropriate treatment [15–17]. Timely diagnosis and treatment can limit or prevent many of the risks associated with ADHD such as significant functional impairment, increased comorbidity with mental illnesses, substance misuse, accidental injury, unemployment, and premature death [18, 19].

Timely diagnosis and treatment can limit or prevent many of the risks associated with ADHD such as significant functional impairment, increased comorbidity with mental illnesses, substance misuse, accidental injury, unemployment, and premature death. The identification of ADHD can also prevent inappropriate treatment with antipsychotics, which is rather common in people with ID/ASD and problem behaviours.

17.2 Trajectory of ADHD

ADHD was initially thought to be a childhood disorder. In recent times, there has been a significant increase in the literature showing that ADHD symptoms can persist into adulthood and can cause functional impairment [20]. Sibley and colleagues [21] showed that 60% of the studied ADHD children continued to experience ADHD symptoms into adulthood and 41% continued to meet both symptom and impairment criteria. Lower IQ was found to be one of the predictors of persistence of childhood ADHD symptoms into adulthood [22]. Therefore, it is important that ADHD should be considered in adults with ID due to the high rate of persistence and the possibility of missed diagnosis in childhood.

However, to make a diagnosis of ADHD in adulthood for the first time, evidence of the onset of symptoms in childhood before age 12 is required. This may be difficult to establish as the memory of childhood events and behaviours is not always accurate. Whereas ADHD symptoms tend to improve in adulthood in many non-ID children, in a much higher proportion of children with ID the ADHD symptoms tend to persist into their adulthood [23]. One study reported a prevalence of ADHD among 2.9–16.4% of non-ID adults [24] which compares with 23% of 78 community-based young ID adults reported in another study [25]. Whereas in non-ID adults, it is the inattention symptoms that tend to persist into adulthood, in the case of children with ID and ADHD, it is the hyperactivity and impulsive symptoms that tend to persist more into adulthood. However, it is important to distinguish these symptoms from problem behaviour which is prevalent among adults with ID [26] (see ► Chap. 7).

17.3 Prevalence

There have been studies across the world, looking into the prevalence rates of ADHD. Polanczyk and colleagues [28] completed a meta-analysis of available literature on the topic, finding an estimated worldwide prevalence of ADHD in children and adolescents to be 5.29%. Differences in diagnostic criteria between DSM-IV [29] and ICD- 10 [8], as well as changes to diagnostic criteria over the last few decades, have made it difficult to compare studies on prevalence rates. Also included studies used different methodologies and variable sources of information to make the diagnosis. How the level of functional impairment was defined by different researchers also made an impact on the recorded prevalence rates [28]. Recent changes to the ADHD diagnostic criteria in the DSM-5 [9], particularly the reduction in the number of core symptoms required for diagnosis in adults, will likely lead to an increased prevalence rate of ADHD.

According to some studies, ADHD is found to be three to four times more prevalent in ID than in non-ID children and adolescents, the rate of which seems to increase with the increasing level of severity of ID [30–32]. However, it is worth pointing out that ADHD

symptoms are more common than an ADHD diagnosis in the ID population [33]. A large Swedish cohort study found that individuals with an ID were at an increased risk of ADHD compared with those without an ID with an odds ratio of 10.26 [34]. Studies on prevalence rates of ADHD in ID have reported a range of 6.8–28% compared with a prevalence rate of 3–5% for the non-ID general population [35]. Faraone and colleagues [34] found that the high prevalence rates of ADHD in the ID population are associated with genetic factors. Studies have also shown that people with ASD are at a higher risk of having ADHD compared with individuals who do not have ASD with an odds ratio of 22.33 [36]. In a recent systematic review and meta-analysis Lai and colleagues found the overall pooled prevalence estimates of ADHD to be around four times higher in people with ASD than in the general population [(28% (95% CI 25–32) vs 7.2% (95% CI 6.7–7.8), respectively] [37]. It can be hypothesised that the high prevalence rate of ADHD in people with ASD may also be a contributory factor for the increased prevalence of ADHD in ID, given the significantly high prevalence rate of ASD in ID [38].

Studies looking at prevalence rates of ADHD in the non-ID population have shown an increased prevalence in men compared to women. Nøvik and colleagues [39] found the ratio for the male to female varied from 3:1 to 16:1 depending on the country in Europe. A higher male to female ratio of 6:1 was found in adults with ID and ADHD in clinical samples [15]. However, in non-clinical samples the gender difference in the prevalence of ADHD among the ID population is not that prominent apart from the exception when there is a comorbid ASD when the male to female ratio increases to 2–3:1 [23]. Larger studies are needed to further establish the difference in prevalence rate between men and women in the ID population and explore the possible reasons.

Epilepsy is common among people with ID, [40] and it has been shown that comorbid epilepsy increases the ADHD symptoms among people with ID [41]. Antiepileptic drugs such as phenobarbitone and primidone which is metabolised into phenobarbitone also tend to precipitate ADHD symptoms in people with ID [42]. Other antiepileptic drugs such as lamotrigine, levetiracetam, topiramate may also lead to ADHD symptoms in the ID population. However, sometimes the same antiepileptic drugs are used to control problem behaviour and affect dysregulation symptoms that are often coexistent with ADHD in people with ID [43].

17.4 Lack of Evidence

There is only low-quality evidence available for the diagnosis or treatment of mental illness in people with ID [26, 27]. Although ADHD in the non-ID population is a widely researched area, the lack of high-quality research in ADHD in ID has been a barrier to the advancement of treatment in this patient group. Most studies name ID as an exclusion criterion. This has potentially led to a lack of emphasis on ADHD in people with ID even when the clinical presentation is suggestive of possible ADHD.

17.5 Diagnostic Classification

The DSM-5 [9] diagnostic criteria for ADHD include symptoms of inattention, hyperactivity, and impulsivity. Hyperactivity and impulsivity are categorised together, and symptoms of inattention are kept within a separate domain. There are nine symptoms in both the hyperactivity/impulsivity domain and the inattention domain. Six or more symptoms of inattention and/or hyperactivity and impulsivity are required for the diagnosis of ADHD in childhood. However, the number of symptoms required to make a diagnosis in an adult over the age of 17 is five from one or both domains. Symptoms need to be developmentally inappropriate which is an important consideration with regard to an individual with lower intellectual functioning. When diagnosing ADHD in an adult, there should be evidence that at least three of the symptoms were present before the age of 12. Symptoms of inattention and/or hyperactivity

and impulsivity need to be present in two or more settings.

For a diagnosis of ADHD to be made, symptoms should cause functional impairment. Defining the functional impairment poses challenges due to subjectivity. Difficulties in education, employment, relationships, and day-to-day activities are considered as some of the areas for assessment of functional impairment. ADHD symptoms can also cause functional impairment in an individual's mental health by causing emotional distress, poor self-esteem, and mental illness such as mood or anxiety disorders. Functional impairment in people with ID and ADHD needs to be assessed in the context of the level of functioning expected for the person's level of ID. People with ID may not have the same level of responsibility and roles in the community as people without an ID. It has been proposed that functional impairment can manifest as challenging behaviour in people with ID [16, 44]. However, one drawback of applying the DSM-5 [9] criteria to people with ID is that, as these symptoms are highly correlated, some may have a total score of 6–10 according to the DSM-5 [9] criteria yet scored less than 6 in each section of inattention and hyperactivity/impulsivity. These people may be more impaired with a higher rate of psychopathology than those who may have a score of more than 6 in any one of the subsections only [23].

Diagnostic assessment includes a detailed neurodevelopmental and psychiatric history followed by specific questions on ADHD symptoms to assess whether a person meets the criteria for ADHD. Perera and colleagues [45] in their study showed that strict application of DSM-5 [9] criteria can lead to low sensitivity (missing the diagnosis of ADHD in people with ID) compared with clinician's overall opinion which had high sensitivity and specificity. Reasons, why the strict application of DSM-5 criteria can lead to underdiagnosis, are examined below.

A comprehensive report aimed at guiding clinicians working with adults with ID to diagnose and treat co-occurrent ADHD has just been published by the Royal College of Psychiatrists (RCPsych), the UK, based on literature review and experts' opinion. It includes adaptations of DSM-5 diagnostic criteria for application to adults with ID [46] (see ◘ Tables 17.1 and 17.2).

Attention deficit hyperactivity disorder (ADHD) is characterised by inattention or hyperactivity/ impulsivity or both.

There is growing evidence that people with ID are at a higher risk of comorbid ADHD compared with people without ID. In general, there is a strong association among ADHD, ID, problem behaviour, and ASD.

The onset of ADHD is in childhood, but in a proportion of cases, some symptoms continue into adulthood.

17.6 Clinical Symptoms

17.6.1 Inattention

Inability to stay focused on tasks, disorganisation, careless mistakes, difficulty following instructions, difficulties in listening, forgetfulness, losing materials, and avoidance of tasks needing sustained attention are symptoms that come under the inattention domain. Some of these symptoms are difficult to assess in people with ID. Therefore, symptoms of inattention can be easily missed or wrongly attributed to having the 'usual' inattention symptoms in people with ID. Inability to pay and sustain attention is a subjective complaint which people with ID may struggle to communicate. Observation of a person's distractibility that is not consistent with their level of development may help when considering the symptom of inattention in an individual with ID (see ◘ Table 17.1). A more pragmatic approach can be helpful when assessing an individual with ID for ADHD. Reports from colleagues, schools, or day centre staff can be invaluable in differentiating between a person with ID alone and a person with the same level of ID and ADHD. Difficulties in sustaining attention and distractibility were found to be the most commonly recognised DSM-5 [9] inattentive symptoms by clinicians diagnosing ADHD in people with ID [45, 46].

Table 17.1 Specific presentation of inattention in persons with ID and ADHD as compared to DSM-5 ADHD diagnostic criteria

DSM-5 criteria [9]	Specific presentation in persons with ID
Often fails to give close attention to details or makes careless mistakes in schoolwork, at work, or with other activities	Struggles to pay attention at school, work, home, and during occupational therapy and other activities
Often has trouble holding attention on tasks or play activities	Difficulty sustaining attention at work, home, and during occupational therapy and other activities
Often does not seem to listen when spoken to directly	Appears not to listen to the conversation, asks for the information again, cannot remember parts of the conversation
Often does not follow through on instructions and fails to finish schoolwork, chores, or duties in the workplace, e.g. loses focus, side-tracked	Starts an activity but does not complete the task. Needs a lot of support and prompting to complete tasks
Often has trouble organising tasks and activities	Observation of a person's difficulty in planning simple tasks and following the steps required to complete tasks in the proper sequence, which is not consistent with their level of development. This symptom does not apply to persons with more severe ID, who are rarely assigned responsibility for organising chores or activities
Often avoids, dislikes, or is reluctant to do tasks that require mental effort over a long period of time (such as schoolwork or homework)	Struggles to complete a task or activity that takes a long period of time or involves concentration
Often loses things necessary for tasks and activities, e.g. school materials, pencils, books, tools, wallets, keys, paperwork, eyeglasses, mobile telephones	Will leave belongings behind even when aware of what is needed for the day. Will not know where something is if it has not been put in its usual place
Is often easily distracted	Gets distracted by every little stimulus. Quickly switches from one task to the next and find it difficult to return to an activity in which they were previously engaged
Is often forgetful in daily activities	Forgets where they have put objects, including favourite items from everyday life, and how to carry out activities, particularly those that are not routine

Adapted from McCarthy et al.[47] and RCPsych [46]

Case Study 17.1

An 18-year-old man with severe ID and autism presents with a long-standing history of behavioural difficulties. He is supported 24 hours a day by caregivers. Assessing symptoms of inattention can be difficult in a person with severe ID. However, close observations by staff who look after him reported that he needs more one-to-one support to keep him focused on tasks compared with other people with a similar level of ID. He was also described as more distractible compared with others. Inattentive symptoms shown by this 18-year-old man can easily be attributed to the person's level of ID. It is challenging to assess whether a person's level of inattention is proportionate to their level of ID as there are no objective measures. This patient was diagnosed with ADHD and was given treatment for ADHD. There was a significant improvement in his level of attention. He was able to sit and enjoy activities. He started to engage in various outdoor activities which were not possible before treatment due to his behaviour. Fewer restrictions were needed to manage behavioural difficulties

Table 17.2 Specific presentation of hyperactivity and impulsivity in persons with ID and ADHD as compared to DSM-5 ADHD diagnostic criteria

DSM-5 criteria [9]	Specific presentation in persons with ID
Often fidgets, taps hands or feet, or squirms in seat	Often cannot be still; always moving around
Often leaves seat in situations when remaining seated is expected	Moves around a lot and finds it difficult to sit still for lengthy periods of time, even while eating or bathing
Often runs about or climbs in situations where it is not appropriate (adolescents or adults may be limited to feeling restless)	Moves around most of the time, frequently changes location or body position, or take objects, even in situations when it is particularly improper such as a waiting area or a public office; internal restlessness can lead to psychomotor agitation, escape, or aggressive behaviour
Often unable to play or take part quietly in leisure activities	Speaks loudly, emits noisy phonations, or behaves inappropriately during leisure activities
Is often 'on the go' acting as if 'driven by a motor'	Shows excessive energy, often moves around, is not able to sit in one place for long, likes to be outdoors most of the time engaging in activities
Often talks excessively	Talks a lot without taking turns or emits continuous phonations while other persons are trying to interact with him
Often blurts out answers before a question has been completed	Blurts out answer to any question asked or physically react before other persons complete their communicative act
Often has trouble waiting his/her turn	Becomes agitated or aggressive when asked to wait for any length of time. Not able to queue for activities, finds it hard to wait if their needs are not met immediately
Often interrupts or intrudes on others	Acts as though he/she lacks a sense of personal space, interrupting others' discussions and actions

Adapted from McCarthy et al.[47] and RCP [46]

17.6.2 Hyperactivity and Impulsivity

The symptoms of hyperactivity and impulsivity include overactivity, inability to sit in one place for long, frequent fidgeting, on the go, talking excessively, intruding into the space or conversations of other people, impatience, and the inability to wait and take a turn during a conversation. Hyperactivity may lead to aggression such as hitting, kicking, biting, pinching others and self, and also arguing with others by people who can speak. Again, these symptoms need to be developmentally inappropriate to consider the diagnosis of ADHD. In contrast to inattention, hyperactivity and impulsivity symptoms can be observed in a person with ID. For example, if they fidget most of the time, leave their seat in situations when they are expected to remain seated or are described as 'on the go'. However, examples such as excessive talking or blurting out answers may not be relevant to people with limited or non-verbal communication skills [35]. An individual's symptoms of impulsivity can be manifested as their inability to wait which can often lead to verbal or physical aggression, irritability, mood fluctuations, or self-harming behaviour [48]. 'Often leaving a situation when remaining seated is expected' and 'on the go' were found to be the most commonly recognised DSM-5 [9] hyperactivity symptoms in people with ADHD and ID [45] (see Table 17.2).

Although inattention is more prevalent in both the ID and the non-ID population, but

as hyperactivity/impulsiveness is associated with problem behaviour, they are more prevalent among the tertiary referrals in the ID clinics, possibly sometimes giving a false sense of a higher prevalence of hyperactivity symptoms in this population. However, hyperactivity symptoms and dysregulated affect cause more impairment and morbidity in people with ID [23].

Case Study 17.2

A 25-year-old man with moderate ID, autism, and anxiety disorder presents with challenging behaviour going back to early childhood. During the assessment of his behaviour, his mother reported how hyperactive he has been all his life. These symptoms were never mentioned in the past as his mother often considered his hyperactivity as part of his ID. Most professionals also overlooked his hyperactive behaviour. Further questions elicited symptoms such as his inability to sit in one place, how busy he is most of the time, and his impulsivity/impatience. A diagnosis of ADHD was made and treated which led to a clear improvement in his behaviour and improved his quality of life

17.7 Diagnostic Validity

The validity of an ADHD diagnosis in the presence of ID has been debated both in the literature and in clinical practice, and to some extent, this issue still remains unresolved. According to the older versions of the DSM [11], ADHD diagnosis could not be made in a person with ID. There are three main areas of confusion, namely (a) an overlap between ID and ADHD symptoms, (b) an overlap between ADHD symptoms and problem behaviour in ID, and (c) the validity of using ADHD diagnostic criteria for people with ID that have been developed based on the studies of people who do not have ID.

Both inattention and hyperactivity are non-specific symptoms that are found in many psychiatric disorders. These symptoms are also common in people with ID which may lead to a 'diagnostic overshadowing' [49] in that even when these symptoms are due to ADHD, they are wrongly attributed to the ID diagnosis. This may lead to an underdiagnosis of ADHD in the ID population. However, as both ID and ADHD are classified under the same hierarchical diagnostic category of 'neurodevelopmental disorder', some symptoms overlap will be expected. Similarly, both conditions may share some genetic as well as environmental aetiologies such as pregnancy related Foetal Alcohol Spectrum Disorder (FASD) or obstetric complication [50]. Underlying brain impairment has also been shown to be similar in both conditions affecting the frontal cortex, basal ganglion, and cerebellum [51]. One study has shown abnormalities in the posterior part of the corpus callosum in people with ADHD and ID associated with velocardiofacial syndrome [51]. However, in the absence of a control group, it is difficult to comment on the specificity of this finding in relation to a combined diagnosis of ID and ADHD.

To establish a separate diagnosis of ADHD in the presence of ID, one has to establish that inattention and hyperactivity/impulsive symptoms are more frequent and severe than what would be expected for the person's level of intellectual capacity. To do that one has to establish the age appropriateness of these symptoms, but there is debate as to whether this should be based on the chronological or developmental age of the person. Developmental age appropriateness may be difficult to establish. Other ways to distinguish the two diagnoses would be to look at the similarities or differences in their (a) symptom profile, (b) trajectory of symptoms, (c) level of associated impairment, (d) pattern of comorbidity, (e) family history, (f) laboratory findings, (g) neuropsychological findings, and (h) treatment response [52, 53].

Core symptoms of ADHD seem to be similar in the presence or absence of ID, but anxiety symptoms may be more prominent in the presence of ID [54]. The factor structure of the ADHD symptoms seems to be the same in the ID and the non-ID population. Using Conners' Parent Rating Scale (CPRS-R) [55],

Deb and colleagues [33] found three factors, namely, inattention, hyperactivity, and conduct disorder among children and adolescents with ID and ADHD. The same three factors were described by Conners and colleagues [55, 56] in the non-ID population with ADHD.

The trajectory of ADHD symptoms, by and large, seems to be similar in the ID and the non-ID population. Hyperactivity/impulsivity symptoms tend to appear at an early age and then diminish as the child grows old. On the other hand, inattention symptoms tend to appear later in childhood perhaps when the child is under academic pressure and tend to persist over time. In the ID population hyperactivity symptoms may be identified more in adulthood as they cause more problems for the service in the form of problem behaviour. In contrast, inattention symptoms may be detected more in the non-ID population in adulthood as they cause more impairment in the person's occupational and social life [57].

The DSM-5 [9] criteria stipulate that to confirm a diagnosis, ADHD symptoms must cause impairment in areas such as communication, daily living skills, social functioning, and educational/occupational achievement. However, the impairment has to be age-inappropriate, and in the case of ID, developmental age inappropriateness may not be easy to determine. However, people with ID who have a diagnosis of ADHD are shown to have more impairment compared with those who have ID but no ADHD and also who have ADHD in the absence of ID [58].

The most common comorbidity found with ADHD in the non-ID population is oppositional defiant disorder (ODD) [53]. The most common comorbidity with ADHD in the ID population is problem behaviour [16, 33]. Therefore, there is a similarity in the ADHD comorbidity pattern between ID and non-ID populations.

Although a number of family studies exist in the non-ID ADHD population, no such study exists involving ADHD in the ID population [59]. Therefore, it is impossible to draw any conclusion about the influence of family factors on ADHD in the ID population.

As for laboratory findings, although many molecular genetic studies are showing a strong genetic vulnerability for ADHD in the non-ID population [6], no such study exists involving people with ADHD and ID.

Similarly, although there are many studies of neuropsychological impairment associated with ADHD in the non-ID population, there is very little empirical evidence available to support any specific pattern of neuropsychological impairment associated with ADHD in ID children over and above the ID-related deficits [51]. One study has shown impaired sustained attention in ID in the presence of ADHD. However, some argue the validity of the assessment of attention in the presence of a global cognitive deficit as ID and attention may be perceived as part of the same overlapping dimension [50].

Similar treatment response to ADHD symptoms, particularly from psychostimulants such as methylphenidate has been shown in both non-ID and ID children and adolescents although the effect size seems smaller in ID children [60].

As problem behaviour is prevalent in people with ID (see ▶ Chap. 7), in many cases problem behaviour may be mistakenly diagnosed (false-positive diagnosis) as ADHD in people with ID. However, problem behaviour is a common co-occurrence of ADHD in both the ID and the non-ID population [16, 44]. Deb and colleagues [33] found conduct disorder is a common factor associated with ADHD among children with ID. However, if the problem behaviour is a comorbidity of ADHD, this has to be more prevalent than what is expected for the person's level of ID. Also, as Deb and colleagues' [33] factor analysis of Conners' Parent Rating Scale [61] items has shown that problem behaviour/conduct disorder is a separate and distinct factor from inattention and hyperactivity factors among ID children with an ADHD diagnosis, the presence of behaviour problem alone in the absence of any other definite ADHD symptoms is not sufficient to make a diagnosis of ADHD in a person with ID (see Case Studies 17.1 and 17.2).

A diagnosis of ADHD is made in the ID population based on the criteria that were originally developed for the non-ID ADHD population. For example, the validity of ADHD screening and diagnostic instruments used in the non-ID population have not been established in the ID population. However, Deb and colleagues [33] have shown that a commonly used screening tool for ADHD in the non-ID population, CPRS-R (Conners Parent Rating Scale-Revised) [55] scores may distinguish between children with ID and with and without ADHD but not the CTRS-R (Conners Teachers Rating Scale-Revised) [62] scores. However, there are many items in the scale such as 'forgets things she has already learned', 'poor spelling', 'spiteful or vindictive', 'not reading up to par', 'argues with adults', 'has difficulty waiting his/her turn', interrupts or intrudes others', 'does not follow through instructions', and 'fails to finish schoolwork' cannot be verified in people who have severe and profound ID and lack communication skills.

In Deb and colleagues' [33] study Conners Teacher's scale yielded a significantly lower score than the Parent's scale. This may reflect a genuine difference between the two scales as the items are not exactly identical in these two scales. It may also mean that whereas Conners Parent rating scale is to some extent validated in the ID population, Conners Teacher rating scale is not valid for use in the ID population. However, more practical reasons are that the children may behave differently in school and in home, because schools may provide a more structured environment and there may be different types of demand placed on children in different environments. Also, parents may perceive their children's behaviour as less of a problem than the teachers. On the other hand, some parents may suffer from burnout and perceive their children as more of a problem than the teachers do.

Therefore, multiple sources of data collection including the use of rating scales, and direct examination and interview of the child and her or his care giver as well as a history of the development of symptoms over time, the impact of symptoms on adaptive behaviour (communication, education/occupation, daily living skills, socialisation, etc.) are needed to reach a diagnosis of ADHD in people with ID.

- Diagnosis of ADHD in people with ID has gone through a long journey of diagnostic challenges. It has finally been accepted in the current diagnostic criteria such as ICD 10/11 [8, 10] and DSM-5 [9].

  The presence of other NDDs can make the diagnosis more challenging. Current diagnostic criteria need to be adapted when making a diagnosis of ADHD in people with ID.

  Certain criteria are not applicable for people with more severe ID [46].

  Functional impairment of ADHD in ID can be different from that of people without ID as societal and self-expectations are lower when a person has an ID.

17.8 Psychiatric Assessment

The diagnosis of ADHD in people with ID involves obtaining a detailed psychiatric and developmental history followed by a structured assessment of symptoms of inattention, hyperactivity, and impulsivity. People with ID often find it difficult to communicate their symptoms, therefore, collateral history from family and support staff would be invaluable. Observation of behaviour in different settings can also be helpful. The aim of the assessment is to determine if the person with ID is experiencing more inattention, hyperactive, or impulsivity symptoms than is expected for their developmental age, with associated functional impairment [63]. Functional impairment from ADHD in a person with ID can present in different ways, including hyperactivity presenting as challenging behaviour [64] which can impact on their access to activities, use of illicit drugs for self-medication, impulsive aggression, or not achieving their maximum potential. Rating scales for ADHD can supplement clinical findings; however, there are not many validated tools for ADHD in people with ID.

17.9 Rating Scales for ADHD in ID

Taylor and colleagues [65] completed a systematic review of 14 ADHD rating scales that are commonly used for non-ID adults. This review found that only Conners' Adult ADHD Rating Scale [66] and the Wender Utah Rating Scale (short version) [67] had robust psychometric properties and content validity. There is a growing need for a validated screening tool for ADHD in people with ID. The ability to screen may help to recognise ADHD more effectively in people with ID. A consensus-based structured diagnostic interview called DIVA-5-ID has been developed by an international group of experts which has been produced by the DIVA Foundation [68] to guide on assessing symptoms of ADHD in a person with ID using the DSM-5 [9] criteria. This instrument has been adapted from the DIVA-5 structured interview schedule, which was originally developed for the diagnosis of ADHD in the non-ID population. Although it is a useful tool in clinical practice, the true validity and psychometric properties of this instrument in the ID population have not been established yet.

17.10 Aetiology and Associated Conditions

Several genetic and chromosomal disorders are associated with ADHD (► Box 17.1) [69, 70]. It is, therefore, worth considering genetic testing for people with ID to understand the possible aetiology of the person's ID. Recognition of a genetic syndrome that is associated with ADHD can be helpful in the diagnostic process. Genetic syndromes highly associated with ADHD shift the diagnosis of ADHD from a phenomenological to an aetiological concept.

ADHD appears to have a multifactorial aetiology, and most of the risk factors discovered thus far appear to be non-specific. Risks such as chromosomal microdeletions, large, rare copy number variants, having a biological relative with a neurodevelopmental disorder, extreme early adversity, pre–/postnatal exposure to lead or other neurotoxins, preterm birth, and extremely low birth weight appear to affect a variety of neurodevelopmental and psychiatric conditions [71]. However, heritable components seem to play a relevant role in the aetiopathogenesis as siblings are reported to present twice the risk of having ADHD than the general population, and there is a much greater concordance in monozygotic twins than dizygotic [72].

More specific risks factors have been identified in few dopaminergic genes [dopamine active transporter gene (DAT1), dopamine D5 receptor gene (DRD5), and especially dopamine D4 receptor gene (DRD4)], catechol-O-methyltransferase gene (COMT), brain-specific angiogenesis inhibitor 1-associated protein 2 gene (BAIAP2). The functional polymorphism in exon III of the D4 receptor gene is associated with ADHD in different meta-analyses, although with substantial heterogeneity across studies [73–77]. COMT gene and its mutation were associated with decreased grey matter volume, cortical thickness, surface area abnormalities, antisocial behaviour, and high vulnerability to early-life negative events and environment in children with ADHD [71, 78–80]. Some isoforms of the BAIAP2 gene, which encodes for a protein that has been suggested to be involved in cerebral asymmetry at embryonic stages, have been repeatedly linked with ADHD-related cognitive and emotional dysfunctions, including issues of sustained attention, working memory, response inhibition, planning, and anger control [77, 81].

One other well-recognised condition associated with ADHD is foetal alcohol spectrum disorder (FASD). FASD is one of the causes of ID [82]. Hence, a detailed history of maternal use of alcohol during pregnancy to rule out FASD in people with ID is important. A diagnosis of FASD is associated with an increased risk for ADHD (Relative risk of 7.6) [83]. Therefore, the presence of FASD in a person with ID should raise the suspicion of ADHD. FASD is associated with a high prevalence of ADHD of up to 75% [84].

Certain genetic syndromes are associated with ADHD (see paragraph on associated conditions and ► Box 17.1), but most people with ID and ADHD do not have a specific genetic syndrome.

Box 17.1 Genetic Syndromes Associated with ADHD in People with ID

- Downs syndrome.
- 22q 11.2 deletion/velocardiofacial syndrome/DiGeorge syndrome.
- Smith-Magenis syndrome.
- Williams syndrome.
- Angelman syndrome.
- Fragile X/ataxia (FRAXT) (also FRAXA, FRAXE).
- Neurofibromatosis.
- Turner syndrome.
- Klinefelter syndrome.
- Jacobsen syndrome.
- Tuberous sclerosis.
- Foetal alcohol spectrum disorder.

It is important to have a good understanding of common coexistent NDDs with ADHD such as ASD and Tourette's syndrome as they add complexity to the clinical presentation. Hyperarousal and restricted and repetitive behaviours can present as hyperactivity in an individual with ASD. Sensory processing difficulties found in people with ASD can also make someone appear hyperactive and restless at times. Ability to differentiate and understand this comorbidity can help to create a bespoke formulation and treatment plan.

Problem behaviour is prevalent in people with ID (see ▶ Chap. 7), but the rate increases in the presence of ADHD. Both inattention and hyperactivity/impulsive symptoms could lead to problem behaviour either directly or indirectly. Therefore, in practice, it may be quite difficult to distinguish problem behaviour from ADHD in people with ID. In this context the developmental history, for example, the presence of ADHD symptoms in childhood before the onset of problem behaviour, and associated features of ADHD may help with the distinction. Because of these difficulties, some clinicians may go for a trial of psychostimulants to assess any advantage of these medications in improving problem behaviour, although the response to a psychostimulant does not necessarily clarify the diagnosis of ADHD.

In a study of 56,462 adult psychiatric patients, ADHD was diagnosed among 2.7% and ASD among 1.3% of the participants. ADHD was associated more with affective disorder and substance use, whereas ASD was associated more with psychosis. The diagnosis of personality disorder remained the same in ASD and ADHD psychiatric patients in the general population [85].

All these conditions add an additional layer of complexity for diagnosis, formulation, and treatment of ADHD in the ID population.

The aetiologies of ADHD are varied including many genetic disorders.

Comorbidity with other NDDs such as ID and ASD is common in ADHD.

Other psychiatric comorbidities such as affective disorder and substance misuse can also be associated with ADHD.

ADHD can often present as a problem behaviour in individuals with ID, so detailed assessment to rule in or out an ADHD diagnosis is vital given the availability of effective treatments.

17.11 Management of ADHD in ID

Management of ADHD in ID follows biopsychosocial principles. In practice, this means reviewing medication options, psychological strategies and optimising environmental factors (see ▶ Chap. 7).

In the UK, National Institute for Health and Care Excellence (NICE) [86] provides the following guidelines for a baseline assessment before starting a pharmacological treatment for ADHD.

Before starting medication for ADHD, children and adults with ADHD should have a full assessment, which should include:

1. A review to confirm they continue to meet the criteria for ADHD and need treatment.
2. A review of mental health and social circumstances, including:

- Presence of coexisting mental health and neurodevelopmental condition.
- Current educational or employment circumstances.
- Risk assessment for substance misuse and drug diversion.
- Care needs.

3. A review of physical health, including:
   - Medical history, taking into account conditions that may be contraindications for specific medicines.
   - Current medication.
   - Height and weight (measured and recorded against the normal range for age, height, and sex).
   - Baseline pulse and blood pressure (measured with an appropriately sized cuff and compared with the normal range for age).
   - A cardiovascular assessment.

An electrocardiogram (ECG) is not needed before starting stimulants, atomoxetine, or guanfacine unless the person has any of the features in the next paragraph, or a coexisting condition that is being treated with a medicine that may pose an increased cardiac risk. Concerns related to an increased risk of sudden cardiac mortality in adults treated for ADHD have been raised; however, observational studies have not consistently demonstrated this to be the case [87].

Individuals should be referred for a cardiology opinion before starting medication for ADHD if any of the following apply:

1. History of congenital heart disease or previous cardiac surgery.
2. History of sudden death in a first-degree relative under 40 years suggesting a cardiac disease.
3. Shortness of breath on exertion compared with peers.
4. Fainting on exertion or in response to fright or noise.
5. Palpitations that are rapid, regular, and start and stop suddenly (fleeting occasional bumps are usually ectopic and do not need investigation).
6. Chest pain suggesting a cardiac origin.
7. Signs of heart failure.
8. A murmur heard on cardiac examination.
9. Blood pressure that is classified as hypertensive for adults.

Refer to a paediatric hypertension specialist before starting medication for ADHD if blood pressure is consistently above the 95th centile for age and height for children and young people.

In the UK, NICE [86] also provides the following guideline for the choice of pharmacological treatment in the non-ID population, but the same guidelines could be applied to people with ID.

17.11.1 Medication Choice: Children Aged 5 Years and Over and Young People

Offer methylphenidate (either short or long acting) as the first-line pharmacological treatment for children aged 5 years and over and young people with ADHD.

Consider switching to lisdexamfetamine for children aged 5 years and over and young people who have had a 6-week trial of methylphenidate at an adequate dose and not derived enough benefit in terms of reduced ADHD symptoms and associated impairment.

Consider dexamfetamine for children aged 5 years and over and young people whose ADHD symptoms are responding to lisdexamfetamine but who cannot tolerate the longer effect profile.

Offer atomoxetine or guanfacine to children aged 5 years and over and young people if they cannot tolerate methylphenidate or lisdexamfetamine or their symptoms have not responded to separate 6-week trials of lisdexamfetamine and methylphenidate, having considered alternative preparations and adequate doses.

17.11.2 Medication Choice for Adults

Offer lisdexamfetamine or methylphenidate as a first-line pharmacological treatment for adults with ADHD.

Consider switching to lisdexamfetamine for adults who have had a 6-week trial of methylphenidate at an adequate dose but have not derived enough benefit in terms of reduced ADHD symptoms and associated impairment.

Consider switching to methylphenidate for adults who have had a 6-week trial of lisdexamfetamine at an adequate dose but have not derived enough benefit in terms of reduced ADHD symptoms and associated impairment.

Consider dexamfetamine for adults whose ADHD symptoms are responding to lisdexamfetamine but who cannot tolerate the longer effect profile.

Offer atomoxetine to adults if they cannot tolerate lisdexamfetamine or methylphenidate or their symptoms have not responded to separate 6-week trials of lisdexamfetamine and methylphenidate, having considered alternative preparations and adequate doses.

However, in the UK, the trend is to use atomoxetine either as a first-line or second-line treatment after methylphenidate in people with ID, particularly among adults.

17.11.3 Further Medication Choices

Obtain a second opinion or refer to a tertiary service if ADHD symptoms in a child aged 5 years or over, a young person, or an adult are unresponsive to one or more stimulants and one non-stimulant.

Do not offer any of the following medication for ADHD without advice from a tertiary ADHD service:

- Guanfacine for adults.
- Clonidine for children with ADHD and sleep disturbance, rages, or tics.
- Atypical antipsychotics in addition to stimulants for people with ADHD and coexisting pervasive aggression, rages, or irritability.
- Medication not included in recommendations.

17.11.4 Medication Choice: People with Coexisting Conditions

Offer the same medication choices to people with ADHD and anxiety disorder, tic disorder, or ASD as other people with ADHD.

Addressing an eventual comorbid anxiety disorder before taking ADHD medication is recommended since there is a risk of increased anxiety and deteriorating behaviour. When initiating ADHD medication, treat anxiety first or utilise medications/strategies to lessen anxiety. Stop ADHD medication if anxiety and associated problem behaviours increase, then continue when the person's anxiety is under control [46].

For children aged 5 years and over, young people, and adults with ADHD experiencing an acute psychotic or manic episode, stop any medication for ADHD and consider restarting or starting new ADHD medication after the episode has resolved, taking into account the individual circumstances, risks, and benefits of the ADHD medication.

17.11.5 Considerations When Prescribing ADHD Medication

When prescribing stimulants for ADHD, think about modified-release once-daily preparations for the following reasons:

1. Convenience.
2. Improving adherence.
3. Reducing stigma (because there is no need to take medication at school or in the workplace).
4. Reducing problems of storing and administering controlled drugs at school.
5. The risk of stimulant misuse and diversion with immediate-release preparations.
6. Their pharmacokinetic profiles.

Immediate-release preparations may be suitable if more flexible dosing regimens are needed, or during initial titration to determine correct dosing levels.

As we have recommended a lower starting dose and slower titration rate, a modified-release preparation may not be suitable as the starting option in many children and adults with ID.

When prescribing stimulants for ADHD, be aware that effect size, duration of effect, and adverse effects vary from person to person.

Think about using immediate- and modified-release preparations of stimulants to optimise effect (e.g. a modified-release preparation of methylphenidate in the morning and an immediate-release preparation of methylphenidate at another time of the day to extend the duration of effect).

Be cautious about prescribing stimulants for ADHD if there is a risk of diversion for cognitive enhancement or appetite suppression.

Do not offer immediate-release stimulants or modified-release stimulants that can be easily injected or insufflated if there is a risk of stimulant misuse or diversion.

Prescribers should be familiar with the requirements of controlled drug legislation governing the prescription and supply of stimulants.

17.12 Pharmacological Management

There are broadly two classes of medications used to treat ADHD: psychostimulants and non-psychostimulants. Cortese and colleagues [88] in their network meta-analysis found 133 double-blind placebo-controlled trials on ADHD medications in the non-ID population based on 10,068 children and adolescents and 8131 adults. However, there are only a limited number of studies on people with ID and ADHD [60]. All the major studies have ID as an exclusion criterion. Therefore, treatment recommendations for ADHD in people with ID are derived from studies from the non-ID population.

A recent systematic review on the effectiveness of methylphenidate in the management of ADHD in people with ID [60] found 13 RCTs, only one of which used parallel design, and the rest are small crossover trials. Overall the quality of evidence is poor. All RCTs included children and none are on adults. It seems that overall the effect size of methylphenidate in children with ID (average around 0.5) is lower than that in the children who do not have ID (average around 0.8–1.3).

Significant adverse effects included sleep difficulties and poor appetite along with weight loss. Other important adverse effects included irritability, social withdrawal, and increased motor activities including tic. The type and the rate of adverse effects among children with ID seem similar to those in the children who do not have an ID (average around 12–24%). Similarly, the placebo effect seen among children with ID seems to be similar to that in the children who do not have an ID (average around 12.5%).

In the past, there was concern that methylphenidate may worsen the symptoms of ASD. However, recent studies [89] showed that methylphenidate could be effective in the general non-ID population of children with ASD (effect size is around 0.5), similar to what has been reported in children with ID [60]. Although a proportion of participants showed adverse events mostly mild, overall it seems that generally methylphenidate was tolerated by most children with ASD. In fact, some studies have shown improvement in core ASD symptoms, such as social communication and self-regulation in children who were treated with methylphenidate [89].

There is some concern that methylphenidate may make epilepsy (which is common among people with ID) [90] worse and interact with antiepileptic medication, although small-scale studies did not show any such adverse effects. There is also some debate as to whether the level of ID is a modifying factor for response to treatment with methylphenidate although this has not been proven unequivocally. Apart from the core symptoms, methylphenidate seems to have some effect specifically on problem behaviour in children who have ADHD but not ID [91]. Among children with ADHD and ID, whereas some studies showed some improvement in problem

behaviour while treated with methylphenidate, other studies have failed to do so [60].

Although not conclusive, it is possible that methylphenidate may have a better effect at a higher dose but possibly at the expense of a higher rate of adverse effects. There is some doubt whether concomitant use of other psychotropic medication (which is common in people with ID) reduces the efficacy of methylphenidate [92]. It is also possible that by countering some of the adverse events such as weight loss and sleep impairment, antipsychotics may indirectly show a better result when used concomitantly with methylphenidate.

17.12.1 Psychostimulants

The most commonly used psychostimulants are methylphenidate and amphetamine. Both amphetamine and methylphenidate act as noradrenaline (norepinephrine) and dopamine reuptake inhibitors, and they both increase monoamine neurotransmitters in the prefrontal cortex. In addition, amphetamine releases dopamine to the synaptic cleft. Methylphenidate and lisdexamfetamine have a strong evidence base with many trials showing efficacy in non-ID people. There are no RCTs in adults with ID for stimulant medications [60, 88].

17.12.1.1 Methylphenidate

Tarrant and colleagues [60] in their systematic review on methylphenidate found only one good quality study parallel design RCT in children with ID. They found that overall the effect size of methylphenidate is lower (0.5) in children with ADHD and ID compared with children without ID [60]. The low response rate in people with ID could be due to various reasons. Aman and colleagues [93] suggested whether the low response rate is due to the use of low doses of methylphenidate in studies done in people with ID. Tarrant and colleagues [60] have cautioned that the findings from the systematic review should be used carefully as studies used in the review were of poor quality.

A crossover trial showed that methylphenidate was effective in reducing ADHD symptoms in four children with FASD [94]. A small crossover trial suggested that methylphenidate might improve cognition and ADHD symptoms in children with velocardiofacial syndrome [95].

17.12.1.2 Different Methylphenidate Preparations

This is the drug of the first choice. This is available as immediate-release and prolonged-release preparations.

Child 6–17 years: Initially 5 mgs one to two times a day, dose increased in steps of 5–10 mgs daily if required, at weekly intervals to a dose of 60 mgs daily, in two to three divided doses, up to 2.1 mgs/kg, higher dose, maximum of 90 mgs daily, under specialist supervision, for a month, to be discontinued if there is no response. If the effect wears off by bedtime with rebound hyperactivity, a bedtime dose may be appropriate (establish need with a trial bedtime dose).

Adults: Initially 5 mg two to three times a day, dose increased at weekly intervals according to response up to a maximum of 100 mgs daily in two to three divided doses, if the effect wears off in the evening with rebound hyperactivity, a bedtime dose may be appropriate. It may be necessary to establish the need with a trial bedtime dose.

Treatment may be established with a modified preparation that contains immediate and modified release components in different proportions to cover the day. The modified release preparations are given in the morning. When switching from immediate to modified release preparations, it is important to check product literature.

Common side effects include decreased appetite, hypertension, growth retardation in children, anxiety, arrhythmias, arthralgia, behavioural difficulties, depression, diarrhoea, dizziness, drowsiness, dry mouth, fever, gastrointestinal discomfort, headaches, nausea, palpitations, sleep problems, vomiting, and loss of weight.

Monitoring requirements: Pulse, blood pressure, psychiatric symptoms, appetite, weight,

and height should be recorded at the initiation of therapy, following each dose adjustment, and at least every 6 months thereafter.

Treatment cessation: avoid abrupt withdrawal.

Methylphenidate is available in short- and long-acting preparations. Short-acting preparations are effective for up to 4 hours. The duration of long-acting preparations varies according to the drug. Short- and long-acting methylphenidates are available in different brands. Ritalin and Medikinet are some of the short-acting brands available in the UK. Concerta XL, Medikinet XL, and Equasym XL are some of the commonly prescribed long-acting methylphenidate preparations in the UK. Concerta XL, Delmosart, and Xaggitin XL are available as 18, 27, 36, and 54 mgs tablets, while Equasym XL capsules are available as 5, 10, 20, 30, 40, 50, and 60 mgs capsules (British National Formulary, BNF 76; ► www.bnf.org). Concerta XL has 8–10-hour duration of action with 22% of the drug released immediately and the rest released slowly over next 8–10 hours. Medikinet XL has 6–8-hour duration of action with 50% of the drug released immediately and the rest released over next several hours. Equasym XL has a similar duration of action to Medikinet XL, but only 30% of the drug is released immediately with 70% released slowly. There are other different brands in the market with similar durations of actions and modified release preparations. These long-acting preparations help people to take one tablet a day as their effect lasts for several hours rather than having to take extra tablets to take to school, day centre, or work. For patients with swallowing difficulties, certain ADHD-modified release preparations (Medikinet XL) can be taken out of their capsules and sprinkled over food.

17.12.1.3 Dexamfetamine

Dexamfetamine is available as short- and long-acting preparations. Short-acting dexamfetamine acts for up to 4 hours. Therefore, several doses are needed to cover longer duration of symptom control. Long-acting preparation available in the UK is called lisdexamfetamine which lasts for up to 14 hours. Dexamfetamine sulphate is available as oral solution, tablets 5, 10, and 20 mgs and as modified release spansules 5, 10, and 15 mgs (BNF, 76; ► www.bnf.org).

Evidence base again is limited for dexamfetamine use in ID [96]. One Cochrane review [97] on amphetamine in children with ADHD and ID only found one study looking at 15 children. This study was limited for 1 week and did not find any difference between treatment and placebo-controlled group. Lack of evidence is again another limitation with regard to the use of dexamfetamine in people with ID.

A small crossover trial with methylphenidate, dextroamphetamine, and placebo suggested that methylphenidate might be more effective and safer than dextroamphetamine in boys with Fragile X syndrome (FXS) [98]. Another study described that dextroamphetamine reduced hyperactivity compared with placebo in a boy with a mild ID [99].

Child: 6–17 years: Initially 2.5 mgs two to three times a day increased in steps of 5 mgs/day weekly if necessary up to 1 mg/kg daily, maintenance dose to be given in two to four divided doses, up to 20 mgs daily: 40 mgs may be necessary in some children.

Adults: Initially 5 mgs twice daily, dose increased weekly depending on the response, maintenance dose in two to four divided doses, maximum 60 mgs per day.

Common side effects include abdominal pain, anxiety, decreased appetite, arrhythmias, joint pain, behaviour problems, depression, dry mouth, headache, altered mood, movement disorders, muscle cramps, nausea, palpitations, poor weight gain, sleep problems, vertigo, vomiting, and weight loss.

Monitoring requirements include growth in children, aggressive behaviour, or hostility. Pulse, blood pressure, psychiatric symptoms, appetite, height, and weight should be recorded at initiation, following each dose adjustment and at least after every 6 months thereafter. Treatment should not be withdrawn abruptly.

17.12.1.4 Lisdexamfetamine Mesilate

This is a prodrug of dexamfetamine.

Child: 6–17 years initially 30 mgs once daily, alternatively, initially 20 mgs once daily,

increased in steps of 10–20 mgs/day every week if required, dose to be taken in the morning, discontinue if the response is insufficient after 1 month; maximum dose 70 mgs per day.

Adult: Initially 30 mgs daily, increased in steps of 20 mgs/day every week if required, dose to be taken in the morning, discontinue if the response is insufficient after 1 month; maximum dose 70 mgs per day.

Common side effects include upper abdominal pain, anxiety, decreased appetite, behavioural problems, constipation or diarrhoea, dizziness, dry mouth, fatigue, feeling jittery, headache, sweating, poor sleep, mood alteration, movement disorders, nausea, palpitations, sexual dysfunction in adults, tachycardia, tremor, and weight loss.

Monitoring requirements are similar to methylphenidate and dexamfetamine and avoid abrupt withdrawal.

Lisdexamfetamine dimesylate is available as 20, 30, 40, 50, 60, and 70 mgs capsules (BNF, 76; ► www.bnf.org).

17.12.2 Non-stimulant Medication

17.12.2.1 Atomoxetine

Atomoxetine is an ADHD medication that acts as selective noradrenaline (norepinephrine) reuptake inhibitor. It is given once or twice a day and can take several weeks to reach plasma levels to lead to symptom improvement. Therefore, this contrasts with psychostimulant medications which can be taken or omitted when needed. Atomoxetine needs to be taken every day to get the maximum benefit. It has 24-hour symptom control which some patients prefer. In people with ID and ADHD, atomoxetine can also be considered as the first choice of medication especially if ADHD is associated with challenging behaviour throughout the day.

There are a limited number of studies on the use of atomoxetine in people with ID and ADHD. Current studies suggest a response rate of 50–65% for atomoxetine [100, 101]. In clinical samples, atomoxetine was reported to be one of the most commonly used ADHD medications in adults with ID [15]. Only two RCTs show the efficacy of atomoxetine in children with ASD and ADHD, a combination of parent training and atomoxetine being the most effective [96]. Atomoxetine was effective in reducing ADHD symptoms in children with ASD with/without ID in recent RCTs [102, 103]. Somnolence, fatigue, anorexia, nausea, and irritability were common adverse events.

Children 6–17 years and adults, weight up to 70 kg: Initially, 0.5 mg per kg daily for 7 days, the dose is increased by the response to 1.2 mg/kg/day weekly, can be given in two divided doses, last dose no later than early evening; up to a maximum of 1.8 mg/kg/day; maximum dose of 120 mgs daily.

Children 6–17 years and adults, weight above 70 kgs: start 40 mgs per day for 7 days, dose increased according to response, maintenance dose 80 mgs daily (adults can go up to 100 mgs/day), can be given in two divided doses, second dose early evening, maximum 120 mgs daily.

Doses above 100 mgs/day are not licenced in children, while those above 120 mgs/day are not licenced in adults.

Common side effects include abdominal pain, poor appetite, low mood, dizziness, feeling sleepy, anxiety, irritability, dry mouth, nausea, dyspepsia, flatulence, fatigue, sleepiness, palpitation, sweating, tachycardia, high blood pressure, tremor, vomiting, sexual dysfunction, rash, taste disturbance, dizziness, and urinary dysfunction.

Preparations: Oral solution: Strattera 4 mg/ml. Capsules: Strattera 10, 18, 25, 40, 60, 80, 100 mgs (BNF, 76; ► www.bnf.org).

17.12.2.2 Guanfacine

Guanfacine is an alpha-2 agonist with similar action to clonidine but with a longer duration of action. It is given once a day. A common side effect specifically related to guanfacine is sleepiness; as a result, it is administered at night. It may be helpful for patients with ADHD struggling with sleep. A small crossover trial of guanfacine in children with developmental disabilities showed benefits in improving hyperactivity and global functioning [104]. A recent large RCT in children with ASD (63% were with IQ < 70) showed that extended release of guanfacine was effective

in reducing hyperactivity/impulsivity and inattention with a large effect size (1.67) [105]. Drowsiness, fatigue, emotional fragility, and irritability were common adverse events.

The *main side effects* include nausea, gastrointestinal disturbances, negative impact on sleep, poor appetite, increased blood pressure, increased heart rate, and weight loss.

As many people with ID are highly sensitive to medications, the advice is to start medication at a lower dose and increase the dose slowly. Individuals with more severe ID may be unable to complain about their side effects; therefore, it is important to observe them closely to detect any side effects.

Monitoring of blood pressure and heart rate in adults due to the risk of increased blood pressure and heart rate is a requirement as per NICE [86] guidelines. Guanfacine can reduce blood pressure due to its mechanism of action. Weight monitoring is also required with height monitoring mainly for children. Blood pressure and heart rate monitoring in particular can be difficult in people with ID who are also challenging in their behaviour. A more balanced and pragmatic view needs to be taken and act in the best interests of the person with ID and ADHD in terms of starting and continuing ADHD medication in such individuals who lack the capacity to make treatment choices. Anecdotal evidence suggests that people with ID are more likely to engage in physical monitoring if their ADHD symptoms are better controlled with ADHD medications.

Increased anxiety is a known side effect of ADHD medications. This can be an issue in people with ID who are experiencing underlying anxiety symptoms especially if they have comorbid ASD. Educating families to monitor for symptoms of increased anxiety is important when starting ADHD medications. If there is a high risk of a further increase in anxiety symptoms, it is worth considering treatment for anxiety before or along with ADHD medications.

A small crossover trial showed clonidine was effective in reducing both inattentive and hyperactive/impulsive ADHD symptoms in children with ID [106]. Drowsiness, dry mouth, and anorexia were common adverse events. One small crossover trial has shown a better efficacy of risperidone than methylphenidate in improving ADHD symptoms in ID [107].

17.12.3 General Principles of Prescribing for People with ID

Starting on a smaller dose and gradually increasing the dose is the general recommendation for psychotropic prescribing in people with ID [108, 109] (see ▶ Chap. 11). It can help patients to tolerate side effects better and improve compliance. This may not be possible especially with slow-release ADHD medications as they cannot be broken into smaller doses than the recommended starting dose; however, non-stimulant medications such as atomoxetine can be started in smaller doses than the recommended BNF (BNF 67) starting dose. Some people with ID may find swallowing tablets or capsules difficult. In such situations, certain ADHD medications which come in liquid preparations can be used. The other strategy is to open capsules of modified ADHD medications and mix the granules with a food item. The medicinal leaflet needs to be followed in such instances. Atomoxetine is the only ADHD medication available in liquid form in the UK at present. Medikinet XL and lisdexamfetamine capsules can be opened and poured onto a food item.

17.13 Non-pharmacological Treatment

Non-pharmacological treatment options cover a wide range of interventions. Lack of studies in non-pharmacological treatment in people with ID can make treatment recommendations difficult for this patient group. However, management options used in the non-ID population can be adapted in people with ID on the premise that treatment principles are similar across both ID and non-ID groups for ADHD.

Despite challenges in building up a stringent evidence base, there is a wide range of

non-pharmacological management options available for ADHD treatment. This has led to difficulties in studying, analysing, and drawing conclusions on the effectiveness of individual strategies. However, on the contrary, a wide range of non-pharmacological interventions can be helpful for people who prefer non-pharmacological strategies. The other challenge for non-pharmacological treatment options is that potential adverse events are not recorded as part of the studies compared with detailed discussions of adverse events associated with pharmacological strategies.

Non-pharmacological options can be broadly divided into

1. Supporting an individual to manage symptoms of ADHD.
2. Reducing functional impairment secondary to ADHD.
3. Managing mental health conditions and emotional disorders secondary to ADHD.
4. Supporting families of children who have ADHD through psychoeducational programmes.

Some of the strategies discussed in the literature are cognitive behavioural therapy (CBT), dialectical behaviour therapy (DBT), coaching, mentoring and other counselling approaches, parent/family/carer training, psychoeducation, relaxation techniques, organisational skills/school or workplace targeted interventions, interventions to improve sleep, exercise, outdoor activities, non-specific supportive therapy, or combination of the above interventions. The use of one or more of these strategies is based on an individual's level of ID, support available to the person, availability of resources, and the environment the person lives in. For example, a person with mild ID and ADHD may be able to engage in CBT therapy adapted to their needs. Such individuals can also be supported through targeted interventions if in employment. Exercise and outdoor activities can be easily accessible depending on where the person lives, resources available, and the amount of support available to the individual especially if the person has moderate to severe ID. Relaxation strategies can be simplified and broken down into smaller steps to help people with any severity of ID.

There are two modes of treatment, namely pharmacological and non-pharmacological. ADHD medications available for children and adults without ID can be used in the ID population.

Psychostimulants are effective in treating ADHD symptoms in people with ID but relatively less effective than the general population.

The side effect profile is similar to that in the general population.

Non-pharmacological strategies can also be useful. There are many different non-pharmacological strategies. Interventions need to be tailored to the individual need [46].

Tip

Researchers and clinicians should pay more attention to the challenges in diagnosing and treating ADHD in persons with ID and/or ASD, with a special focus on person-centred outcomes and specific symptoms in individuals with more severe cognitive and communicative impairment.

Key Points

- There is growing evidence that people with ID are at a higher risk of comorbid ADHD compared with people without ID.
- Despite this much higher prevalence, there is minimal high-quality research specific to people with ID and low-functioning ASD in comparison to the general population.
- In people with ID and/or ASD, ADHD is frequently misdiagnosed, under-recognised, and improperly treated.
- The stringent use of categorical diagnostic criteria might limit the identification of the disorder since some criteria may not be applicable, particularly for those with more severe cognitive and communication impairment.
- Observation of a person's distractibility that is not consistent with their develop-

mental level may help when considering the symptom of inattention in an individual with ID and/or low-functioning ASD.

- In people with ID/ASD and more severe cognitive and communication impairment, hyperactivity and impulsivity can present with fidgeting most of the time, appearing 'on the go', and being unable to remain seated for a long time and wait for one's turn. Verbal or physical aggression, irritability, mood fluctuations, or self-harming behaviour are also possible expressions or consequences of these ADHD symptoms.
- ADHD appears to have a multifactorial aetiology with most of the risk factors discovered thus far resulting to be shared with many other psychiatric and neurodevelopmental disorders. More specific risk factors have been identified in few dopaminergic genes, catechol-O-methyltransferase gene, and brain-specific angiogenesis inhibitor 1-associated protein 2 gene. A well-recognised condition associated with ADHD is foetal alcohol spectrum disorder.
- ADHD therapy should be thorough, collaborative, and multimodal, with a variety of pharmacological and non-pharmacological options available to fit the individual requirements of the person with ID and/or ASD.

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Specific Learning Disorders, Motor Disorders, and Communication Disorders

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Contents

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Learning Objectives

- To learn about definitions and diagnostic criteria of specific learning disorders within the main classification systems
- To understand difficulties of diagnosing a specific learning disorder in people with ID and ASD
- To learn about most appropriate assessment for every specific learning disorder
- To learn about definitions and diagnostic criteria of motor and communication disorders
- To understand specific characteristics of motor and communication disorders in persons with ID
- To gain a better understanding of the current state of research and its implications for the development of treatment and support methods for specific learning, motor, and communication disorders

18.1 Introduction

Developmental learning, communication, and motor disorders affect persons with intellectual disability (ID), autism spectrum disorders (ASD), and/or other neurodevelopmental disorders more often than neurotypical persons. These disorders, which occur during the first years of life, have a significant influence on the emotional and adaptive development of children and adolescents, but they also have considerable consequences throughout the whole lifespan of affected persons, influencing their lives and personalities. Some of these disorders can be effectively remediated if recognized and treated early. Others are chronic and necessitate the development of compensatory skills.

Specific learning disorders (SLDs) are conditions that cause a discrepancy between potential and actual levels of academic performance as predicted by the person's intellectual abilities. Learning disorders involve impairments or difficulties in concentration or attention, language development, or visual and aural information processing. These specific learning disorders should be distinguished from a global cognitive impairment as found in people with ID although they can coexist with ID.

Motor disorders are lifelong conditions that make it hard to learn motor skills and coordination and may cause a lack of intended movement or an excess of involuntary movement. Symptoms of motor disorders include tremors, jerks, twitches, spasms, contractions, or gait problems.

Communication disorders consist of an impairment in the ability to receive, send, process, and comprehend concepts or verbal, nonverbal, and graphic symbol systems. A communication disorder may be evident in the processes of hearing, language, or speech, or any combination of these, and may range in severity from mild to profound. Developmental communication disorders differ from acquired ones in that they express conditions of uneven development of communication skills, while acquired communication disorders come after development and are generally the result of traumatic brain injury or neurological disorders.

The phenomenology, classification, aetiology, and treatment outcomes of communication and motor problems in people with ID and/or ASD are still poorly understood. It is also difficult to sort out the numerous causal relationships between these disorders and the often co-occurring psychiatric disorders. The co-occurrence of specific learning, motor, and communication disorders cause significant adjunctive impairment and result in negative physical, psychological, and social consequences.

18.2 Specific Learning Disorders (Specific Developmental Disorders of Scholastic Skills)

Specific learning disorders (SLD) or developmental learning disorder (in ICD-11) [1] represents a group of disorders characterized by specific deficits in one domain of cognitive processing that manifest in persistent learning difficulties with significant interference with academic or occupational performance or with activities of daily life.

They are chronic disorders that modify their expression over time in relation to age and environmental requirements, manifesting with different characteristics over the course of development and the phases of scholastic learning. The term 'specific' refers to the fact that the impairment refers to specific cognitive domains and is not solely explained by ID (intellectual developmental disorder; IDD) or impairments of overall cognitive functioning. If an ID is present, a specific learning disorder can be adjunctively diagnosed only if the specific learning difficulties are more severe than those attributable to ID [2]. Specificity has to be ascertained also in presence of global developmental delay, hearing or visual disorders, neurological or motor disorders, lack of availability of education, lack of proficiency in the language of academic instruction, psychosocial adversity, etc.

SLDs occur early during the first years of formal schooling, with difficulty in learning, reading, writing, and mathematics. They are persistent, consequently also resistant to targeted educational interventions. These disorders, which do not depend on maturation processes, emerge when reading, writing, and mathematics are taught at school in an explicit manner, but could also manifest later, when the person can no longer compensate for the environmental requirements, for example due to the speed required or excessive academic demands. They are not due to lack of educational opportunity, ID, trauma or acquired brain-related diseases, or uncorrected visual or hearing impairment.

The neurobiological dysfunction underpinning the disorders interferes with the normal learning process of reading, writing, and mathematics and is influenced by environmental factors such as school, family, and social context, determining the phenotypic expression and a greater or lesser maladjustment [3].

Thus, it involves a neurobiological impairment of academic skills like reading, writing, and mathematics, which are considered as functional disabilities. These functional aspects weigh heavily on the organization of the day of students with SLD diagnoses, a day mainly occupied with schoolwork during school hours, but also on the time used in the afternoon or weekend for completing the schoolwork, limiting the time to dedicate to more recreational activities or sports, believed by many parents of SLD students to be a type of release valve necessary for maintaining their psychological health.

SLDs are typically diagnosed in childhood, but they often continue into adulthood influencing a wide range of occupational, relational, and mental health issues.

Compared with the previous edition of the DSM [4] and the ICD-10 [5], DSM-5 [2] sets out a single diagnostic category for SLDs. While in ICD-10 [5] we included specific disorder of reading (DD, developmental dyslexia, F81.0), the disorder of spelling (dysorthographia, F81.1), and specific disorder in mathematical abilities (dyscalculia, F81.2), in DSM-5 [2], the continuity of the disorders is highlighted, included in the single category of SLDs, but with the specification of different impairments such as reading impairments (included dyslexia), mathematical impairment (included dyscalculia), and impairment of written expression (included dysorthographia and dysgraphia). The ICD-11 classification differs significantly from the ICD-10 and is very much aligned with the DSM-5, dividing developmental learning disorder (6A03) into (0) with impairment in reading; (1) with impairment in written expression; (2) with impairment in mathematics; (3) with other specified impairment of learning; and (Z) unspecified (see Table 18.1).

According to the DSM-5, in people with ID the formulation of an adjunctive diagnosis of a specific learning disorder is possible only if the specific learning difficulties are more severe than those attributable to ID.

18.2.1 Prevalence

SLDs are one of the most common neurodevelopmental disorders (NDD), the prevalence of which is estimated as 5–15% in school-aged children across different languages and cultures, and approximately 4% in adults. These percentages vary from country to

Table 18.1 SLDs in ICD-10 [5], ICD-11 [1], and DSM-5 [2]

SLD	ICD-10	ICD-11	DSM-5
Dyslexia	F81.0 Specific reading disorder	6A03.0 Developmental learning disorder with impairment in reading	315.00 Specific learning disorder with impairment in reading
Dysorthographia	F81.1 Specific spelling disorder	6A03.1 Developmental learning disorder with impairment in written expression	315.2 Specific learning disorder with impairment in written expression
Dysgraphia	F81.8 Other developmental disorders of scholastic skills	6A03.3 Developmental learning disorder with other specified impairment of learning	315.2 Specific learning disorder with impairment in written expression
Dyscalculia	F81.2 Specific disorder of arithmetical skills	6A03.2 Developmental learning disorder with impairment in mathematics	315.1 impairment in mathematics

country, as they depend on the characteristics of the language studied (transparent language vs. opaque language and more or less regular spelling), on the varying definitions of SLD, and on the different methods and instruments used for diagnosis. The cut-off scores reported in the literature for dyscalculia diagnosis are also very variable, ranging from the 5th percentile [6] to the 46th, [7] so the reported prevalence is not uniform. The DSM-5 [2] reports the rate of dyslexia among 4–9%, while dyscalculia 3–7% [2].

There is a slight dominance among males compared with females (ratios range from about 2:1 to 3:1) [2, 8] probably because more males than females come to clinical attention for their behavioural manifestations, in particular, for the frequent comorbidity between SLD and attention deficit hyperactivity disorder (ADHD), a condition which itself is associated with a high rate of problem behaviour [9].

SLDs seem to be rather frequent also in children, adolescents, and adults with borderline intellectual functioning and mild ID, though an adjunctive diagnosis of a specific learning disorder requires special care and in-depth evaluation in these individuals [10]. In general, SLDs' prevalence is inversely correlated with IQ level, even when other factors such as age, gender, and socio-cultural context are taken into account [11].

Specific learning disorders occur early during the first years of formal schooling, with difficulty in learning, reading, writing, and arithmetic.

These disorders originate in childhood and often continue into adulthood among many.

These disorders seem to be prevalent among children and adults with borderline intellectual functioning and mild intellectual disabilities, although in these persons adjunctive diagnosis of a specific learning disorder requires particular attention and in-depth assessment [10].

18.2.2 Aetiopathogenesis

The identification of the causes of SLD is complicated by their heterogeneity and the influence of multiple neuropsychological factors [12, 13]. Nevertheless, most experts agree on the biological origin of the cognitive anomalies that underlie behavioural symptoms of the disorders. The biological origin includes the interaction of genetic, epigenetic, and environmental factors that affect the brain's ability to perceive or process verbal or nonverbal information efficiently and accurately [2]. For example, left temporo-parieto-occipital impairment is associated with dyslexia.

Particularly, functional neuroimaging studies identified hypoactivation of the left parieto-temporal and occipito-temporal regions in children and adults with SLDs when they perform reading-related tasks [14–20].

Although genetic abnormalities associated with SLDs are described in the literature, the 'dyslexia gene' has not yet been identified. SLDs seem to have complex multi-factorial aetiology [21] because it is believed that the transmission of the disorder does not occur according to the Mendelian inheritance model. Genes such as DYX1C1, DCDV2, KIAA0319, ROB01, C2Orf3, and MRPL19 are implicated as risk factors for dyslexia [22, 23]. The SLDs, like the other NDDs, have overlapping traits, which derive from the interaction among multiple genetic risk factors and the epigenetic regulation by environmental factors [24].

Neural plasticity is also affected by the quality of the external environment, both in terms of life experiences and biological factors, such as diet, drugs, endocrinal abnormalities, or pathogenic agents [25]. Risk factors for SLDs are divided into environmental, genetic, and physiological factors. Environmental factors include premature birth or very low birth weight, and prenatal exposure to nicotine. A family history of SLDs is frequently present in persons with SLDs. In fact, recent studies have revealed that the whole spectrum of SLDs is strongly determined by genetic predisposition with the highest risk among first-degree relatives [26, 27].

18.2.3 Criteria and Clinical Features

The diagnosis of an SLD in reading, writing, and arithmetic can be made based upon individually applied standardized methods for testing scholastic achievement and IQ. The evaluation is based on a variety of methods, including medical history, clinical interview, school report, teacher evaluation, rating scales, and psychometric tests. For individuals aged 17 years and older, standardized achievement measures and comprehensive clinical assessment may be substituted by a documented history of impairing learning difficulties.

In DSM-5 [2] the IQ discrepancy criterion was abandoned. To make a diagnosis of SLD, the impaired ability must be significantly lower than age and class attended (discrepancy criterion). In statistical terms, this corresponds to two standard deviations below the mean, or performance below the 5th percentile [5]. The results depend largely on the tests used, which vary according to country, but which must present adequate psychometric characteristics, with a good reference sample and valid and reliable norms.

The first diagnostic criterion of DSM-5 [2] (Criterion A) is that difficulties learning and using academic skills must be present for at least 6 months, despite targeted interventions. Indication of the number of months of persistence of the disorder and the resistance to the targeted intervention represent an important innovation compared with previous editions, consistent with the literature on response to intervention (RT) [28–30].

To make an SLD diagnosis, one of these symptoms must be present (a) slow or inaccurate word reading, (b) difficulty in reading comprehension, (c) difficulty in spelling (spelling errors), (d) difficulty in written expression (grammatical, punctuation, morphosyntactic errors), (e) difficulty in mastering the number sense, (f) arithmetical facts or calculation, and (g) difficulty with mathematical reasoning.

The second diagnostic criterion (Criterion B) relates to performance in the affected academic skill, which must be below those expected for the age. It is, therefore, necessary to verify a particularly low scholastic performance, for example in a person who requires specific skills, compared with others in which the student succeeds easily. The school achievements may also be adequate but obtained only with great effort and struggle and, therefore, are inconsistent with the time and effort dedicated to studying.

The learning difficulties cannot be due to atypical academic experiences, for example prolonged absences from school, frequent changes of school or teachers, inadequate teaching, or inadequate educational instruction. SLD

cannot be attributed to more general external factors, such as economic or environmental disadvantage, chronic absenteeism, or lack of education as typically provided in the individual's community context. Uncorrected visual or auditory acuity, other mental or motor or neurological disorders (e.g. paediatric stroke), psychosocial adversity that could cause academic difficulties must be excluded (Criterion D).

Academic skills and intelligence must be evaluated with individually administered standardized tests. In the case of borderline IQ (70–85), it is recommended to use a multicomponent test for intellectual assessment, since the cognitive profile is more informative than the mere IQ scores. The difficulties must interfere with academic or occupational performance and the activities of daily life.

The DSM-5 [2] describes three specific learning disorders, namely (a) impairment in reading, (b) impairment in the written expression, and (c) impairment in mathematics. Each disorder is described by subskills that must be recorded separately.

A disorder in reading (dyslexia) is characterized by (a) the inability to decode a text; (b) inaccurate or dysfluent loud reading, for example when words are read incorrectly or slowly and hesitantly, or when the child tries to guess words or has difficulty sounding out words (may add, omit, or substitute vowels or consonants); or (c) when there are difficulties in reading comprehension. Difficulties in understanding the relationships, inferences, or deeper meanings are included, even if reading is adequate. According to DSM-5 [2] and ICD-10 [5], the diagnosis of reading impairment can be made even if there is only impairment in comprehension of the text without difficulty in loud reading, but in some countries like Italy, the Consensus Conference promoted by the Italian National Institute of Health [3] recommend to diagnose dyslexia, not to include reading comprehension as a diagnostic parameter, since persons with comprehension problems but good decoding skills do not meet the criteria for dyslexia. For transparent languages such as Italian, for the diagnosis of dyslexia, it is recommended to consider reading fluency in addition to the parameter of accuracy as reported in the ICD-10 [5] criterion [31].

In adults, most common symptoms of dyslexia include visual problems while reading (high sensitivity to glare, the colour of the paper, font, etc.), difficulty focusing when reading such as losing place, confusing very similar words or letters when writing or reading, feeling like words are moving or jumbled up, finding reading very stressful, rarely or never reading for pleasure, difficulties with written communication or tests, difficulty writing down messages or reports, and confusing left and right, or struggling with spatial reasoning [32, 33].

Another disorder included in SLDs is dysorthography, characterized by impairment of orthographic skills and phonographic coding. In the DSM-5 [2] the impairment in written expression includes also errors in spelling, grammar, and punctuation, or poverty in organization and clarity of written expression.

There may be a disorder in numeracy and calculation skills (dyscalculia) when there are difficulties in mastering number sense, number facts, or calculation. The calculation is slow or incorrect, understanding of numbers is poor, arithmetic facts are not memorized, fingers are used to count instead of recalling the fact, and procedures are not automated. Difficulties in mathematical reasoning can be present, with difficulty applying mathematical concepts, facts, or procedures to solve quantitative problems. Skills assessed with standardized tests must fall outside the limits of 2 standard deviations (SD) from the level expected based on the child's chronological age and his overall intellectual level, but no parameter (fluency, accuracy) is specified.

This lack of precise references, which includes calculation difficulties that are rather different from those characterizing a real disorder, is probably the cause of the extreme variability of the way dyscalculia is defined in scientific literature. Dyscalculia seems to fall at the highest severity end of a wide spectrum of mathematical difficulties. In addition, many studies confound exact and approximate numerical processing because they involve qualitatively different task designs [34].

It is difficult to establish a reliable prevalence rate for the impaired mathematical skills because of the heterogeneity of definitions, diagnostic criteria, and different assessment scales used across the studies [35–37], with a considerable increase when the discrepancy between intellectual efficiency and mathematical skill is not adequately considered and persons with ID are included [38]. Kaufmann [39] argues that in many studies on mathematical difficulties only a few participants show a real calculation disorder [35, 40]. Also, aetiopathological models of dyscalculia are contradictory. According to both the 'core deficit theory' [41] and the 'defective module hypothesis' [42], individuals with dyscalculia would suffer from impaired number processing due to a defective number sense, while the 'triple code model' argues for alterations in one or more of the three representational codes for number, which are Arabic digits, verbal number words, and analog non-symbolic magnitude representations, each subserved by functionally dissociated neural substrates [43]. A meta-analysis suggested that specific difficulty in number facts [44] or number processing [45] is particularly meaningful as a criterion.

- **DSM-5 [2] Describes Three Levels of Severity for SLDs, Namely Mild, Moderate, and Severe**

1. Mild: Some difficulties in learning skills in one or two academic domains, but of mild enough severity that the individual may be able to compensate or function well when provided with appropriate accommodations or support services, especially during the school years.
2. Moderate: Marked difficulties in learning skills in one or more academic domains, so that the individual is unlikely to become proficient without some intervention of intensive and specialized teaching during the school years. Some accommodations or supportive services at least part of the day at school, in the workplace, or at home may be needed to complete activities accurately and efficiently.
3. Severe: Severe difficulties in learning skills, affecting several academic domains, so that the individual is unlikely to learn those skills without ongoing intensive individualized and specialized teaching for most of the school years. Even with an array of appropriate accommodations or services at home, at school, or in the workplace, the individual may not be able to complete all activities efficiently [2].

The timeliness and adequacy of the rehabilitative measures, the IQ score (which in SLD may be at the mild ID end, but also above average), the areas involved in the disorder (one or more among reading, calculation, reading comprehension, expressive ability, graphic skill, spelling), and any contemporary presence of other disorders (most frequently, language and attention disorders) influence the severity of the disorder. The severity of impairment may vary from case to case and affect the academic skills by age in a different way.

There is high comorbidity among different types of learning disabilities (dyslexia, dysorthographia, dysgraphia, and dyscalculia), and they often co-occur with other NDDs, ADHD, communication disorders, developmental coordination disorder, ASD, and other mental disorders (anxiety disorders, depressive and bipolar disorders). In children with SLDs in multiple learning domains, both the rates and the types of psychopathology are higher than in children with an isolated SLD [46].

Dyslexia has particular comorbidity with ADHD, developmental dyscalculia, specific language impairments, and speech-sound disorders [47]. Overlap among various learning disorders leads to sharing of some risk factors among them, different according to the individual disorders involved [48]. Therefore, it becomes particularly difficult to clearly distinguish main symptoms from associated symptoms [49].

18.2.4 Specific Assessment

The assessment involves professionals with expertise in SLDs and psychological/cognitive assessment.

According to DSM-5 [2], low scores are at least one and a half standard deviations below the mean of the population by age, which is

below the 7th percentile, in one or more tests. In ICD-10 [5], this threshold is two standard deviations below the mean, or the 5th percentile.

To verify the reading impairment, it is necessary to use evidence that evaluates the word recognition, the ability to read out loud, and the ability to automatically understand what is read. Reading tests include a word, non-word, sentence, and passage reading. In childhood word and non-word reading tests show higher reliability and predictability as compared with whole text reading. In dyslexic and/or compensated adults use of non-word reading tests is recommended for its relevance in this particular population. Given that decoding may be slow or incorrect, it is necessary to measure both the parameters of speed and correctness. For a correct functional diagnosis, it is also necessary to make a qualitative analysis of the errors committed, to correctly attribute them to phonological, articulation, or lexical difficulties.

It is also important to verify expressive and receptive language skills, given that frequently, reading impairments are preceded by a history of speech and language disorders, which, during school age, may be compensated or still show some specific markers. To evaluate reading comprehension, it is necessary to use evidence that assumes autonomous reading by the person, usually including passages with questions and multiple-choice replies that the student must respond to. To distinguish how much a decoding difficulty affects fatigue, and thereby the correct comprehension of what was read, it is also useful to administer comprehension tests in listening form, read by the examiner.

To evaluate the impairment of writing skills, it is necessary to include dictations of words, sentences, or passages, checking that the spelling is correct. For the diagnosis of dysorthographia, the use of word and non-word dictation tests is recommended along with the production of written texts and sentences. In early school years, it is important to assess grapheme-phoneme conversion processes, while during primary school, assessment of spelling at the lexical level is becoming progressively more important. Errors in grapheme-phoneme conversion at the end of the primary school or later is a marker of a particularly severe disorder. To evaluate the correctness of the grammar and punctuation and the morphosyntactic aspects, it is also necessary to test spontaneous writing skill, which involves regulations for the number of words and sentences used, the expository richness (use of qualifying adjectives, personal pronouns, number of subordinations, adequate vocabulary, etc.), punctuation correctness.

To evaluate the calculation impairment, arithmetic tests must be used, that involve subtests on numerical ability, arithmetic facts, calculation, and mathematical problem-solving. These tests evaluate the concept of numbers, counting, reading and writing of numbers, repetition of numbers, semantic coding, ordering of numbers, and size comparison. With regard to calculation, it is necessary to involve written calculation tests, to evaluate the learning of the algorithm procedure, mental calculation with complex numbers to evaluate the ability for calculation strategies, mental calculations with small quantities to verify the learning of the so-called arithmetic facts, namely those calculations that, for their solution, do not require the application of the algorithm (e.g. sums and subtractions under ten), learning of multiplication tables. Murphy [35], Mazzocco [45], and Chong [50] suggest defining 'dyscalculia' only when the child's test performance level is lower than the 10th percentile in at least two specific tests of basic arithmetic skills. Children who score between 11th and 25th percentiles are considered 'low achievers', while those above the 25th percentile are considered 'typical achievers'. In these tests, it is necessary to evaluate the correctness, the number and type of errors made [7, 35, 51], and the speed of execution that, with age, becomes a significant parameter to highlight the presence of dyscalculia. It is also crucial to evaluate the persistence of immature computation strategies (e.g. long-term use of the fingers) [52].

As working memory and visuo-spatial skills support and facilitate the acquisition and consolidation of arithmetic skills [53], for the diagnosis of dyscalculia they must be investigated with standardized tests assessing [54] different types of dyscalculia. For example, different treatment programmes may be needed depending on the type of numerical cognition or procedural deficits.

Neither ICD-10 [5] nor DSM-5 [2] explicitly involves the diagnostic label of dysgraphia, that is difficulty in writing in the absence of a motor coordination disorder. To verify the presence of dysgraphia, it is, therefore, necessary to exclude this disorder and involve tests that evaluate both the quality of the written product and the speed of execution. For the ICD-10 [5], it is possible to use diagnostic code F81.8 'Other development disorders of scholastic skills'.

It is indisputable that to exclude the presence of an IDD, it is necessary to evaluate the intelligence through a standardized test that may be mono-component or multicomponent. There is a recent debate on how necessary it is to make a complete cognitive evaluation to diagnose a specific learning disorder [55].

Diagnosis of SLD includes cognitive, speech and language, medical, psychological, and educational assessment, which should be made by professionals with specific training and expertise.

18.2.5 Treatment

Early intervention is fundamental for all SLDs in order to prevent or limit maladaptive consequences in school and working career [56, 57] as well as psychological distress and risk of mental health issues [58, 59]. The effectiveness of rehabilitation programmes on reading, writing, calculation, and overall adaptive skills during the developmental age has been repeatedly proven, but evidence on long-term outcomes is lacking.

Numerous studies on the aetiopathogenesis of dyslexia in recent years have helped to identify a multifunctional deficit model [60, 61], but studies on treatment have been few [62], due to several factors such as the national health policies, considerable heterogeneity of interventions, and cost. Rehabilitation programmes vary from country to country, focusing more on correctness or speed depending on the characteristics of the language. In less regular spellings (as in English) correctness represents the main issue, while in regular spellings (as in the Italian language) it is the speed that has to be impaired more [63–65]. In general, studies on the enhancement of correctness are more numerous than those on speed [66–67]. The literature suggests that the skills to train in the presence of dyslexia are verbal working memory, meta-phonological skills (phonological processing), and rapid automatized naming (RAN). Proposed treatments are different according to the aetiopathogenetic theories (phonological awareness theory, rapid auditory processing theory, magnocellular-dorsal theory, attentional deficit theory) embraced by those who developed them.

Systematic reviews of the literature, with and without meta-analysis, show that specialist interventions aimed at improving the correctness and fluency of reading are effective. These include the reading of words isolated or inserted in a context as well as exercises on meta-phonology, grapheme-phoneme conversion, repeated readings, which must be done several times in a week for at least 20 min each, and for at least 15–20 sessions [68–74]. In transparent languages, such as Italian, fast reading of whole words or reading with facilitated syllables identification are effective, also through the use of modern technologies, which allow to realize remote rehabilitation programmes, reduce costs, and are more fun for kids [66, 67, 75–81]. Many studies show that increased letter spacing can facilitate reading in dyslexics with visual crowding deficits [82–90]. In recent years transcranial magnetic stimulation (TMS), particularly rapid-rate TMS has been successfully used for treating dyslexia and for improving reading performance by exciting or rebalancing underactive reading pathways in the brain [91–93]. Also, transcranial direct current stimulation is showing promising results [94].

Studies on the treatment of dysorthography (or spelling disorder) are even less than those for dyslexia, probably because dyslexia and dysorthography frequently co-occur and some prerequisite skills underlie both conditions [95–97]. However, this does not justify the lack of studies, given that spelling correctness also requires skills other than the recognition process, such as the ability to segment words into phonemes [98]. In fact, students who have spelling deficits do not necessarily have word recognition deficits [99].

Interventions on dysorthography must involve the various sub-lexical or lexical components or both [100]. Studies carried out on English- or German-speaking children have shown the effectiveness of interventions on both components, sub-lexical [101, 102] and lexical [103–105]. The effectiveness of treatments depends on many factors; the literature indicates that the most important are represented by the clarity of instructions, regularity of exercise repetition, and timeliness of feedbacks [106].

Computer-based programs seem to offer benefits beyond traditional interventions, especially for spelling in writing through word processing programs with speech synthesis, although the evidence is far from conclusive [107, 108].

Treatments for dyscalculia also vary depending on pathogenic models. The most common focus on the Approximate Number System [41] and the access deficit hypothesis [109].

Slowness with numerical magnitude processing is a crucial target for treatment, for both symbolic (digits) and non-symbolic (dots) number formats, although data are more consistent and robust across studies for symbolic numbers [110–112].

In addition to the duration (at least 20 sessions of 30 min each), other characteristics of the intervention proved highly relevant for effectiveness, such as the inclusion of direct and self-instruction (more than mediated instruction), use of adequate support tools (i.e. number line), and teacher experience. Computer-assisted interventions can improve motivation to practice, automatization of math facts, and direct feedback provision but showed smaller effects than traditional interventions with humans as teachers [113–117].

Early intervention is critical for all SLDs to avoid or reduce negative repercussions in academic and occupational career, as well as psychological distress and the likelihood of mental health problems.

Various treatments and support programmes are available for SLDs, showing a varied rate of success.

18.2.6 Prognosis

SLDs occur early during the first years of formal schooling and persist into adulthood. As for NDDs, their manifestation may change over time, determined in part by genetic and biological factors, and influenced by environmental aspects. Therefore, the progression depends both on the severity of the impairment and the general clinical situation and on the external stimuli that the student receives from school and family. The IQ score also has an influence, which, as a diagnostic criterion, must not be lower than 70, but that delineates very different profiles of function between students that have a medium-low IQ and those with a very high IQ. Early diagnosis and successive intervention are fundamental, differentiating the prognosis of children who are diagnosed in adolescence compared with those who receive early diagnoses. Subjects who have undergone a course of rehabilitation treatment may compensate for the initial learning difficulties, while some are resistant to treatment and continue, therefore, to have significant problems later in life [118, 28, 30].

In addition, comorbidities have to be considered as the sum of numerous disorders makes the situation more complex and does not favour a positive prognosis. In particular, the coexistence of SLD and ADHD is predictive of worsening results of mental health. The situation may be worsened by histories of scholastic failure, with early school leaving (dropout) and a greater probability of psychological disadvantage. Early school leaving and symptoms of depression lead to a higher suicide rate, while social and emotional support are predictive factors.

Tip

Researchers and clinicians should focus more on identifying and treating specific learning disorders in adults who have not previously received treatment.

The neurological, genetic, and cognitive basis of learning impairments should be addressed in future studies and prac-

tice, particularly in those who do not respond to typical treatments. A continued empirical study on the function of cognitive processes in detection and intervention is required. Interdisciplinary study based on a rigorous, scientific approach and focused on integrating information across domains will provide a better understanding of the communalities and distinctions between specific and non-specific developmental learning problems.

18.3 Motor Disorders

Developmental motor and coordination disorder represent a group of lifelong conditions characterized by difficulties in acquiring motor and coordination skills. They are not learning disorder, but they can impact learning and many associated activities. Children with these conditions may be substantially delayed in reaching motor milestones, make repetitive and involuntary movements, or have physical or verbal tics, which cause impairment and result in negative physical, psychological, and social consequences across the whole lifespan. Motor disorders frequently co-occur with intellectual disability and/or autism spectrum disorders, supporting the notion that motor, cognitive, and social-communication functioning are interrelated [119, 120].

The latest editions of the main classification systems of disorders and diseases (DSM-5 [2] and ICD-11 [1]) show significant changes in the consideration of developmental motor disorders compared with previous editions. The DSM-5 [2] created a new subcategory called 'Motor Disorders' under the new meta-structure called 'Neurodevelopmental Disorders', while in the DSM IV-TR [4], developmental coordination disorder, stereotypic movement disorder, and tic disorders were included under the cluster 'Disorders Usually First Diagnosed in Infancy, Childhood, and Adolescence', with only developmental coordination disorder being specified as a motor skills disorder. This new neurodevelopmental framework also blurred the hierarchy of a disorder in relation to another; for example, stereotypies occurring in children who are otherwise developing normally, which were referred to as primary, are no longer distinguished from those occurring in children who have developmental problems, which were referred to as secondary. Another significant change concerns tic disorders, for which the DSM-IV-TR [4] required the youth to be tic-free for no more than 3 months before being diagnosed. This requirement has been removed in the DSM-5 [2], which takes a more dimensional approach to diagnosis and, as a result, places a greater emphasis on the likelihood that tics might come and go in terms of frequency and incidence. In the DSM-5 'motor disorders' category includes developmental coordination disorder, stereotypic movement disorder, and tic disorders [2].

Within ICD-11, only the developmental motor coordination disorder and stereotyped movement disorder are included under the new chapter of 'neurodevelopmental disorders' (number 6), while primary tics or tic disorders are included in the chapter of 'diseases of the nervous system' (number 8), and more specifically in the subchapter named 'movement disorders', which also includes disorders as parkinsonism, choreiform disorders, dystonic disorders, or myoclonic disorders [1].

Concerns about the presence of a developmental motor disorder should be raised if the following developmental milestones are missing or delayed; (a) 4–6 months: sits with support, rolls, reaches out and grasps objects; (b) 6–9 months: crawls, sits without support, pulls to stand, transfer objects between hands; (c) 7–12 months: pincer grasp, walks with hands held; (d) 12–15 months: drinks from a cup, builds two-brick towers; (e) 18 months: walks up steps, walks independently, builds three-brick towers; (f) 2 years: kicks and throws a ball, jumps, runs, builds six-brick towers, uses spoons, turns pages, helps with dressing, draws circular scribbles; (g) 3 years: stands on one leg momentarily, eats with forks and spoons, draws circles; (h) 4 years: hops, can dress and undress, draws a person with head, body, and legs [121].

Motor disorders such as motor coordination difficulties, tic, and Tourette's syndrome are common in children and adults with intellectual disabilities.

18.3.1 Developmental Coordination Disorder

- **DSM-5 Diagnostic Criteria [2]**

A. The acquisition and execution of coordinated motor skills are substantially below that expected given the individual's chronological age and opportunity for skill learning and use. Difficulties are manifested as clumsiness (e.g. dropping or bumping into objects) as well as slowness and inaccuracy of motor skills (e.g. catching an object, using scissors or cutlery, handwriting, riding a bike, or participating in sports).
B. The motor skills deficit in Criterion A significantly and persistently interferes with activities of daily living appropriate to chronological age (e.g. self-care and self-maintenance) and impacts academic/school productivity, prevocational and vocational activities, leisure, and play.
C. The onset of symptoms is in the developmental period.
D. The motor skills deficits are not better explained by intellectual disability or visual impairment and not attributable to a neurological condition affecting movement (e.g. cerebral palsy, muscular dystrophy, degenerative disorder).

The diagnosis is made by a comprehensive developmental and medical history, physical examination, school or workplace reports, and culturally appropriate standardized individual tests. Disorders that commonly occur with developmental coordination disorder include speech and language disorder, SLDs especially reading and writing, problems of attention including ADHD, ASD, disruptive and emotional problems, and joint hypermobility syndrome [2].

The diagnostic criteria exclude ID, but the two conditions appear together in several studies. Gillberg and colleagues [122] found that after adjusting for epilepsy, there remained a significant association between febrile seizures and ASD, developmental coordination disorder, and ID. Bernier and colleagues [123] were able to follow up two cohorts, one with 16 p11.2 deletion and the another with 16 p11.2 duplication, and study their early developmental trajectories and emergence of the phenotype. The most commonly diagnosed conditions for the deletion carriers were speech sound disorder (67%), developmental coordination disorder (67%), and language disorder (54%). For the duplication carriers, the most common diagnoses were developmental coordination disorder (56%) and ADHD (39%). Fifteen per cent of the deletion group and 22% of the duplication group also had an ID. Although the verbal IQ of duplication children with ID improved, they showed more motor skills problems over time ($P = 0.02$) and showed a trend toward increased challenges in daily living skills. There appeared to be a complex interplay of possibilities between the children developing ASD, motor disorders, or ID.

Barnevik Olsson and colleagues [124] presented the neuropsychiatric profiles of children aged 11 years with ASD, assessed before age 4.5 years, and after interventions. Developmental coordination disorder rates were equal in the average intelligence, borderline intelligence, and ID group. The authors make a very important point about ASD which is equally important about ID that it is rarely an isolated disorder but a co-occurring one.

Cunningham and colleagues [125] found that indicative developmental coordination disorder was associated with full-scale IQ children with 22q11.2DS ($P = 0.038$) which suggests that the observed coordination difficulties seen in this population can be partially explained by a general deficit in IQ. They suggest that this agrees with studies of children with developmental coordination disorder not selected for having a chromosomal disorder and suggest that within the ID population, the level of impairment is associated with motor dysfunction.

> Although the presence of ID is an exclusion criterion for the diagnosis of developmental coordination disorder, the two disorders have been found together in several studies.

18.3.2 Stereotypic Movement Disorder

According to ICD-11, stereotyped movement disorder is characterized by the presence of persistent voluntary, repetitive, apparently purposeless movements that are not caused by the direct physiological effects of a substance or medication, and markedly interferes with normal activities [1]. Like developmental coordination disorder, stereotypic movement disorder has an early onset during the developmental age but can continue across the whole lifespan [126]. Around 80% of children with complicated motor stereotypies show symptoms before the age of 24 months, 12% between the ages of 24 and 35 months, and 8% at the age of 36 months or beyond [127]. Stereotypic movement disorder affects more males than females [128].

Symptoms of stereotypic movement disorder include repetitive and involuntary motor behaviours like shaking, rocking, finger-flicking mannerisms, or hand flapping. Stereotyped movements with self-injury, such as head banging, face slapping, or self-biting, represent a specific sub-category of 'stereotyped movement disorder' within ICD-11 while they are considered as a specifier of a unique category of 'stereotypic movement disorder' in DSM-5. However, most of the international scientific community views self-injurious behaviour as a broad category of problem behaviours with a variety of aetiologies and explanatory hypotheses, including the possibility of underlying causative mechanisms that are more closely related to the spectrum of OCD/impulse dyscontrol than to stereotypic movement disorder [129]. For further details on self-injury see ► Chap. 7.

Youngsters with stereotypic movement disorder are unable to cease repeated motions even when distracted or given attention, although they can try to limit their movements by sitting on their hands or wrapping their arms in their clothing. Nevertheless, it is also possible that distraction may attenuate stereotypic movements to some extent and for a limited time.

In terms of body location, stereotypies frequently involve hands, arms, legs, the upper body, or the entire body, which represent quite different locations in respect to other repetitive movement disorders such as tic disorders, which commonly involve eyes, face, head, and shoulders. Stereotypies are also more stable, rhythmic, and long-lasting than tics, which tend to be variable, irregular, and quick. Also in contrast to tics, stereotypies are not accompanied by premonitory desires, prior sensations, or an inward desire to execute. Both could be precipitated or exacerbated by some psychological conditions such as distress, anxiety, excitement, focused concentration, or boredom, but stereotypic movements are also frequent when a person is immersed in an activity.

Stereotyped or repetitive motor movements can also be present in ASD and are not uncommon in persons with ID, especially in those with a moderate-to-severe degree of severity. In these cases, stereotypic movement disorder is diagnosed only when there is self-injury or when the stereotypic behaviours are sufficiently severe to constitute an adjunctive clinical focus. Further information on stereotypies associated with ID and ASD has been included in the ► Chaps. 6 and 16.

Despite the frequency of its occurrence, stereotypic movement disorder still presents diagnostic difficulties, confusion around comorbidities, and management uncertainty. Also, terminology, definition, and aetiology have been subject to recent debate [130].

18.3.3 Tic Disorders

A tic is a sudden, rapid, recurrent, non-rhythmic motor movement or vocalization. Tics are common in childhood and transient in most cases. The estimated prevalence of Tourette's syndrome ranges from 3 to 8 per 1000 school children with boys being more commonly affected than girls [2]. Onset of tics is typically between 4 and 6 years of age, peak severity occurs between 10 and 12 years, with a decline during adolescence. They can wax and wane and change muscle groups affected and patterns of vocalizations over time.

Tics can be simple or complex. Simple motor tics are of short duration and can include eye blinking, shoulder shrugging, and

extension of extremities. Simple vocal tics include throat clearing, sniffing, and grunting caused by the contraction of the diaphragm or the muscles of the oropharynx. Complex tics can appear purposeful. Complex tics are of longer duration and often include a combination of simple tics such as head turning and shoulder shrugging. They can appear purposeful, such as a tic like sexual gesture (copropraxia), tic like imitation of someone else's movements (echopraxia), tic like an imitation of what one has said (palilalia), and tic like an imitation of what someone else has said (echolalia), uttering socially unacceptable words (coprolalia). Coprolalia is an abrupt, sharp bark or grunt utterance and lacks the normal prosody of speech [2].

Tics are worsened by anxiety, stress, and exhaustion. Observing a gesture or sound in another person may precipitate tic which may be misconstrued as being purposeful by authority figures such as teachers.

The key DSM-5 [2] diagnostic criteria for different tic disorders are listed below.

Tourette's Disorder

A. Both multiple motor and one or more vocal tics have been present at some time during the illness, although not necessarily concurrently.
B. The tics may wax and wane in frequency but have persisted for more than 1 year since the first tic onset.
C. Onset is before age 18 years.
D. The disturbance is not attributable to physiological effects of a substance (e.g. cocaine) or another medical condition (Huntington's disease, post viral encephalitis).

Persistent (Chronic) Motor or Vocal Tic Disorder

A. Single or multiple motor or vocal tics have been present during the illness, but not both vocal and motor.
B. The tics may wax and wane in frequency but have persisted for more than 1 year since the first tic onset.
C. Onset is before age 18 years.
D. The disturbance is not attributable to physiological effects of a substance (e.g. cocaine) or another medical condition (Huntington's disease, post viral encephalitis).
E. Criteria have never been met for Tourette's disorder.

Specify if with motor tics only or with vocal tics only.

Provisional Tic Disorder

A. Single or multiple motor and/or vocal tics.
B. The tics have been present for less than 1 year since the first tic onset.
C. Onset is before the age of 18 years.
D. The disturbance is not attributable to physiological effects of a substance (e.g. cocaine) or another medical condition (Huntington's disease, post viral encephalitis).
E. The criteria have never been met before for Tourette's disorder or persistent (chronic) motor or a vocal tic disorder.

Tic disorders are hierarchical in order, that is Tourette's disorder, followed by persistent (chronic) motor or vocal tic disorder, followed by provisional tic disorder followed by other specified or unspecified tic disorders, such that once a tic disorder at one level of the hierarchy has been diagnosed, a lower level diagnosis cannot be made.

Cervantes and Matson [131] found that people with autism and ID were significantly more likely to experience tics and stereotypies than people who only had ID. Tourette's disorder and ASD share clinical features and possibly an overlapping aetiology. Darrow and colleagues [132] recruited 535 participants with Tourette's disorder and 234 of their family members and got them to complete Social Responsiveness Scale (SRS), 2nd edition, to characterize ASD symptoms. More children with Tourette's disorder met cut-off criteria for ASD (22.8%) than adults (8.7%). The elevated rate in children was primarily due to high scores on the SRS Repetitive and Restricted Behaviours (RRB) subscale. Higher observed rates of ASD among children affected by Tourette's disorder may in part be due to difficulty in discriminating complex tics and OCD symptoms from ASD

symptoms. Careful examination of ASD-specific symptom patterns (social communication vs. repetitive behaviours) is essential.

Self-injurious behaviour has been reported in up to 60% of people with Tourette's disorder. Mathews and colleagues [133] found that 29% of their sample had self-injury and 4% had severe self-injury. Mild to moderate symptoms correlated with obsessive and compulsive symptoms, while severe self-injury was correlated with variables related to affect or impulse dysregulation.

In their cross-sectional study of a cohort of people with tuberous sclerosis, Raznahan and colleagues [134] found two people with a tic disorder; it is not clear whether they also had ID. Shelley and colleagues [135] reported a case of a young man with Smith-Magenis syndrome and Tourette disorder suggesting that the co-occurrence of the two conditions may reflect common endophenotypic mechanisms underpinning the complex genetic disorders.

Barabas and colleagues [136] reported three cases of people with Down syndrome who also had characteristic features of Tourette's disorder with multiple motor and vocal tics. Kerbeshian and Burd [137] examined the North Dakota Tourette Registry retrospectively and found that five people also had Down syndrome, out of 70 adults and 188 children and adolescents, which gave an association rate of 2% between Down syndrome and Tourette's disorder.

Tartaglia and colleagues [138] reported on medical and psychological aspects of XXYY syndrome having looked at a large cohort of 95 individuals from the USA, Canada, the UK, and Australia and found that 18.9% had a tic disorder.

Schneider and colleagues [139] described five male patients from three unrelated families with fragile X syndrome who presented with motor and phonic tics. Four of them fulfilled the criteria for the diagnosis of Tourette's disorder and the fifth for adult onset tic disorder. As the onset of tics in all the individuals was considerably later than usual, authors suggest testing for Fragile X syndrome in people with Tourette's disorder complicated by ID and dysmorphic features.

Treatment of Tic disorders include both pharmacological intervention such as risperidone, haloperidol, pimozide, aripiprazole, clonidine, and non-pharmacological interventions such as symptom focused behaviour therapy like habit reversal training and exposure and response prevention therapy [140].

Tics and stereotypies are more common in people with more complex neurodevelopmental disorders, such as those with co-occurring ID and ASD, than in people with ID alone.

18.4 Communication Disorders

People with mild and moderate ID are usually able to communicate using speech. Those with more severe degrees of ID may be able to utter single words, and caregivers use body language, pictures, signs, and other enhanced forms of communication to anticipate the needs of these people. Language disorder is strongly associated with other NDDs such as ADHD and ASD [2].

People with ID may have communication needs that must be fully recognized in order to understand how to support them. For example, articulation difficulties in expression may lead to stammering, slurred speech, echolalia (repeating the same word or repeating the last word that has been said to them), fluctuating pitch, and intonation. Restrictive speech and repetitive speech are common in people with ASD.

Communication impairments can be described in three aspects of communication. These are (a) comprehension (how much someone understands what is being spoken), (b) expression (how someone communicates their needs), and (c) social Interaction: how someone seeks interactions with others. Most people would be expected to have better understanding skills (comprehensive speech) than expressive speech. However, some people can understand more language than they can communicate themselves. Developmental language disorders are not always easily identified and might not become apparent until a child begins school. This can cause problems with academic performance and overall functioning.

18.4.1 Speech Sound Disorder

Speech sound production is the clear articulation of individual sounds that in combination produce spoken words. Learning to produce speech sounds clearly and later connect speech fluently are developmental skills that follow a pattern. Normally developing children may shorten words and syllables during this process but produce mostly intelligible speech by 3 years. Most speech sounds and most words should be pronounced accurately by the age of 7 years according to age and community norms with the most frequently misarticulated sounds being *l*, *r*, *s*, *z*, *th*, *ch*, *dzh*, and *zh*, up to the age of 8 years.

Speech sound production requires phonological knowledge of the sound and the ability to coordinate the movements of the jaw, tongue, and lips – the articulators with breathing and vocalizing. Children with speech sound disorder could have difficulties with knowing what the sound is and also with coordination, but the underlying causes could be heterogeneous. A diagnosis can be made only when the level of speech is not what would be expected at that developmental stage for the child. The examples are (a) no gestures or response to words at 9 months, (b) no babbled phrases at 12 months, (c) no clear words at 18 months, (d) has less than 50 words and does not produce 2 words sentences at 2 years, (e) does not ask 'what' and 'who' questions, does not use pronouns (e.g. 'I', 'me'), and does not understand 3 words commands at 3 years, and (f) does not ask 'why', 'when', and 'how' questions and cannot count up to 20 by age 4 years.

- **DSM-5 Diagnostic Criteria [2]**

A. Persistent difficulty with speech sound production that interferes with speech intelligibility or prevents verbal communication of messages.
B. The disturbance causes limitation in effective communication that interferes with social participation, academic achievement, or occupational performance, individually or in any combination.
C. The onset of symptoms is in the early developmental period.
D. The difficulties are not attributable to congenital or acquired conditions such as cerebral palsy, cleft palate, deafness or hearing loss, traumatic brain injury, or other neurological conditions.

In many children with ID conditions like cerebral palsy, cleft palate, hearing problem, and other neurological conditions will be present.

A study carried out in Edinburgh [141] found that in a large sample of ID adults, 12.7% had no speech and 53% had speech problems, primarily in the form of difficulties in intelligibility. Shriberg and colleagues [142] also found a high percentage (43%) of persistent speech errors in their Idiopathic ID group which would affect their intelligibility. Extensive work has been carried out in the acquisition of language by children with Down syndrome, and this shows differences emerging at the transition into first words with delays characteristic of developmentally younger children, while consonant errors, as well as phonological processes (patterns of sound errors such as deletion of final consonants), are similar to the patterns in younger typically developing children at similar mental age levels [143, 144]. Although children with Down syndrome use phonological processes or sound patterns that are similar to those used by typically developing children, they eliminate these processes at a slower rate. The boys with Down syndrome generally use reduced word shapes, by omitted syllables (e.g. 'bana' for 'banana'), reduced consonant clusters (e.g. 'bu' for 'blue'), and deleted consonants, e.g. spoo for spoon [145].

Delayed emergence of speech and restricted and atypical phoneme repertoires are common findings in people with 22q deletion syndrome which results in poor speech intelligibility, particularly in younger children. Although there is progression and improvement in speech in school age children, speech sounds deficits may persist into late childhood and adolescence. Common speech sound disorders include hypernasality, high pitch, dysphonia, restricted and delayed speech sound acquisition, articulation impairments, abnormal speech prosody, childhood apraxia of speech, and speech motor delay [146].

The FOX2P gene has been called the speech gene encoding a transcription factor involved in speech and language acquisition. There is increasing evidence that dysregulated FOXP2 activity may also be instrumental in human oncogenesis [147]. All FOXP2-related speech and language disorders, regardless of the underlying genetic alteration, have a core phenotype: childhood apraxia of speech (CAS), a disorder of speech motor programming or planning that affects the production, sequencing, timing, and stress of sounds, syllables, and words. All individuals with CAS (whether caused by an alteration of FOXP2 or of an unknown cause) have difficulties in automatically and accurately sequencing speech sounds into syllables, syllables into words, and words into sentences with the correct prosody [148]. Some of these individuals may have global developmental delay and ASD too.

Speech delay and articulation problems occur in children and adults with ASD. Rapin and colleagues [149] analysed language data of children with ASD and reported more than a quarter of the group as having phonological speech problems. Cleland and colleagues [150] found speech problems in 12% when assessing a group of children with ASD without ID; however, 41% of the children produced at least some speech errors. Similarly, Shriberg and colleagues [151] reported 'speech delay', defined as mainly phonological speech errors affecting intelligibility (but also including articulation errors) occurring in 15% of their sample of ASD children.

18.4.2 Childhood Onset Fluency Disorder (Stuttering)

Essentially, this is a disorder with a disturbance in the normal fluency and time patterning of speech which is inappropriate for the individual's age. It is characterized by frequent repetitions or prolongations of words or syllables and by other types of speech dysfluencies including broken words, audible or silent blocking, word substitutions to avoid difficult words, words produced with an excess of physical tension, and monosyllabic whole word repetitions. This may interfere with academic or occupational achievement and social interactions and is usually worse when there is pressure to communicate. It may be absent when talking to pets or inanimate objects or singing. The child may develop motor movements such as eye blinks, tics, tremors, etc. together with dysfluency. The onset of the condition can be insidious or sudden, with the age of onset ranging from 2 to 7 years. Longitudinal studies show that 65–85% of children recover from dysfluency, and severity of the disorder at age 8 years predicts recovery or persistence into adolescence and beyond [2].

DSM-5 Diagnostic Criteria [2]

A. Disturbances in the normal fluency and time patterning of speech that are inappropriate for the individual's age and language skills, persist over time, and are characterized by frequent and marked occurrences of one (or more) of the following:
   1. Sound and syllable repetitions.
   2. Sound prolongations of consonants as well as vowels.
   3. Broken words (e.g. pauses within a word).
   4. Audible or silent blocking (filled or unfilled pauses in speech).
   5. Circumlocutions (word substitutions to avoid problematic words).
   6. Words are pronounced with excessive physical tension.
   7. Monosyllabic whole word repetitions (e.g. 'I-I-I-I see him').

B. The disturbance causes anxiety about speaking or limitation in effective communication, social participation, or academic or occupational performance, individually or in any combination.

C. The onset of symptoms is in the early developmental period. (Note: Later onset cases are diagnosed as adult onset fluency disorder.)

D. The disturbance is not attributable to a speech motor or sensory deficit, dysfluency associated with neurological insult (e.g. stroke, tumour, trauma), or another medical condition and is not better explained by another mental disorder.

Data collected through Centres for Disease Control and Prevention indicate a prevalence rate of 1.6% of children between 3 and 17 years of age [152]. Stansfield [141] carried out a study on the prevalence of idiopathic dysfluency or stuttering in adults with ID using NHS and local authority provision in Edinburgh and found a total prevalence of 6.31% of speech dysfluency. Half of the dysfluent group had Down syndrome and they formed 14.7% of the Down syndrome group. Kent and Vorperian [153] have reviewed the studies on children with Down syndrome and concluded that stuttering has been demonstrated in 10–45%, on an average of 31% of individuals. Briley and Ellis [154] found that the presence of at least one disabling developmental condition was 5.5 times higher in children who stutter when compared with children who do not stutter. The presence of stuttering was also associated with higher odds of each of the following coexisting developmental and neurological conditions: ID (odds ratio [OR] = 6.67, p < 0.001), SLDs (OR = 5.45, $p < 0.001$), ADHD (OR = 3.09, $p < 0.001$), seizures (OR = 7.52, $p < 0.001$), ASD (OR = 5.48, $p < 0.001$), and any other developmental delay (OR = 7.10, $p < 0.001$).

Stuttered speech has been reported in people with Fragile X syndrome. Paul and colleagues [155] reported 2.9% stuttered syllables on average in the spontaneous speech of a group of adult males with fragile X, compared with 2.75% in a group with non-specific ID and 2% in a group with ASD. Ferrier and colleagues [156] reported a mean percentage of stuttering of 4.9% in fragile X syndrome, 1.6% in ASD, and 6.1% in Down syndrome. Van Borsel and Tetnowski [157] concluded that individuals with fragile X syndrome appear to have a higher prevalence of stuttering than non-specific forms of ID and ASD, but a lower prevalence than individuals diagnosed with Down syndrome.

Prader Willi syndrome is also associated with speech dysfluency. Van Borsel and Tetnowski [157] suggested that an in-depth analysis of the distribution of stuttering moments has been shown to differentiate the stuttering associated with Prader-Willi syndrome from developmental stuttering.

18.4.3 Social (Pragmatic) Communication Disorder

This condition is characterized by a primary difficulty with the pragmatics or social use of language and communication. The individual is unable to understand and follow social cues of verbal and nonverbal communication according to context and the needs of the listener or the situation, and follow the rules of conversation and storytelling. This could lead to functional limitations in effective communication, social participation, and developing social relationships, academic achievements, or occupational performance. The most common association is language impairment with a history of language delay. Individuals may avoid social interactions [2].

As the diagnosis is dependent on adequate language development, this disorder is rare before the age of 4 years. The outcome of the disorder is variable, with some children improving substantially, and others continuing to have difficulties into adulthood.

A diagnosis of social communication disorder can only be considered if the child did not display repetitive restricted patterns of behaviours, interests, and activities during the early developmental period. Social communication skills may also be deficient in individuals with ID, but a separate diagnosis is not made unless social communications are clearly in excess of the intellectual limitations [2].

- **DSM-5 Diagnostic Criteria [2]**

A. Persistent difficulties in the social use of verbal and nonverbal communication as manifested by all of the following:
   1. Deficits in using communication for social purposes, such as greeting and sharing information, in a manner that is appropriate for the social context.
   2. Impairment of the ability to change communication to match the context or the needs of the listener, such as speaking differently in a classroom rather than in a playground, talking differently to a child than an adult, and avoiding overly formal language.

3. Difficulties following rules of conversation and storytelling, such as turn taking in conversation, rephrasing when misunderstood, and knowing how to use verbal and nonverbal signs to regulate interaction.
4. Difficulties understanding what is not explicitly stated (e.g. making inferences) and nonliteral or ambiguous meanings of language (e.g. idioms, humour, metaphors, multiple meanings that depend on the context for interpretation).

B. The deficits result in functional limitation in effective communication, social participation, social relationships, academic achievement, or occupational performance, individually or in combination.
C. The onset of the symptoms is in the early developmental period (but deficits may not become fully manifest until social communication demands exceed limited capacities).
D. Symptoms are not attributable to another medical or neurological condition or low abilities in the domain of word structure and grammar and are not better explained by autism spectrum disorder, intellectual disability (intellectual developmental disorder), global developmental delay, or another mental disorder.

Given that the concept has been recently expanded in DSM-5 [2], it will be necessary to examine and evaluate the validity of the criteria for the disorder before estimating the prevalence [158]. However, Ouss et al. [159] found that two of their patients with Dravet syndrome with ID also met the criteria for social communication disorder. Despite DSM-5 [2] excluding ID and ASD for the above diagnosis, the social communication disorder is very common in ASD and also to some extent in the ID population. It has been shown that prior to 2013, before social communication disorder was added to the DSM-5 as a stand-alone diagnosis, individuals with this condition would have been diagnosed with ASD, most often pervasive developmental disorder not otherwise specified or Asperger's syndrome [160].

18.4.4 Unspecified Communication Disorder

This category is applied to presentations in which there is a communication disorder significant enough to cause clinically significant distress or impairment in social, or other important areas of functioning but does not meet full criteria for any of the communication disorders or any of the disorders of the NDDs. DSM-5 [2] specifies that this category is used in situations when the clinician chooses not to specify the reason that the criteria are not met and includes presentations in which there is insufficient information to make a more specific diagnosis.

Stansfield [141] in the comprehensive prevalence study of health and local authority provision in Edinburgh found that 53% had speech problems including difficulties in intelligibility, comprehension, expressive language, voice, fluency, and no speech.

The role of functional communication assessment is highlighted by Cascella [161] in his review of standardized speech and language tests. He suggests that a functional assessment approach lets the clinician consider the variety of communication contexts in which the student participates (e.g. home, school, and community). When the context is considered, the speech and language practitioner can evaluate communication opportunities and teachers' and family members' expectations in any given situation. This can be used in other situations too (Table 18.2).

Children and adults with intellectual disabilities manifest various types of communication disorders including speech sound disorder, speech fluency disorder (stuttering), and social (pragmatic) communication disorder.

Table 18.2 Tips to improve communication with people who have mild to moderate ID and can speak *(see Deb & Iyer, 2005)* [162]

The nature of the person's relationship with the interviewer may influence the interview process.	Some may have set ways of responding to questions irrespective of their content. For example, they may answer 'yes' or alternatively 'no' to all questions irrespective of their contents. This may tell you they are not understanding what you are saying.	The tendency of the person with ID to echo the last option can be minimized by asking her/him to confirm both options and choose one.
Be aware that the person with ID may have negative experiences of the interview process.	Avoid closed questions and always use open questions.	Avoid complex phrasing and those involving abstract concepts such as 'extent', 'all the time', and 'low spirits'.
They may also be concerned about the consequences of the interview.	Establish first the person's preferred method of communication and inform other professionals about that.	Ask the person with intellectual disabilities to clarify statements using examples.
Wait for them to respond to your question and assess whether you feel they have understood you. If necessary, repeat the question.	Be flexible about the length of interviews, such as using several shorter interviews instead of one long interview.	Avoid double negatives. For example, 'You do not refuse medication, do you?' may be usefully phrased as 'Do you take your meds?'
Ensure you have made all reasonable adjustments for the interview.	Make sure you allow plenty of processing time when you have asked a question.	Events could be used as anchors to get a time frame. For example, 'Are you feeling sad since your birthday?'
Interview in an unfamiliar location may pose its own anxieties.	Start with easy questions to gain the person's confidence.	Use simple sentences using short words, in the present tense.
The interview may involve a change in routine which may precipitate unwanted behaviour.	Be patient and wait for the answer to the first question before moving to the next.	Watch out for non-verbal signs especially distress and discomfort.
Remember that sensory difficulties such as impaired hearing or vision are likely to affect the interview.	Be aware of body language. For example, during the discussion of a recent bereavement, an autistic person started to rock in his chair, which is an early sign of distress. You may need to stop the discussion or change the topic.	You should be aware of autistic masking where someone says something, but it is not exactly concise. The interviewer needs to use insight and interpretation to establish what the person actually means.
Reduce background noise, allow plenty of time and ensure you understand each individual's communicative needs, and try to establish eye contact to get their attention.	You can use reverse wording. For example, the question 'Do you have trouble sleeping?' can be followed by 'Do you sleep well?' in the latter part of the interview.	For example, BJ, a 13-year-old boy assessed for depression talked about having his 'tummy cut'. His mother clarified that this referred to his preoccupation with his weight of late. He had recently watched a programme on television about surgical treatments for obesity.

18.4.5 Strategies to Support People with ID Who Have Communication Difficulties

- **Useful Links**

► https://toolsfortalking.wordpress.com/resources/

► http://helensandersonassociates.co.uk/person-centred-practice/person-centred-thinking-tools/

Tips to improve communication with people who have severe to profound ID and cannot speak (► https://spectrom.wixsite.com/project).

People with severe and profound ID and without verbal skills may need to use non-verbal methods of communication. It is advisable to have a 'communication partner' usually the key care/support staff or a family member who has developed a good relationship with the person and has developed skills to communicate with the person non-verbally. This 'communication partner' can act as an interpreter and could inform other people, particularly professionals about the best way to communicate with the person with ID [163]. The person with ID may respond better to environmental cues such as a 'familiar voice', 'gesture', and 'touch'.

Following are the examples of non-verbal communication techniques that could be used (augmentative and alternative communication (AAC)/communication aids) [163].

► https://www.youtube.com/watch?v=YzVvYj1RfKI

- **Examples of AAC**

- *Signing* includes the use of signs and symbols to convey relevant key messages to aid comprehension. For example, signing 'we need to visit the park after you finish your lunch' would include signs 'lunch', 'shoes', and 'park'.
- *On-body singing* is used for people with visual impairments. Examples of this include placing one person's hand on top of or under the partner's hand so the communicative partner can read the signs.
- *Objects of references* include the use of objects to convey messages. For example, using a football to signify that the activity football game is about to take place.
- You can use *photos*, *pictures*, and *symbols* to convey any message. For example, you can use a photo of a relative to show that his or her relative has come to visit.
- Picture Exchange Communication System (*PECS*) is a training method that uses symbols to teach the person communication skills of increasing complexity. For example, the person first learns to use simple symbols for things she or he wants and then learns to use this in different settings. The person then learns to use multiple pictures to form sentences, expand their sentences, respond to questions, and comment on things.
- A person can communicate using their eyes to point to a specific symbol on an *E-tran* frame. For example, a person can look at food symbols to show that she or he wants to eat.
- *Talking mats* includes using symbols or pictures to communicate about a topic. For example, a person can stick symbols or pictures of how they feel about swimming, playing football, or any other activity if the topic is an activity.
- High-tech devices such as *VOCAs* can generate speech for people who have no means to communicate. For example, it can be used to inform support staff of what the person wants.
- *Intensive interaction* is a person-centred approach to enhance social communication, particularly among people who have severe and profound ID, sensory impairment, and autism. Following are the main principles on which this intervention is based.
  - Doing a sequence of activities with the person
  - Attending and concentrating
  - Taking turns in exchange for behaviour
  - Sharing personal space
  - Using and understanding eye contact (face and mind-reading)
  - Using and understanding facial expression (face and mind-reading)
  - Using and understanding physical contacts

- Using and understanding non-verbal communication
- Vocalizing and using vocalization meaningfully (including speech)
- Regulating and controlling arousal level
- Considering emotional and well-being issues

Here is a link for more information on intensive interaction: ► https://www.intensiveinteraction.org/

Use short videos and visual aids such as photographs, line drawings, visual scripts or social stories, and communication boards or PECS symbols.

Always seek the advice and support of an experienced professional such as a speech and language therapist specializing in ID to guide which of these approaches are likely to be the most successful with an individual.

Use '*Tools for talking*' to identify what the person with ID wants or needs to improve her or his quality of life. Tools for talking can be used for people from ethnic minorities and who have low communication skills.

► https://toolsfortalking.wordpress.com/resources/

Example of person-centred tools developed by Helen and Sanderson Associates to understand communication and communicate effectively.

► http://helensandersonassociates.co.uk/person-centred-practice/person-centred-thinking-tools/

Always try to understand and meet the needs of the person. If a person enjoys routine, then a visual timetable of the activities she or he will participate in may be created. The use of sign languages such as Makaton or British Sign Language, and objects of reference such as plates to signify dinner/lunch time is another option.

Pay attention to the body language of a person with ID. For example, you may identify a person who is about to display aggressive behaviour through cues such as fist clenching, lip biting, or stomping of feet. The facial expression, body posture, and tone of voice will provide many cues. Examples of positive cues include smiling, eye contact, relaxed body, responding well to social interaction, and so on. Examples of negative cues include restless, rigid body, feet tapping, slumped posture, a relaxed tone of voice, and so on.

Useful Links

► https://www.mencap.org.uk/sites/default/files/2016-06/hospitalcommunicationbook.pdf

► http://www.bild.org.uk/EasySiteWeb/GatewayLink.aspx?alId=3338

► https://www.asha.org/NJC/AAC/

Key Points

- Specific learning, motor, and communication disorders start at childhood and often persist into adulthood, with changes of symptomatology across time depending on age and environmental requirements.
- If an ID is present, a specific learning disorder can be diagnosed additionally only if the specific learning difficulties are more severe than those attributable to ID.
- Current classificatory systems introduced significant changes to diagnostic criteria of specific learning, motor, and communication disorders in comparison to previous editions.
- Specific learning disorders are among the most common neurodevelopmental disorders, with a prevalence of 5–15% in school-age children across different languages and cultures, and approximately 4% in adults.
- The identification of the causes of SLD is complicated by their heterogeneity. Nevertheless, most experts agree on a biological origin of the cognitive anomalies that underlie behavioural symptoms of the disorders.
- The assessment of SLDs should be made by specialists with appropriate training and expertise and address cognitive, speech and language, medical, psychological, and educational aspects.
- Early intervention is critical for all SLDs to avoid or reduce negative outcome.

- Various treatments and support programmes are available for SLDs, showing a varied rate of success.
- Children with motor disorders may be substantially delayed in reaching motor milestones, make repetitive and involuntary movements, or have physical or verbal tics, which cause impairment and result in negative physical, psychological, and social consequences across the whole lifespan.
- Motor disorders frequently co-occur with ID and/or ASD, supporting the notion that motor, cognitive, and social-communication functioning are interrelated.
- Stereotyped or repetitive motor movements represent a distinguishing feature of ASD and are not uncommon in persons with ID, especially in those with a moderate-to-severe degree of severity. In these cases, stereotypic movement disorder is diagnosed only when it is sufficiently severe to constitute an adjunctive clinical focus.
- Persons with more complicated neurodevelopmental disorders, such as ID and ASD co-occurring, have more tics and stereotyped or repetitive motor movements than people with ID alone.
- Speech sound disorder, speech fluency disorder (stuttering), and social (pragmatic) communication disorder are all common communication disorders in children and adults with ID.

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Behavioural and Emotional Disorders with Onset Usually Occurring in Childhood and Adolescence

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Contents

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Learning Objectives

- Conduct disorder (CD) in people with intellectual disabilities
- Oppositional defiant disorder (ODD) in people with intellectual disabilities
- Intermittent explosive disorder (IED) in people with intellectual disabilities
- Stereotyped movement disorders in people with intellectual disabilities
- Other behavioural disorders with onset in childhood and adolescence
- Assessment of CD, ODD, and IED in people with intellectual disabilities

- Disruptive, impulse-control, and conduct disorders is a new category under DSM-5 classification system.
- These disorders originate in childhood and often continue into adulthood.
- There is a strong association between these disorders and intellectual disabilities (ID).
- They include conduct disorder, oppositional defiant disorder which is a less severe form of conduct disorder, and intermittent explosive disorder, which was part of impulse control disorder in the previous DSM classifications.
- By definition many of these disorders have to show an intention behind the behaviour, which is not easy to determine, particularly among those who have severe and profound ID.
- Other impulse control disorders include pyromania, kleptomania, gambling disorder, compulsive sexual behaviour disorder, trichotillomania, and excoriation disorder.
- Body-focussed repetitive behaviour, on the other hand, includes stereotyped movement disorder.
- The aetiology of these disorders is complex and often is the outcome of an interaction between internal factors within the person and external factors such as the environment.
- Therefore, the management of these disorders requires input from a multitude of professionals with relevant and specific skills to provide a person-centred assessment and management.
- The ultimate aim should be to improve the quality of life of the person with ID and their family members.

Tip

- The DSM-5 criteria are often difficult to apply to people with intellectual disabilities for conduct disorder, intermittent explosive disorder, and oppositional defiant disorder.
- Therefore, instead of making a psychiatric diagnosis as per DSM-5 criteria, it is more important to describe the behaviour which will lead to a better management.

19.1 Disruptive, Impulse-Control, and Conduct Disorders

19.1.1 Introduction

Disruptive, impulse-control, and conduct disorders (CD) are characterised by impairment in the ability to control emotions and behaviours to an extent that the resulting behaviours have a clinically significant impact on an individual and her or his interactions with others and/or society. The six specified disruptive, impulse-control, and CD are (a) antisocial personality disorder, (b) CD, (c) oppositional defiant disorder (ODD), (d) intermittent explosive disorder (IED), (e) kleptomania, and (f) pyromania. In a way ODD is considered as the less severe form of CD. In order to meet DSM-5 [1] diagnostic criteria for disruptive, impulse-control, and CD, there should be evidence that an individual present with behaviours that violate the rights of others (e.g. aggression, destruction of property) and/or bring the individual into significant conflict with societal norms or authority figures.

In the previous DSM classification such as the DSM-IV-TR [2], CD and ODD were included in the attention deficit hyperactivity

disorder (ADHD) and disruptive behaviour disorder category. In the previous version of DSM, 'Impulse-Control Disorders' included IED, kleptomania, and pyromania. These are now classified within the new DSM-5 classification as disruptive, impulse-control, and CD. Also included in this category are CD, ODD (both previously classed as Disorders Usually First Diagnosed in Infancy, Childhood, or Adolescence) and antisocial personality disorder (see ▶ Chap. 31).

Previously, trichotillomania and pathological gambling were also included in Impulse-Control Disorders Not Elsewhere Classified. However, in the DSM-5, these disorders are now included within obsessive-compulsive and related disorders and substance-related and addictive disorders respectively. Habit disorders such as finger sucking, body rocking, and bruxism are classed as stereotypic movement disorders and are included among motor disorders under the 'neurodevelopmental disorders' section of DSM-5 [1]. Antisocial personality disorder is included in DSM-5 [1] both under the 'Disruptive, Impulse-control, and CD' category as well as under the 'Personality Disorder' category.

Diagnoses of other specified or unspecified disruptive, impulse-control, and CD may be given when an individual has symptoms which are characteristic of the specific disorders listed above but do not meet the full diagnostic criteria. These diagnoses may be used for individuals with presentations in which there is insufficient information to make a more specific diagnosis.

19.1.2 Issues in the Use of Diagnosis in Intellectual Disabilities

There are a number of difficulties when using these terms in relation to people with intellectual disability (ID). These diagnostic categories are hardly used in day-to-day practice, instead a broader term such as problem (challenging) behaviour is used more widely both in clinical practice and in research. The term 'problem behaviour' is used to describe 'socially unacceptable behaviour that causes distress, harm or disadvantage to the persons themselves or to other people, and usually requires some intervention' [3] and includes behaviours such as verbal or physical aggression to others, physical aggression to property and/or self, screaming, shouting, indiscriminate wandering, objectionable personal habits including smearing of faeces and pica, etc., antisocial behaviour including taking others' possession without permission, etc., unacceptable sexual behaviours, and stereotyped movement not caused by a neurological disorder.

DSM-5 [1] reports that these disorders usually develop in childhood or adolescence. This criterion applies equally to people with ID. However, for individuals with ID there may be a higher likelihood that symptoms remain unrecognised or are misinterpreted until adulthood because of the difficulties encountered when assessing and diagnosing psychiatric and behavioural disorders in this group [4, 5], particularly among children and adolescents. For individuals with ID, before making a diagnosis one has to consider whether the behaviour is both age and developmentally appropriate as in many people with ID, developmental age does not correspond to their chronological age.

One of the reasons that diagnoses such as CD and ODD are rarely used in ID literature is that these disorders assume that the person is behaving with an intent, which is difficult to determine in most people with ID who exhibit problem behaviour. Another problem is with the DSM-5 criterion that stipulates that a diagnosis of IED cannot be made before the age 6 years. As individuals with severe and profound ID are likely to have a mental age below 6 years, this diagnosis cannot be applied to this group of people [6].

Among the general population who do not have ID, disruptive, impulse-control, and CD are known to commonly occur with each other, substance use disorders, and ADHD. This might as well be the case for people with ID. Because of these difficulties with diagnostic categories Deb and colleagues [6] suggested that a diagnosis based on a group of behaviours may be better for an appropriate formulation of management than a syndromic categorisation.

The DSM-5 [1] acknowledges that many of the behaviours associated with disruptive, impulse-control, and CD are present in the general population and that when formulating a diagnosis 'the frequency, persistence, pervasiveness across situations, and impairment associated with the behaviors…[should] be considered relative to what is normative for a person's age, gender, and culture'. For individuals with ID, attention should also be paid to what is considered normative relative to an individual's developmental level.

19.1.3 Assessment of Behaviour

As a diagnosis of disruptive, impulse-control, and CD cannot be made in most people with ID, particularly for those who have severe and profound ID, Deb and colleagues in DM-ID-2 [6] recommended that in practice, clinicians should make a diagnosis of 'problem (challenging) behaviour' outside the DSM-5 [1] framework and describe the frequency and severity of these behaviours along with severity ratings (mild, moderate, severe) and frequency ratings (low, medium, high). They recommend a multidisciplinary, person-centred assessment of the behaviours using a schema based on the assessment of the *B*ehaviour, the *P*erson showing the behaviour, *P*sychological and psychiatric factors, *M*edical and organic factors, and *S*ocial factors (BMPPS) (see ► Chap. 7 and ► https://spectrom.wixsite.com/project).

Assessment of behaviour should include the person's history of problem behaviour; baseline behaviour prior to the onset of the current behaviour; how the behaviour started, whether they appeared gradually over time or relatively abruptly, perhaps precipitated by an acute event; the frequency, severity, and duration of the behaviour; the nature, content, and context of the behaviour, as some behaviour may occur in certain circumstances/settings but not in others, associated behaviours; and the impact of the behaviour on the person's life, others' lives, and the environment. The behaviour may lead to reduced quality of life for the individual and her/his caregiver. The behaviour may lead to reduced access to services including education, day service, and employment opportunity, and may lead to a threatened or actual loss of placement in a residential setting or day placement. The behaviour may lead to reduced social activities including leisure activities, access to friends, etc.

In severe cases the individual may end up being physically restrained, inappropriately medicated, or taken to a hospital or police station. All these scenarios may subsequently have their impact on the individuals, their behaviour, and their caregivers. For example, some of these outcomes may work as perpetuating factors for the ongoing behaviour. These outcomes may be seen as inappropriate or excessive and the individuals or their caregivers may perceive them as punishments.

A risk assessment should be carried out which should include (a) risk to others, (b) risk to the individual, (c) risk to the environment, and (d) other risks. Where possible use a standardised method of risk assessment but always make a record of the assessment and review them periodically.

19.1.3.1 Assessment of Medical and Organic Factors Should Include

- Chronic physical conditions such as headache, toothache, and pain in other parts of the body.
- Medical conditions such as acid reflux, chest infections that are common in people with ID.
- Epilepsy which is common in people with ID [7].
- Neurological conditions such as cerebral palsy.
- Genetic conditions such as Prader-Willi syndrome, Fragile X syndrome, and Lesch-Nyhan syndrome which are known to be associated with problem behaviour [10].
- Hearing and visual impairments which can lead to problem behaviour form frustration.
- Sensory processing issues in people with ASD.

- Communication problems where the individual may use problem behaviour to communicate or a discrepancy between expression and comprehension.
- Physical disabilities causing frustration.
- Illicit drug and alcohol use, although this may be more of an issue in people with borderline intelligence.
- Adverse effects of prescribed medicines.

19.1.3.2 Assessment of Psychological and Psychiatric Factors

Psychiatric disorders such as mood disorders, schizophrenia, other psychoses, and anxiety-related disorders may lead to problem behaviour in people with ID [4]. ASD and ADHD are also common comorbidities resulting in higher rates of problem behaviour. It is important to investigate for these conditions as the management of the problem behaviour will depend on the management of the underlying condition.

Psychological and emotional factors such as bereavement and trauma and abuse (physical, emotional, or sexual), historical, ongoing, recurring or new, or stress and relationship difficulties may lead to loss of self-esteem and isolation, causing and perpetuating problem behaviour.

19.1.3.3 Assessment of Social Factors

Environmental factors have an important role to play as some people with ID may present with problem behaviour if they find themselves in an environment which they find too demanding or understimulating. Personalities and attitudes of others towards the person, including care givers can also affect the person with ID. Needless to say, management styles could impact on the individual too, for example confrontational attitude.

19.1.4 Conduct Disorder

The essential feature of Conduct Disorder (CD) is a repetitive and persistent pattern of behaviour that violates the basic rights of others or that breaks major age-appropriate societal norms or rules. Behaviours are grouped into four major categories: (a) aggression to people or animals, (b) destruction of property, (c) deceitfulness or theft, and (d) serious violation of rules. As with all psychiatric conditions, the presentation must cause clinically significant impairment in social, academic, or occupational functioning.

These disorders could be of (a) childhood onset where the behaviour is present before the age of 10 years or (b) adolescent onset where no symptoms are observed before the age of 10 years. Unspecified onset category applies to those for whom where there is insufficient information to determine the age of onset of symptoms.

Following are the DSM-5 [1] criteria for diagnosis of CD, none of which could be applied to those who have severe and profound ID as it is not possible to determine the underlying intention behind the behaviour. All three criteria (A, B, C) must be met in order to qualify for the diagnosis.

- **DSM-5 Diagnostic *Criteria A* [1]**

Repetitive and persistent pattern of behaviour in which the basic rights of others or major age-appropriate societal norms or rules are violated, as manifested by the presence of three or more of the following criteria in the past 12 months, with at least one criterion present in the past 6 months. The clinician should assess age appropriateness of the behaviour and ascertain that the individual understands societal rules and norms.

19.1.4.1 Aggression to People and Animals

- Often bullies, threatens, or intimidates others.
- Often initiates physical fights.
- Has used a weapon that can cause serious physical harm to others (e.g. bat, brick, broken bottle, knife, gun).
- Has been physically cruel to people.
- Has been physically cruel to animals.

In case of people with ID, it is important to ascertain that the individual displaying the

behaviour has understood the significance and impact of her or his actions.

19.1.4.2 Destruction of Property

- Has deliberately engaged in fire setting with the intention of causing serious damage.
- Has deliberately destroyed others' property (other than by fire setting).

In case of people with ID, clinician should assess intent behind the behaviour. Even some people with mild ID may have difficulties with communicating their thoughts and feelings. The intent is even more difficult to determine in people with severe ID.

19.1.4.3 Deceitfulness or Theft

- Has broken into someone else's house, building, or car.
- Often lies to obtain goods or favours or to avoid obligations (i.e. 'cons' others).
- Has stolen items of nontrivial value without confronting a victim (e.g. shoplifting, but without breaking and entering; forgery).

In case of people with ID, it is important to establish that the individual understands the concept of ownership.

19.1.4.4 Serious Violations of Rules

- Often stays out at night despite parental prohibitions, beginning before age 13 years.
- Has run away from home overnight at least twice while living in parental or parental surrogate home (or once without returning for a lengthy period).
- Is often truant from school, beginning before age 13 years.

In case of ID, it is important to carefully examine the home environment and the reasons why the individual is running away. For example, if there is evidence of inadequate care, neglect, or abuse, the relevance of this behaviour to the diagnosis should be questioned. Careful examination of school environment is necessary to rule out any reasonable cause such as distress due to bullying.

- **DSM-5 Diagnostic *Criterion B* [1]**

The disturbance in behaviour causes clinically significant impairment in social, academic, or occupational functioning.

- **DSM-5 Diagnostic *Criterion C* [1]**

If the individual is 18 years or older, criteria are not met for antisocial personality disorder.

- **Severity:**

Mild: few if any conduct problems in excess of those required to make the diagnosis *and* conduct problems cause only minor harm to others.

Moderate: number of conduct problems and effect on others intermediate between 'mild' and 'severe'.

Severe: many conduct problems in excess of those required to make the diagnosis *or* conduct problems cause considerable harm to others.

It cannot be assumed that the person with ID understands the basic rights of others and appropriate social norms. The four categories of behaviours to be rated in CD cannot be viewed as equivalent in the case of people with ID. There are examples of people with ID showing aggression towards people or animals and destroying property, but there are far fewer instances of deceitfulness or serious violation of rules. The difference is in the nature of the intent. Aggression and destruction can occur when the person is frustrated and does not have any other available outlet to express their frustration. Serious violation of rules could also occur in the absence of intent, either because the person does not understand the rules or because the person does not appreciate the consequences of their actions. Deceit, however, is by definition an intended act.

Often people with ID function at a level much lower than their chronological age, which makes it difficult to determine the age appropriateness of the behaviour. When the function

of the behaviour (what the behaviour is trying to achieve) is understood, making a diagnosis of CD becomes difficult, even for those functioning at borderline levels of intelligence.

Clinicians should focus on the interaction between social environment and the individual and not assume that the responsibility lies with the individual for which it is necessary to use multiple sources of information during assessment. People with ID themselves can be essential sources of information.

Assessment should focus on obtaining the most direct information regarding current behaviours. Information on previous problem behaviour if any should be collected to determine onset, predisposing, precipitating, and perpetuating factors for the individual's behaviour. Frequency and severity of each current and past behaviours should be assessed [11] along with repetitiveness and chronicity to indicate the onset and stability over time, and developmental appropriateness.

The determination of age appropriateness depends on an understanding of both normal developmental patterns and the behavioural patterns presented by the person. For example, one might be reluctant to assign the label 'stealing' to a person for whom it is unclear whether she or he understands the concept of personal ownership.

Assessment of environment plays an important part in determining the diagnosis of CD. Behaviours that are inconsistent across settings suggest environmental influence on behaviour. The behaviours may be generated by environmental factors which would weigh against a diagnosis of CD. Importance of making assessments across multiple settings is that multiple informants are likely to be used, thus somewhat mitigating reporting bias.

19.1.5 Oppositional Defiant Disorder

The essential feature of Oppositional Defiant Disorder (ODD) is a recurrent pattern of angry, irritable mood with defiant, disobedient, and hostile vindictive behaviour towards at least one individual who is not a sibling. The behaviour must occur more frequently than would occur typically in individuals of comparable age and development and has to cause significant impairment in social, academic, or occupational functioning. All three DSM-5 [1] criteria (A, B, C) must be met in order to qualify for the diagnosis.

The DSM-5 [1] criteria could not be applied for those who have severe and profound ID and who lack communication skills but could be applied to those who have mild to moderate ID and could communicate with some caveats (see next section).

- **DSM-5 Diagnostic** *Criteria A* **[1]**

Pattern of angry/irritable mood, argumentative/defiant behaviour, or vindictiveness lasting at least 6 months, during which four or more of the following are present:

1. Often loses temper
2. Often touchy or easily annoyed
3. Often angry and resentful
4. Often argues with authority figures or, for children and adolescents, with adults
5. Often actively defies or refuses to comply with requests from authority figures or with rules
6. Often deliberately annoys others
7. Often blames others for his or her mistakes or misbehaviour
8. Spiteful or vindictive at least twice within the past 6 months

To apply this diagnosis to people with ID, the clinician must rule out any other reasons for the behaviour (see ▶ Chap. 7). The clinician also needs to carry out a language and cognitive assessment to rule out comprehension and specific cognitive difficulties that might otherwise explain the noncompliance and other associated behaviour. Assessment of both intent and developmental age is also required. Consider a criterion met only if the behaviour occurs more frequently than is typically observed in individuals of comparable age and developmental level.

- **DSM-5 Diagnostic** *Criterion B* **[1]**

The disturbance in behaviour is associated with distress in the individual or others in the

immediate social context. Further, it causes clinically significant impairment in social, academic, or occupational functioning.

DSM-5 Diagnostic *Criterion C* [1]

The behaviours do not occur exclusively during the course of a psychotic disorder, mood disorder or substance use.

Rates of problem behaviour which may or may not include CD and ODD vary widely between 2% and 60% [4, 12, 13] depending on the diagnostic criteria used and the type of population studied. The rates may also vary depending on factors like age, gender, etc. (see ► Chap. 7) [4, 14, 15]. Impairments in impulse control may be associated with aggressive behaviour in people with ID and also borderline intellectual functioning [15, 16].

Given that the locus of control in their lives is often outside their control, people with ID are more likely to feel frustrated and for this to manifest itself as insufficient self-regulation and defiance. For this reason, the clinicians have to be mindful of the danger of possible overdiagnosis of ODD in people with ID.

People with severe ID may not have the necessary language and cognitive abilities to be able to meet some of the criteria, such as argue with others, blame others, or be vindictive. It is thus incumbent on clinicians to conduct a comprehensive assessment of all relevant factors, including physical and mental health status, quality of the environment, possible sources of stress, and possible abuse and come to an accurate formulation which may point away from such a diagnosis. A diagnosis of ODD often lends the impression that the responsibility for the problem lies mainly within the individual themselves, rather than an interaction between the individual and the social environment or with other systemic factors [6].

A thorough assessment is necessary addressing the following issues in order to make the correct diagnosis and formulate an appropriate management plan.

- The nature of the behaviours must be explored in detail, including the circumstances in which they occur, including the predisposing, precipitating, and perpetuating factors.
- Assess the amount of control the individual seems to have over these behaviours.
- Make a list of all current and previous behaviours being displayed.
- Clarify the referral question.
- Determine who are affected by the behaviour, where and when do they occur?
- Define them in each area of the individual's life.
- Determine whether these behaviours are developmentally appropriate or not?
- Obtain a careful biopsychosocial history, to chart onset, course, and contributing risk factors.
- Assess the strengths and weaknesses of the individual that may have an impact on the behaviour.
- Assess the individual's cognitive, communication, occupational, and social function.
- Exclude comorbidities such as anxiety disorder, ASD, ADHD, PTSD, history of abuse that may have a bearing on the behaviour.
- How has the individual's behaviour affected the social environment and vice versa?
- What systematic factors contribute to or maintain the disruptive behaviour?
- What has the individual been referred for and by whom?
- Evaluate the reliability of the referral and informant sources.
- Gather information from as many sources as possible including the individual with ID and from direct observation of behaviour in different settings where possible.
- Consider the fact that transient oppositional behaviour may be common among preschool children and adolescents.

Emerson and Hatton [13] reported ODD among 13.3% of youth with ID compared with 2.3% in typically developing youths. In contrast Christensen and colleagues [17] reported among 5–9-year-old children, a point prevalence of ODD between 21% and 29% among the typically developing children compared with 35–44% among those who had borderline intellectual functioning and 39–48% among children who had ID. They noticed comorbidities with other disorders,

particularly ADHD, among children with ID. However, they did not find any intergroup differences in the rate of ODD according to gender, age of onset, or stability of the behaviour.

19.1.6 Intermittent Explosive Disorder

Intermittent Explosive Disorder (IED) is characterised by recurrent impulsive aggressive outbursts that are grossly out of proportion to any provocation or psychosocial stressor. Explosive episodes may be frequent and less severe (non-injurious) and less frequent but involving severe aggressive behaviour, such as destruction of property or physical assault with injury against an animal or a person. An important qualifier is that the aggressive outbursts are impulsive in nature and not premeditated or instrumental.

The recurrent aggressive outbursts cause marked distress or impairment in occupational or interpersonal functioning or are associated with social, financial, or legal consequences. The aggression in IED is characteristically more intense than what is observed in ADHD, CD, ODD, and antisocial and borderline personality disorders. IED should not be diagnosed in children younger than 6 years of age or in children aged 6–18 years if aggression occurs in the context of an adjustment disorder. This criterion makes this diagnosis invalid for people who have severe and profound ID, whose developmental age is likely to be lower than 6 years. This diagnosis may also be difficult to apply to those with less severe degree of ID (see notes in the next section).

IED should not be diagnosed in people with disruptive mood dysregulation disorder (DMDD), or whose impulsive aggressive outbursts are attributable to another medical condition or to the physiological effects of a substance or another mental disorder. All three DSM-5 [1] criteria (A, B, C) must be met in order to qualify for the diagnosis.

- **DSM-5 Diagnostic** *Criteria A* **[1]**

Pattern of angry/irritable mood, argumentative/defiant behaviour, or vindictiveness lasting at least 6 months, during which four or more of the following are present:

1. Often loses temper
2. Often touchy or easily annoyed
3. Often angry and resentful
4. Often argues with authority figures or, for children and adolescents, with adults
5. Often actively defies or refuses to comply with requests from authority figures or with rules
6. Often deliberately annoys others
7. Often blames others for his or her mistakes or misbehaviour
8. Spiteful or vindictive at least twice within the past 6 months

To apply this diagnosis to people with ID, the clinician must rule out any other reasons for the behaviour (see ▶ Chap. 7). The clinician also needs to carry out a language and cognitive assessment to rule out comprehension and specific cognitive difficulties that might otherwise explain the noncompliance and other associated behaviour. Assessment of both intent and developmental age is required. Consider a criterion met only if the behaviour occurs more frequently than is typically observed in individuals of comparable age and developmental level.

- **DSM-5 Diagnostic** *Criterion B* **[1]**

The disturbance in behaviour is associated with distress in the individual or others in the immediate social context. Further it causes clinically significant impairment in social, academic, or occupational functioning.

- **DSM-5 Diagnostic** *Criterion C* **[1]**

The behaviours do not occur exclusively during the course of a psychotic disorder, mood disorder, or substance use.

The onset of recurrent, problematic impulsive aggressive behaviour is most common in late childhood or adolescence [18]. The core

features of IED typically are persistent and continue for many years. The course of the disorder may be episodic, with recurring periods of impulsive aggressive outbursts. Coccaro and colleagues [19] reported a lifetime prevalence rate of 11% and a 1-month prevalence of 2.3% in a community sample of adults. These prevalence rates depend on the definition used for IED.

Certain factors make people with ID vulnerable to IED. For example, they may find it difficult to control their impulse compared with those who possess better psychological defence mechanism and stress management skills to deal with their inner frustration and anxiety. It may be difficult to be sure of the instrumental nature of behaviour in people with significant intellectual and communication impairment because the underlying intention could not be assumed. Other factors such as comorbid seizure disorders, developmental brain abnormalities, and environmental factors may all contribute to the behaviour (see ▶ Chap. 7). Some highlighted a role of life events as possible contributing factor to IED [20] among adults with ID, which may suggest a connection between explosive aggression and hyperarousal provoked by specific conditioned fear experiences in these individuals.

A multidisciplinary assessment is needed to confirm a diagnosis of IED in an individual with ID. The multidisciplinary team may include a psychiatrist, a psychologist or behaviour specialist, a speech and language therapist, a nurse, an occupational therapist, a social worker, and a neurologist where necessary. Caregivers including family members and other close informants such as teachers should also contribute to the discussion. Individual with ID should also take part in the discussion where necessary and appropriate.

A thorough physical and mental state examination is necessary to exclude any medical and psychiatric cause of aggressive behaviour. A neurologist may assist in the diagnosis and management of epilepsy and other neurological disorders which may be contributing to the aggressive behaviour [7–9] (see ▶ Chap. 7). The psychologist or behaviour specialist/nurse may help in gathering data on the nature, frequency, target, and impact (damage or injury) of aggression and carrying out a functional assessment of the behaviour. Finally, any history of abuse or trauma should be discussed in the context of identifying stimuli that may trigger an aggressive fear response.

Biological including genetic, developmental, psychosocial including environmental and family factors should be carefully assessed. The following conditions could be coexistent with or should be differentiated from IED.

- Disruptive mood dysregulation disorders (DMDD)
- Antisocial personality disorder or borderline personality disorder
- Delirium
- Major neurocognitive disorders
- Aggressive behaviour due to another medical condition
- Substance intoxication or substance withdrawal
- ADHD
- ODD
- CD
- ASD

19.1.7 Disruptive Mood Dysregulation Disorders

In DSM-5 [1] a new diagnostic category called 'Disruptive Mood Dysregulation Disorders (DMDD)' has been created under the overall 'Depressive disorder' category. This was done to categorise youths who display severe irritability and aggression chronically. DMDD is defined primarily by two features: (a) frequent severe temper outburst, and (b) persistent irritability evident every day for most of the day. Unlike the previous diagnosis of 'severe mood dysregulation', DMDD excludes hyperarousal (e.g. insomnia, agitation, distractibility, racing thoughts) from the essential criteria. Although theoretically DMDD is distinguishable from CD and ODD on the basis of severity, it is not so easy in practice to apply this distinction.

This causes confusion among the DMDD diagnosis and CD and ODD diagnoses. Therefore, some suggested that DMDD should be used as a specifier of ODD rather than a separate diagnosis [21]. Lochman and colleagues [21] also highlighted the danger of potential misdiagnosis of severe chronic irritability as a bipolar disorder which may lead to inappropriate use of medication.

19.1.8 Pyromania

A diagnosis of pyromania is made when a person repeatedly sets fires deliberately to relieve arousal or obtain pleasure from or relief of unpleasant tension upon watching the fire. The act is repetitive, deliberate, and for the purpose of relief of tension. This is not done for any other purpose or caused by any other mental disorder.

Only a small proportion of people with ID who set fires can be diagnosed with pyromania since most cases of fire setting occur for reasons that would exclude the diagnosis. The presence of ID particularly when severe and profound is likely to cause difficulty in assessing motivation and the presence of tension and relief of tension. In the case of severe or profound ID such assessments would be very difficult, if not impossible to carry out. Similarly, the observer of this act would have difficulty assessing whether there is a motive behind the act. DSM-5 [1] criteria also exclude a diagnosis of pyromania in the presence of impaired judgement which may be the case for many people with ID. Therefore, we do not think that this diagnosis could be applied to those who have severe and profound ID and who are unable to communicate their thoughts, feelings, and emotions.

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DSM-5 Diagnostic Criteria [1]

A. Deliberate and purposeful fire setting on more than one occasion.
B. Tension or affective arousal before the act.
C. Fascination with, interest in, curiosity about, or attraction to fire and its situational contexts (e.g. paraphernalia, uses, consequences).
D. Pleasure, gratification, or relief when setting fires or when witnessing or participating in their aftermath.
E. The fire setting is not done for monetary gain, as an expression of sociopolitical ideology, to conceal criminal activity, to express anger or vengeance, to improve one's living circumstances, in response to a delusion or hallucination, or as a result of impaired judgement (e.g. in major neurocognitive disorders, intellectual disabilities, substance intoxication).
F. The fire setting is not better explained by CD, a manic episode, or anti-social personality disorder.

This diagnosis has to be distinguished from abnormal rituals and interests that may be seen in a person with ASD. The clinician has to consider the difficulty in ascertaining the exact inner feelings of a person with ID.

Whereas the acts of fire setting and arson are committed by people with ID, it is not clear how many of these acts will qualify for a diagnosis of pyromania which is likely to be made of a very small subgroup of fire-setters [22–27]. Published reports identify individuals as fire setters or arsonists in a forensic setting but not in the community. It is not clear whether fire-setting is more or less prevalent among people with ID than in the general population who do not have ID. However, Jacobson [28] reported an association between the rate of fire-settings and an increasing severity of ID. Taylor and colleagues [29, 30] showed that group intervention could be effective for women with ID who set fires.

19.1.9 Kleptomania

The diagnosis requires repetitive stealing not due to the need for personal use, monetary value, anger, vengeance, or in response to delusions, hallucinations, or other mental disorders. The stealing is done to alleviate tension. It may be difficult to determine

whether these conditions are met in mild and moderate ID and it is not possible to make this diagnosis in people with severe and profound ID.

The diagnosis of kleptomania requires ascertainment of motives and the presence of tension and gratification upon stealing. Some people with ID will have difficulty describing these measures due to poor verbal skills, lack of understanding of what these measures mean, and even, inability to understand the concept of ownership and personal property.

- **A.DSM-5 Diagnostic Criteria [1]**

B. Recurrent failure to resist impulses to steal objects that are not needed for personal use or for their monetary value.
C. Increasing sense of tension immediately before committing the theft.
D. Pleasure, gratification, or relief at the time of committing the theft.
E. The stealing is not committed to express anger or vengeance and is not in response to a delusion or a hallucination.
F. The stealing is not better explained by CD, a manic episode, or antisocial personality disorder.

In case of people with ID, it is important to assess that the individual understands the sense of ownership and normal societal rules regarding possession. The difficulty of assessing inner feelings of a person with ID should be considered.

No information is available on the development and course of this disorder. The prevalence is unknown, although stealing without meeting the criteria for kleptomania is not unusual among people with ID. This may be seen as part of mania, CD, or antisocial personality disorder. Kleptomania would severely interfere with social adaptation. No research literature concerning kleptomania in people with ID could be found except a case report in the context of naltrexone treatment. Jacobson [28] reported an increased rate of property theft associated with increased severity of ID.

19.1.10 Trichotillomania

Also known as 'hair pulling disorder', involves recurrent, irresistible urge to pull hair from scalp, eyebrows, eyelids, and other areas of the body, despite repeated attempts to stop or decrease hair pulling. In DSM-5 [1], this is classified under Obsessive Compulsive and Related Disorders.

- **DSM-5 Diagnostic Criteria [1]**

A. Recurrent pulling out of one's hair, resulting in hair loss.
B. Repeated attempts to decrease or stop hair pulling.
C. The hair pulling causes clinically significant distress or impairment in social, occupational, or other important area of functioning.
D. The hair pulling or hair loss is not attributable to another medical condition (e.g. a dermatological condition).
E. The hair pulling is not better explained by the symptoms of another mental disorder (e.g. attempts to improve a perceived defect or flaw in appearance in body dysmorphic disorder).

Hair pulling can happen in people with ID, as part of a syndrome such as Down syndrome, Prader-Willi syndrome as part of the behavioural phenotype or in people with autism and sensory issues. While they would meet Criterion A, the rest of the criteria would not be met. Pulling out hair and consuming it is a cause of bowel obstruction in people with ID [31].

19.1.11 Excoriation (Skin-Picking) Disorder

- **DSM-5 Diagnostic Criteria [1]**

A. Recurrent skin picking resulting in skin lesions.
B. Repeated attempts to decrease or stop skin picking.
C. The skin picking causes clinically significant distress or impairment in social,

occupational, or other important areas of functioning.

D. The skin picking is not attributable to the physiological effects of a substance (e.g. cocaine) or another medical condition (e.g. scabies).
E. The skin picking is not better explained by symptoms of another medical condition (e.g. tactile hallucinations or delusions in a psychotic disorder, attempts to improve a perceived defect or flaw in appearance in body dysmorphic syndrome, stereotypies in stereotypic movement disorder, or intention to harm oneself in non-suicidal self-injury).

As with hair pulling, skin picking is not uncommon, particularly in people with ID and autism and people with behavioural phenotypes such as Smith Magenis syndrome.

19.1.12 Gambling Disorder

- **DSM-5 Criteria**

A. Persistent and recurring problematic gambling behaviour leading to clinically significant impairment or distress as indicated by individual exhibiting four or more of the following in a 12-month period:
   1. Needs to gamble with increasing amounts of money in order to achieve the desired excitement.
   2. Is restless or irritable when attempting to cut down or stop gambling.
   3. Has made unsuccessful efforts to control, cut back, or stop gambling.
   4. Is often preoccupied with gambling (e.g. having persistent thoughts of reliving past gambling experiences, handicapping or planning the next venture, thinking of ways to get money with which to gamble).
   5. Often gambles when feeling distressed (e.g. helpless, guilty, anxious, depressed).
   6. After losing money gambling, often returns another day to get even (chasing one's losses)
   7. Lies to conceal the extent of involvement with gambling.
   8. Has jeopardised or lost a significant relationship, job or educational or career opportunity because of gambling.
   9. Relies on others to provide money to relieve desperate financial situations caused by gambling.
B. The gambling behaviour is not better explained by a manic episode.

Gambling disorder can be episodic or persistent and can be mild (4–5 criteria met), moderate [7, 10], or severe [11, 12].

The prevalence of gambling disorder in people with ID is not known. People with more severe degrees of ID will be supervised and may not have access to finances to gamble. It may be fair to say that there is a complex interplay of factors in people with ID. Tromans and colleagues [32] describe the association of temporal lobe epilepsy, ASD, and gambling disorder in a man with mild ID.

19.2 Secondary Impulse Control Disorders

19.2.1 Hoarding Disorder

Hoarding disorder affects both men and women. Epidemiological studies report significantly higher prevalence among men which contrasts clinical samples where women predominate. Community surveys estimate point prevalence of clinically significant hoarding to be approximately 2–6% [1]. Hoarding symptoms are three times more common in older adults, aged 55–94 years compared with younger adults.

Hoarding begins early in life, starts interfering with everyday functioning by mid-twenties and significant impairment by mid-thirties. Individuals have persistent difficulties discarding or parting with possessions, regardless of their actual value. The main reasons given are the perceived utility or the aes-

thetic value of the item or the strong sentimental attachment to the possessions. Commonly saved items include newspapers, magazines, old clothing, books, bags, mail, and paperwork, but virtually any item can be saved. Individuals experience distress when faced with the prospect of discarding them which emphasises the intentional aspect of hoarding. They accumulate large number of items which fill up and clutter living areas so that they are no longer habitable so much so that the individual may not be able to sleep on their bed or cook in their kitchen. Third parties such as family members, local authorities may have to intervene to remove clutter. Symptoms such as discarding items must cause clinically significant distress. People with hoarding disorder may show features of indecisiveness, perfectionism, avoidance, procrastination, difficulty in planning and organisation, and distractibility. Half of these people have a relative with the condition.

DSM-5 Diagnostic Criteria [1]

A. Persistent difficulty discarding or parting with possessions, regardless of their actual value.
B. This difficulty is due to a perceived need to save the items and to distress associated with discarding them.
C. The difficulty discarding possessions results in the accumulation of possessions that congest and clutter active living areas and subsequently compromises their intended use. If living areas are uncluttered, it is because of the interventions of third parties (e.g. family members, cleaners, authorities).
D. The hoarding causes clinically significant distress or impairment in social, occupational, or other important areas of functioning (including maintaining a safe environment for self and others).
E. The hoarding is not attributable to another medical condition (e.g. brain injury, cerebrovascular disease, Prader-Willi syndrome).
F. The hoarding is not better explained by the symptoms of another mental disorder (e.g. obsessions in obsessive compulsive disorder, decreased energy in major depressive disorder, delusions in schizophrenia or another psychotic disorder, cognitive deficits in major neurocognitive disorder, restricted interests in ASD).

Specify if: *with excessive acquisition:* if difficulty discarding possession is accompanied by excessive acquisition of items that are not needed or for which there is no available space.

Specify if:

with good or fair insight: The individual recognises that hoarding related beliefs and behaviours (pertaining to difficulty discarding items, clutter, or excessive acquisition) are problematic.

with poor insight: The individual is mostly convinced that hoarding-related beliefs and behaviours (pertaining to difficulty discarding items, clutter, or excessive acquisition) are not problematic despite evidence to the contrary.

with absent insight/delusional beliefs: The individual is completely convinced that hoarding related beliefs and behaviours (pertaining to difficulty discarding items, clutter, or excessive acquisition) are not problematic despite evidence to the contrary.

Although ID itself is an exclusion in the diagnosis of hoarding disorder, this behaviour occurs in people with ID. Testa and colleagues [33] found that among 61 children with ID, 16.4% exhibited hoarding as a major clinical issue. When the children with ID and hoarding were compared with age-, sex-, and IQ-matched ID participants without hoarding, hoarders exhibited a slower learning curve on word list-learning task.

Diogenes syndrome is characterised by marked self-neglect, domestic squalor, social withdrawal, and hoarding of rubbish and is named after Greek philosopher Diogenes, known for his pursuit of self-sufficiency, freedom from material possessions, and disregard for social niceties and creature comforts. Williams and colleagues [34] describe two women with mild ID who fit the criteria for Diogenes syndrome and discuss the possible differential diagnoses. Hoarding was a significant feature in both of them. One of them made a significant improvement when she returned to the care of her mother and the hoarding stopped completely but the situation continued with the second one.

Hoarding is part of the behavioural phenotype of Prader-Willi syndrome. Dykens and colleagues [35] examined the nature, severity, and correlates of non-food obsessions and compulsions in 91 people with Prader-Willi syndrome and found that 47% showed hoarding behaviour. Participants hoarded a variety of objects such as old batteries, trash, papers, pens, and toiletries. Respondents often noted that some objects were collected and stored in a specific manner, for example books or toys in a series had to be purchased and then arranged in sequential order. Many care givers noted the need for subjects to sort and arrange toys, clothing, or papers according to specific rules based on size, shape, colour, or simply until they were 'just right'. As repetitive behaviours occur in 86% of people with Williams syndrome, Royston and colleagues [36] examined the profile of behaviours, comparing them with those with Prader-Willi and Down syndromes. They administered the Repetitive Behaviour Questionnaire to care givers of adults with Williams (n = 96), Prader-Willi (n = 103), and Down (n = 78) syndromes, respectively. There were few group differences, although participants with Williams syndrome were more likely to show body stereotypies. Individuals with Williams syndrome also showed more hoarding and less tidying behaviours than those with Down syndrome.

Lane and colleagues [37] suggest that hoarding can be complicated for individuals with developmental disabilities living in congregate settings because it can interfere with staff and peer relationships and affect potential community placement. Berry and Schell [38] evaluated the effects of individualised reinforcement and item return procedures on hoarding behaviour in a multiple baseline across three persons with severe ID. Systematic preference assessment procedures identified items used in the individualised reinforcement procedures. There was a reduction in hoarding behaviour for each person when individualised treatment and where item return procedures were applied, and these reductions were maintained when direct support staff were trained to provide treatment. Kellet and colleagues [39] provided 12 cognitive behaviour therapy (CBT) sessions to 14 people with mild ID on domiciliary visits at their residential facilities in the community and followed them up 6 months later. They found no evidence of relapse or adverse events and suggest from this open label trial that CBT may be a safe intervention for people with ID who exhibit hoarding behaviour.

19.2.2 Compulsive Sexual Behaviour Disorders

Compulsive sexual behaviour disorder has been proposed for inclusion as an impulse control disorder in the ICD-11 [40]. It is characterised by a persistent pattern of failure to control intense, repetitive sexual impulses or urges, resulting in repetitive sexual behaviour over an extended period (e.g. six months or more) that causes marked distress or impairment in personal, family, social, educational, occupational, or other important areas of functioning [41].

The pattern is manifested in one or more of the following:

(a) Engaging in repetitive sexual activities has become a central focus of the person's life to the point of neglecting health and personal care or other interests, activities, and responsibilities.
(b) The person has made numerous unsuccessful efforts to control or significantly reduce repetitive sexual behaviour.
(c) The person continues to engage in repetitive sexual behaviour despite adverse consequences (e.g. repeated relationship disruption, occupational consequences, negative impact on health).
(d) The person continues to engage in repetitive sexual behaviour even when she or he derives little or no satisfaction from it [41].

Compulsive sexual disorder or sexual addiction is not recognised in the latest DSM-5 [1] classification system. It was previously included in the DSM-III-TR [42] as a descriptor under the general diagnosis of 'Sexual Disorder Not Otherwise Specified'.

Pang and Masiran [43] described a 22-year-old man referred from the genitourinary clinic where he had been treated for gonococcal urethritis. He experienced high libido from the age of 19 years, admitted to having sexual intercourse with more than one hundred different commercial sex workers in the last three years, with increase in frequency to gain satisfaction, having to widen his search of area to find sex workers, having to spend huge amounts of money on this activity which he financed by working as a shop assistant and by borrowing from relatives. There were withdrawal symptoms from abstinence and the sexual activity was carried out to the exclusion of all other activities. He has been physically abused by his father as a child. His academic functioning at school was poor, having failed examinations. He could not handle money or carry out age appropriate calculations. He is reported as meeting the criteria for compulsive sexual behaviour disorder in the context of mild ID and was treated by addiction services. Psychological work reduced his craving significantly.

Singh and Coffey [44] reported a case of a young man with mild ID, autism, OCD, and relapse of bipolar affective disorder who had developed ulcers on his penis from excessive masturbation. While an inpatient, he was treated with lithium and as his mood stabilised, so did the sexual behaviour. Nishimura and colleagues [45] also described a young woman with ID who started masturbating in public, with no other features of a mood disorder who did not respond to antipsychotics but improved with lithium treatment. Lamy and colleagues [46] describe a woman with ID who had herself been experienced sexual and physical abuse. She then perpetrated sexual abuse on her children.

Thom and colleagues [47] in their review suggested that the prevalence of inappropriate sexual behaviour in people with ID can be as high as 15–33%, but the nature tends to be inappropriate than violative. It may be associated with ID syndromes. For example, Fisher and colleagues [48] found a complex association between Klinefelter syndrome and hypersexuality, paraphilic behaviours, and gender dysphoria, which were mediated by obsessive compulsive and autistic traits. Börjeson-Forssman-Lehmann syndrome (BFLs) is an X-linked inherited disorder characterised by unusual facial features, abnormal fat distribution, and ID. Behaviour in persons with BFLs is in general friendly but can be challenging with externalising and thrill-seeking features but their social skills are good. de Winter and colleagues [49] described a group of four BFLs patients with inappropriate sexual behaviour as part of their behavioural phenotype.

Finally, as Sajith and colleague [50] point out, men with ID sometimes exhibit inappropriate sexual behaviours, which may be termed challenging behaviours or offending behaviours depending on the circumstances and whether criminal justice system is involved. Hawk and colleagues [51] have shown that the rate of sex offence charges was nearly twice as high among defendants with ID as among defendants without ID.

In conclusion, compulsive sexual behaviours can happen in people with ID for various reasons; as part of affective disorder, behavioural phenotype, due to environmental reasons, or as an entity in itself. However, it is important to explore the knowledge of the person with ID of sexual function, sexual relationship, and social norms before assuming that any socially inappropriate sexual acts on their part including sex offences are intentional.

19.3 Body-Focussed Repetitive Behaviour Disorder

19.3.1 Stereotypic Movement Disorder

- **DSM-5 Diagnostic Criteria [1]**

A. Repetitive, seemingly driven, and apparently purposeless motor behaviour (e.g. hand shaking or waving, body rocking, head banging, self-biting, hitting own body).
B. The repetitive motor behaviour interferes with social, academic, or other activities and may result in self-injury.

C. Onset is in the early developmental period.
D. The repetitive motor behaviour is not attributable to the physiological effects of a substance or neurological condition and is not better explained by another neurodevelopmental or mental disorder (e.g. trichotillomania, OCD).

- **Specify if:**

With self-injurious behaviour or behaviours that would result in an injury if preventive measures were not used.

Without self-injurious behaviour.

- **Specify if:**

Associated with a known medical or genetic condition, neurodevelopmental disorder, or environmental factor (e.g. Lesch-Nyhan syndrome, ID, intrauterine alcohol exposure).

Mild, if symptoms can be suppressed by sensory stimulus or distraction; moderate, if explicit protective measures and behaviour modifications are required; and severe if continuous monitoring and protective measures are required to prevent self-injury.

Essential features of stereotypic movement disorder are that they are repetitive, seemingly driven, and apparently purposeless [1]. They are often rhythmical movements of head, hand, or body without obvious adaptive function and may not respond to effort to stop them. These may be seen in typically developing children, but they can be stopped when attention is drawn to them or when the child is distracted. The child may also demonstrate self-restraining behaviour, that is sit on their hands.

In typically developing infants, there is a predictable sequence of rhythmic motor development, which appears stereotypic and declines by 24 months of age [52]. This stereotypic, rhythmic motor behaviour may appear at later ages in children with Down syndrome and those with significant motor impairment [53, 54]. Moreover, children with significant developmental disabilities engage in relatively high rates of stereotypies as compared with typically developing infants and toddlers [54, 55]. Hoch and colleagues [56] compared two groups of young children, one with developmental delay and one without, and asked parents to complete standardised measures on the presence of stereotypies and self-injury. They found that stereotyped behaviour was reported significantly more frequently by parents of children with developmental delay than by parents of typically developing children (85.7% vs. 42.8%). Over half of the children with delay (53%) were described as having at least one stereotypy that was considered a moderate or severe problem in comparison with only 2% of the children without developmental delay on a standardised measure of repetitive behaviour.

Stereotypic behaviour is variable as each individual presents with her or his own signature behaviour. Examples of non-self-injurious stereotyped behaviour include body rocking, hands flapping, finger flicking, and head nodding. Examples of self-injurious stereotypic behaviour can be repeated head banging, face slapping, eye poking, and biting parts of the body. Frequency of occurrence of these behaviours can vary and be related to stress, excitement, boredom, fatigue, and multitude of factors specific to the person. In both humans and nonhuman primates, aberrant stereotyped behaviour including self-injury has been associated with adverse environmental circumstances and early CNS damage [57].

Stereotypic behaviours occur in a variety of neurodevelopmental conditions, particularly in people with ID and ASD. Bodfish and colleagues [58] found a prevalence of stereotypy of 60.9% and self-injury 46.6%; and compulsion when they surveyed people with severe and profound ID. Rojahn and colleagues [59] found 54% of 432 randomly selected residents from a developmental centre had stereotyped behaviour and 43%, self-injury. The determination of the functional profile of a targeted behaviour has important implications for the design of customised behavioural interventions. Medeiros and colleagues [60] investigated the relationship between the level of ID and the functional profile of aggression, stereotypy, and self-injurious behaviour. They found that individuals with mild ID tended to use self-injurious

behaviour less often for tangible rewards or to escape social demands than individuals with severe ID who tended to use them for these same purposes significantly more often. This could be helpful in devising interventions, together with language and communication training which may help people with severe ID express themselves about when they wanted a break from social demands. Research also showed that aggressive and stereotypic behaviour problems can function, regardless of the severity of ID, for the purposes of attention, sensory stimulation, pain reduction, social escape, and tangible reinforcement. A functional assessment is, therefore, an essential part of the assessment process (see Assessment section in the ► Chap. 7).

Despite their core significance in ASD, it is not clear whether there are distinct groups of stereotypic behaviours with different specificity to autism. Carcani-Rathwell and colleagues [61] found that subjects with a Pervasive Developmental Disorder (PDD) diagnosis were more likely to have repetitive stereotyped behaviours than those with only ID. Within the PDD group, those with ID had higher rates of all repetitive stereotyped behaviours. The study suggests that there may be two distinct sub-groups of repetitive behaviours whereby the sensory and motor repetitive behaviours are generally associated with lower developmental age and less specific to the autistic syndrome whereas the 'higher-order' behaviours, that is cognitive rigidity symptoms, may be a more 'autism specific' feature. Bodfish and colleagues [62] compared adults with autism with adults with ID and found that both groups had significant patterns of repetitive behaviour co-occurrence. Autistic subjects had significantly greater severity ratings for compulsions, stereotypy, and self-injury. Repetitive behaviour severity also predicted severity of autism. Pedersen and colleagues [63] found that restricted interests or repetitive behaviours significantly differentiated between children with autism and ID.

Behavioural features of children with severe ID remain difficult to disentangle from those of children with autism. Polyak and colleagues [64] suggest that current estimates of autism prevalence fail to take into account the impact of comorbidity of ID and epilepsy on autism diagnosis.

Self-injury also co-occurs in a number of syndromes associated with ID including Lesch-Nyhan, Smith Magenis, Rett, Cornelia de-Lange, Prader-Willi, and fragile X syndrome [10]. Disruption to the activity dependent neuroprotector homeobox (ADNP) gene is among the most common heterozygous gene mutation seen associated with ASD. Affected individuals have a constellation of features including autism, ID, dysmorphic features, and hypotonia. Arnett and colleagues [65] compared individuals with ADNP syndrome with those with autism due to CHD8 mutation and idiopathic autism and found that ADNP syndrome group's profile of social communication and restricted and repetitive behaviours deficits to be notably different from those of other autism subtypes. The youths with ADNP syndrome had relatively mild social communication deficits (despite impaired verbal intelligence and low expressive language abilities) coupled with stereotyped motor restricted repetitive behaviours. The social communication deficits were mild enough as to not to warrant a diagnosis of DSM-5 [1] ASD in about 30% of cases, although all of them had a diagnosis of ID. The aetiology of social communication and repetitive stereotyped behaviours in autism is not well understood. Arnett and colleagues [65] suggest that genetic subtypes of ASD may each be associated with a unique aetiology of autism trait covariance. Within the idiopathic group, it could be polygenic. However, within single gene disorder, individual differences in the location and effect of the genetic variant could have vastly different effects on protein functioning. Yet, these disparate events frequently produce a similar behavioural, physical, and/or medical phenotype, implying convergence to a common neurobiological endophenotype.

Haw and colleagues [66] carried out a study on the movement disorders seen in a cohort of 145 people with Down syndrome living both inpatient and community facilities in Surrey, UK. They found that over a third of these people experienced stereotypies and no association was found between stereotypies

and age, hospital or community placement, exposure to neuroleptics, behavioural disorder, or dementia.

For treatment of all the conditions described in ► Chaps. 11 and 12.

Key Points

- The DSM-5 criteria are often difficult to apply to people with intellectual disabilities for conduct disorder, intermittent explosive disorder, and oppositional defiant disorder.
- Therefore, instead of making a psychiatric diagnosis as per DSM-5 criteria, it is more important to describe the behaviour for a better formulation leading to a better intervention.
- Both pharmacological and non-pharmacological interventions are used but the evidence in their support is poor.
- It is, therefore, important to make a very thorough multidisciplinary assessment of these behaviours in order to achieve a successful treatment outcome.
- The ultimate goal for the intervention is to improve the quality of life of people with intellectual disabilities and their care givers.

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Psychotic Disorders

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Contents

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Learning Objectives

- To learn about the definition and ascertainment of psychotic disorders in people with intellectual disabilities (ID) across the lifespan
- To become aware of differential diagnoses in people with ID
- To understand the issues around diagnosis and management at different levels of intellectual functioning
- To list treatment options and approaches to psychotic disorders in this population

20.1 Introduction

Psychosis is a severe mental disorder defined by its chronic course and the significant levels of disability that it is associated with. It is found in all parts of the world with an often reported prevalence of approximately 1% whilst incidence (median) is estimated to be 15.2/100.000 persons with a ratio for males:females of 1.4:1 [1].

The lifetime risk for psychosis is almost half of the incidence (7.2/100.000). People with psychosis have almost threefold increase in all-cause mortality, and multiple factors are implicated in its onset including urbanicity, migrant status, socio-economic status, genetic predisposition, etc. Psychotic disorders place a significant burden on individuals, their families and societies; most of the economic impact is due to use of health resources, inpatient and social care, homelessness, legal issues, and loss of productivity (these are also called direct and indirect costs). A study by Cloutier et al. [2] indicated that the annual cost was estimated at $155.7 billion at 2013 prices. The three main reasons driving expenditure were unemployment, caregiver loss of productivity, and healthcare costs.

Psychosis is a serious mental disorder with a chronic course in most cases and consequent high level of disability. It is also associated with increase in all-cause mortality, high costs for health care, low economic and decision-making autonomy as well as a considerable burden on caregivers.

20.2 Psychosis and Intellectual Disabilities

Clouston, Murray, and Bramon [3] were the first to put forward the theory of psychosis as a neurodevelopmental disorder. Kraepelin described an entity he termed 'dementia praecox' which was associated with poor prognosis and recent theorists suggest that potentially both a neurodevelopmental process and neurodegeneration may be implicated. This hypothesis suggests abnormalities have taken place in the brain early on with consequent impact in functioning including in social development [4]. However, neither hypothesis fully explains psychotic disorders with the possibility of endophenotypes existing with different aetiological background [5].

Further, recent advances in genetics suggest that genomic copy number variants may not only increase the risk of psychosis but are also associated with also associated with intellectual disability (ID), autism spectrum disorder (ASD), attention-deficit/hyperactivity disorder, and epilepsy [6].

The causes of psychotic disorders are multiple and include neurodegenerative, neurodevelopmental, and genetic hypotheses which are supported by some evidence.

20.3 Diagnostic Criteria and Clinical Presentation

Psychosis or schizophrenia spectrum disorders are non-specific terms which include a number of conditions with both a narrow and a broader definition. These conditions may vary in duration, intensity, and course mainly clustered around clinical signs and symptoms.

Two classification manuals (the Diagnostic and Statistical Manual of Mental Disorders, DSM-5 [7] and the International Classification of Diseases, ICD-10 [8]) are mainly in use but both have adaptations for people with ID (DM-ID-2; NADD 2017 [9] and DC-LD; Royal College of Psychiatrists 2001 [10], respectively). DC-LD specifically addresses the challenges of diagnosing mental conditions in people with more severe ID. For

example, it suggests that that early signs of psychosis could be new behaviours, especially if odd/bizarre/uncharacteristic for that person or increased frequency/severity of pre-existing behaviour. It places less significance to negative symptoms than other diagnostic criteria.

In 2019, member states of the World Health Organization agreed to adopt the new (11th) revision of ICD (ICD-11; WHO 2019 [11]), whose long-text version has come out at the beginning of 2022 [12]. ICD-11 new diagnostic categories are included in ▶ Box 20.1, for comparison with the former ICD-10 and DSM-5.

Whilst ICD-10 includes a number of diagnostic sub-categories (see ▶ Box 20.2 from Sheehan, Fodor-Wynne, and Hassiotis [13]), DSM-5 does not as specific sub-types appear to have limited diagnostic stability, low reliability, and poor validity; there has been an attempt to define disorders along a spectrum with a gradient of symptoms and continuum of severity. Generally, application of diagnostic criteria appropriate to peers of average intelligence has been shown to be restrictive and may lead to the under-diagnosis of serious and treatable mental disorders in children and adults with ID. The prevalence rates of psychotic disorders according to the DSM-IV-TR [14] have been found to be lower than prevalence according to the DC-LD, suggesting that DSM is not as effective in diagnosing psychotic disorders in this patient group [15]. Usually, the criteria have to be applied in consultations with qualified practitioners who are expert in the assessment and diagnosis of people with ID.

The core symptoms of psychotic disorders – also called first rank symptoms – [17] include:

- Auditory hallucinations (indistinguishable from a normal percept but cannot be corroborated by experience)
- Thought echo (*Gedankenlautwerden*; an auditory hallucination in which the patient hears his/her thoughts spoken aloud)
- Passivity experiences (commonly delusions of being controlled by an external force or agency)
- Thought withdrawal (a belief that the patient's thoughts have been 'taken out' of the patient's mind)
- Thought insertion (thoughts, ascribed to other people, intruding into the patient's mind)
- Thought broadcasting (patient's own thoughts shared with others)
- Delusional perception (linking a normal sensory perception to a bizarre conclusion, for example, seeing an aeroplane means the patient is the president).

Delusions are fixed, false beliefs that will not be changed when patients are exposed to new, conflicting evidence. These beliefs can include a variety of themes and specifiers (e.g. grandiose, religious, persecutory). Hallucinations are sensory experiences that do not have an external stimulus. Formal thought disorder is generally observed through disorganized speech, thought blocking, derailment. For example, a patient might rapidly change topic when speaking and when asked a question, and their answer may be unrelated. This category of symptoms also includes 'catatonic behaviour' which is characterized by a marked decrease in reactivity to the environment, for example, resistance to instruction, rigid posture, lack of verbal and motor response.

Grossly disorganized or abnormal motor behaviour can manifest as unprovoked agitation, child-like behaviour, and reacting inappropriately to the environment (e.g. laughing inappropriately).

Negative symptoms are characterized by a withdrawal or lack of normal function. Two common negative symptoms are diminished emotional expression and avolition (reduction in drive to fulfill goals and activities).

Some of these core symptoms can present also in other psychiatric disorders. Catatonia in particular can occur in neurodevelopmental, affective, and organic mental disorders, or represent an unspecified diagnostic category when associated with symptoms that cannot be attributed to any mental disorder or other medical condition, as in the DSM-5 [7]. In ICD-11, catatonia is defined as 'a syndrome of primarily psychomotor disturbances that is characterized by the simultaneous occurrence of several symptoms such as stupor; catalepsy; waxy flexibility; mutism; negativism; posturing; mannerisms; stereotypies; psycho-

motor agitation; grimacing; echolalia and echopraxia', and has been split into four types based on the following characteristics: (a) occurrence in the context of specific mental disorders, including schizophrenia or other primary psychotic disorders, mood disorders, and neurodevelopmental disorders; (b) being induced by psychoactive substances, including medications; (c) being caused by a medical condition not classified under mental, behavioural, or neurodevelopmental disorders; and (d) being not otherwise specified.

The diagnosis of psychotic disorders in people with ID may be confounded by many other factors related to symptom identification and interpretation. Communication difficulties are one of the most common confounders and as yet there are no biomarkers to confirm the diagnosis of a psychotic disorder. Therefore, clinical skill is important in the ascertainment of psychosis with good reliability achieved between experienced clinicians [18]. A systematic review of the clinical presentation of psychosis in people with ID ($n = 280$) found that participants in the five included studies had an IQ range 50–81 with higher scores on scales of negative features (effect size of 1.72). Other studies indicate that people with ID show an excess of formal thought disorder and limited delusional content [19].

Psychotic disorders are characterized by a wide variety of conditions that share a cluster of clinical signs and symptoms (traditionally defined as *first-rank*) including auditory hallucinations, passivity experience, thought insertion, thought broadcasting, thought withdrawal, and delusion, often associated with disorganized behaviour and negative symptoms. It can be difficult to identify these symptoms in people with ID and/or low-functioning ASD (LF-ASD), especially in the case of severe communication difficulties, thus requiring good diagnostic skills from clinicians. The diagnostic manuals currently available for use with ID and LF-ASD are DM-ID-2 and DC-LD, which represent adaptation of the DSM-5 and ICD-10, respectively.

Box 20.1: Comparative Schema of Classification of Schizophrenia Spectrum Between ICD-10, ICD-11, and DSM-5

ICD-10		ICD-11		DSM-5
Schizophrenia, schizotypal and delusional disorders		Schizophrenia or other primary psychotic disorders		Schizophrenia and other psychotic disorders
F20 Schizophrenia		6A20 Schizophrenia		295.90 Schizophrenia
	F20.0 Paranoid schizophrenia		6A20.0 Schizophrenia, first episode	
	F20.1 Hebephrenic schizophrenia		6A20.1 Schizophrenia, multiple episodes	
	F20.2 Catatonic schizophrenia		6A20.2 Schizophrenia, continuous	

ICD-10		ICD-11		DSM-5
	F20.3 Undifferentiated schizophrenia		6A20.Y Other specified schizophrenia	
	F20.4 Post-schizophrenic depression		6A20.Z Schizophrenia unspecified	
	F20.5 Residual schizophrenia			
	F20.8 Other schizophrenia			
	F20.9 Schizophrenia, unspecified			
F21 Schizotypal disorder		6A22 Schizotypal disorder		Schizotypal (personality) disorder
F22 Persistent delusional disorder		6A24 Delusional disorder		297.1 Delusional disorder
	F22.0 Delusional disorder		6A24.0 Delusional disorder, currently symptomatic	
	F22.8 Other persistent delusional disorders		6A24.1 Delusional disorder, in partial remission	
	F22.9 Persistent delusional disorder, unspecified		6A24.2 Delusional disorder, in full remission	
			6A24.Z Delusional disorder, unspecified	
F23 Acute and transient psychotic disorders		6A23 Acute and transient psychotic disorders		298.8 Brief psychotic disorder
	F23.0 Acute polymorphic psychotic disorder without symptoms of schizophrenia			

ICD-10		ICD-11		DSM-5
	F23.1 Acute polymorphic psychotic disorder with symptoms of schizophrenia			
	F23.2 Acute schizophrenia-like psychotic disorder			
	F23.3 Other acute predominantly delusional psychotic disorders			
	F23.8 Other acute and transient psychotic disorders			
	F23.9 Acute and transient psychotic disorder, unspecified			
F24 Induced delusional disorder				
F25 Schizoaffective disorders		6A21 Schizoaffective disorders		295.70 Schizoaffective disorder
	F25.0 Schizoaffective disorder, manic type		6A21.0 Schizoaffective disorders, first episode	
	F25.1 Schizoaffective disorder, depressive type		6A21.1 Schizoaffective disorders, multiple episodes	
	F25.2 Schizoaffective disorder, mixed type		6A21.2 Schizoaffective disorders, continuous	
	F25.8 Other schizoaffective disorders		6A21.Y Other specified schizoaffective disorders	
	F25.9 Schizoaffective disorder, unspecified		6A21.Z Schizoaffective disorders, unspecified	

ICD-10		ICD-11		DSM-5
F28 Other nonorganic psychotic disorders		6A2Y Other specified schizophrenia or other primary psychotic disorders		298.8 Other specified schizophrenia spectrum and other psychotic disorder
F29 Unspecified nonorganic psychosis		6A2Z Schizophrenia or other primary psychotic disorders, unspecified		298.9 Unspecified schizophrenia spectrum and other psychotic disorder
				Substance/medication-induced psychotic disorder
				Psychotic disorder due to another medical condition

Box 20.2: Schizophrenia Spectrum Disorders Defined in ICD-10

1. Schizophrenia
   Manifests as a disorder of significantly distorted thinking, perception and affect. Main symptoms include thought disorder, delusions, hallucinations, and negative symptoms. Cognitive deficits develop over time. Subtypes have been delineated by course and major features.
   - Paranoid schizophrenia
     - *The most common subtype. Paranoid or persecutory delusions and auditory hallucinations dominate.*
   - Hebephrenic schizophrenia
     - *Delusions and hallucinations are less prominent. Behaviour is grossly disorganised and emotional response is lacking or inappropriate.*
   - Catatonic schizophrenia
     - *Bizarre motor manifestations are the hallmark of catatonia. These include, but are not limited to, posturing, waxy flexibility, stupor, and mutism.*
   - Residual (and simple) schizophrenia
     - *A slow but progressive decline in functioning accompanies social withdrawal and the appearance and deepening of negative symptoms.*
2. Schizotypal disorder
   Sometimes considered a form of personality disorder, schizotypal disorder is characterised by social and interpersonal deficits. There may be suspiciousness, odd beliefs, eccentric behaviour and unusual perceptual experiences, though no single feature predominates. The disorder runs a chronic course.
3. Persistent delusional disorder
   Chronic, frequently lifelong delusions occur in the absence of other psychotic symptoms.
4. Acute and transient psychotic disorder

Psychotic symptoms develop rapidly and are not due to intoxication or an organic condition.

There is an equally rapid and usually spontaneous recovery.

5. Induced delusional disorder
Occurs when a delusional belief, and sometimes other psychotic symptoms, are shared by two or more people with close emotional links. The sufferers are typically socially or physically isolated from others. Symptoms resolve in at least one of the sufferers following geographical separation. The illness is also known by the French term folie à deux, literally 'madness of two' [16].

20.4 Incidence and Prevalence

Cooper et al. [20], found that in a Scottish epidemiological sample, the 2-year incidence of psychosis was 1.4% and for first episode of psychosis 0.5%. One of the earliest studies [21] found that incidence was higher in people with mild compared to severe ID but this may be due to inability to make a diagnosis in the latter.

A recent systematic review and meta-analysis of 25 cross-sectional studies estimated the prevalence for psychosis in people with ID to be 3.46% and the authors observed a variation in reported rates between those with mild compared to people with more severe ID (5.55% in mild vs 0.88%, respectively) [22]. Prisoners with ID are also shown to be twice as likely to suffer with psychosis compared to peers without ID [23].

20.5 Issues Related to Diagnosis in Persons with ID

Individuals with a dual diagnosis of ID and psychosis are significantly younger at first contact with mental health services compared with individuals with schizophrenia alone although the reasons for this are unclear [24]. Given the impact of untreated psychosis on the disease course, it is essential for family and paid carers to be aware of relevant symptoms that should be brought to the attention of professionals. One such example is the case of distinguishing true hallucinations from pseudohallucinations in people with ID [25]. Pseudohallucinations are defined as hallucinations where the individual is aware that their experience is in their own mind and not perceived as external stimuli. This distinction is important in terms of the therapeutic approach and advice for future management. Such diagnostic conundrums are particularly important in the diagnosis of mental disorders in people with severe ID and lack of verbal ability.

Higher levels of aggression have been found in people with ID and psychosis when compared to individuals with ID without psychosis possibly underpinned by 'impulse control, mood dysregulation and perceived threat' [26].

Bradley et al. [27] explored the association between ID and psychiatric disorders, including psychosis. The authors suggest that the symptoms of ID may sometimes be overlooked during psychiatric assessments which, they argue, may lead to inaccurate diagnosis and incorrect treatment. They argue that patients are not regularly reviewed and a patient may have a diagnosis for years when another diagnosis may better suit their symptoms. They suggest that medication is overused in this patient group and that more emphasis should be given to psychosocial therapies and good quality care. Axmon et al. [28] found that the prescription of antipsychotic medication without thorough psychiatric diagnosis is common also in older persons with ID, mainly for the management of problem behaviours.

Finally, there is a danger of misdiagnosing schizophrenia when a patient presents with positive symptoms that are caused by childhood trauma or abuse and not necessarily indicative of a psychotic disorder.

Finally, Hurley and Moore [29] suggest that cases of erotomanic type delusional disorder in adults with ID may be underreported because it is assumed to be an immature fantasy that is a product of the cognitive limitations.

Frequently reported issues in diagnosing psychotic disorders in persons with ID and LF-ASD are distinguishing hallucinations from pseudohallucinations, distinguishing other positive psychotic symptoms from symptoms caused by childhood trauma or abuse, identifying aggressive behaviour as a behavioural equivalent of one or more psychotic symptoms, and recognizing erotomanic delusion in those with assumed immature fantasy.

20.6 Aetiology

The increased comorbidity between ID and psychosis may be explained by the fact that cognitive impairment is an independent risk factor and a feature of the disorder [6, 30–35]. Evidence from epidemiological studies supports a shared genetic risk for lower IQ and schizophrenia with the strength of the genetic effect increasing with decreased IQ [36].

Very likely, the same factors that have been implicated in the onset of psychosis in the general population may play a role in people with ID too and could possibly form future research strands [36, 37]. This is an important point, as people with ID are routinely excluded from research in a variety of medical fields, and therefore, conclusions drawn from those studies cannot be fully applied to this population due to the differences in health patterns.

New advances in genetics, particularly in the study of CNVs, show that they increase the risk for other neurodevelopmental disorders [38]. In a study of 39,000 people referred to a diagnostic laboratory, about 1000 had a CNV at one of the loci identified in schizophrenia. Main clinical presentation included psychiatric disorders, developmental delay, ID, and autism-related disorders. Examples of neurodevelopmental genes implicated in schizophrenia include genes that regulate cell growth and behaviour, DISC1, NRG1, DTNBP1, KCNH2, AKT1, PAK7, and RGS4 [39, 40]. Specific genetic syndromes that have been considered as examples of the biology of psychosis include the velo-cardio-facial syndrome (VCF, also known as DiGeorge syndrome) [41, 42]. VCF syndrome is a relatively common genetic disorder (14,000) associated with ID (usually mild-moderate) and psychosis develops in about 40% of cases [43].

Other genetic syndromes linked with ID and associated with psychosis include fragile X syndrome [44], Kleinfelter syndrome [45], Prader-Willi syndrome [46, 47], Dandy Walker syndrome [48, 49], Nieman Pick disease [50], Usher syndrome [51], Williams syndrome [52], and Charlexois ARSACS [53]. A recent study reported a higher rate of psychosis in adolescents and young adults with Down's syndrome [54]. Most of the studies on syndromes are single cases of sporadic morbidity and therefore further research is needed to confirm such associations.

Imaging studies are sparse in people with ID and psychosis, but available literature appears to show that the brains of people with ID and schizophrenia are more similar to the brains of those with schizophrenia in the general population than those with ID alone suggesting a common pathophysiological process at work [55].

Multiple biochemical pathways are likely to contribute to schizophrenia so identifying a single abnormality is difficult. A number of key neurotransmitters have been linked to schizophrenia including dopamine, glutamate, serotonin, noradrenaline, and GABA. Some abnormalities in these pathways are also associated with ID [56] and ASD [57].

Another possible explanation for the increased association between ID and psychosis may be derived from the vulnerabilities conferred by cognitive deficits, for example in processing information, problem solving and planning, and emotional and social deficits [58].

Finally, whilst life events [59] and stigma [60] may be associated with psychological morbidity in people with ID there is not enough evidence from research studies to hypothesis causality in the onset of psychosis. Nevertheless, it is well established that the development of psychotic disorders is associated with aversive childhood and adulthood experiences such as bullying and trauma [61] and that individuals with neurodevelopmental disorders such as ID and ASD may be at an increased risk of victimization [62–64].

However, the impact of psychosis on people with ID was studied by Bouras et al. [65] who found that the ID group showed serious impairments in social and occupational functioning (Global Assessment of Functioning scale), compared with the non-ID group. The ID group was likely to have fewer or no friends and to have problems maintaining employment. Friedlander and Donnelly [66] found that participants with a diagnosis of psychosis not otherwise specified and moderate or severe ID were the most functionally impaired group in the study, based on the Global Assessment of Functioning scale and the Clinical Global Impression.

In a recent large prospective study carried out in Sweden, 23% of the risk for non-affective psychosis associated with urban and deprived settings at birth could be attributable to indirect effects on cognition, measured by IQ at 18 years of age [67].

Genetic factors seem to play an important role in the aetiopathogenesis of psychotic disorders in persons with ID and LF-ASD as suggested by the presence of common CNV alterations in psychotic disorders and ID/ASD, and the high prevalence of psychotic symptoms in specific genetic syndromes including ID/ASD such as DiGeorge syndrome, Prader-Willi syndrome, and Fragile X syndrome. Various neurotransmitter alterations have been identified in psychotic disorders as well as in ID and ASD. A relevant psychological vulnerability factor is represented by low coping with adverse life events, stressors, and traumas.

20.7 Differential Diagnosis

As mentioned earlier, it can be difficult to diagnose psychosis in people with an ID due to many factors, such as communication problems and variable presentation of symptoms and cultural norms. There are also several other disorders that can present with symptoms that overlap with those of psychosis. While many of these disorders are separate from schizophrenia spectrum disorders, it is important to be mindful of the fact that some could be involved in the development and course of psychosis.

Differential diagnoses to psychosis are shown in ► Box 20.3 [68].

Dementia in people with ID is more common than in the general population [69, 70]. Dementia can often present with psychotic symptoms or abnormal speech [71] and so dementia may be confused with psychotic disorders in people with ID.

Box 20.3: Differential Diagnoses

Physical illnesses including: deliriums

Epilepsy (particularly temporal lobe epilepsy)

Traumatic brain injury

Intracerebral conditions (e.g. space occupying lesion)

Sleep disorders

Infection (e.g. respiratory or urinary tract infection, meningitis)

Endocrine causes (e.g. hyper/hypothyroidism)

Hearing or visual impairment (e.g. individuals with Charles Bonnet syndrome who have a visual impairment may experience hallucinations or delusions)

Causes of pain or distress (e.g. toothache, earache, constipation, menstrual pain)

Drug or alcohol related causes including:	Delirium tremens Alcoholic hallucinations Drug-induced psychosis Sensitivity to prescribed medication
Other psychiatric illness including:	Affective illness Anxiety disorder Post-traumatic stress disorder (PTSD) Dementia (e.g. visual hallucinations in Lewy-body dementia) Autistic spectrum disorder

Other differential diagnosis (e.g. challenging *behaviour* caused by ID, or just ID itself may be diagnosed as a psychotic disorder)

20.8 Comorbidity with Autism Spectrum Disorder

A great complexity underlies the association [72, 73]. ID, psychotic disorders, and ASD may be treated as separate entities but are likely to have common underlying basis [74].

There is increasing evidence on a clinically significant degree of co-occurrence between ASD and psychotic disorders [75, 76]. Non-affective psychoses in particular are reported to be tenfold or higher in persons with ASD than in the general population [77] and childhood onset psychosis has been found to co-occur with ASD in 30–50% of common family genetic imaging findings [78]. Diagnostic confusion may also arise due to symptom overlap, for example, negative symptoms, social interaction/communication difficulties and repetitive stereotyped behaviours in people with psychosis [79] especially for those psychiatrists who have not had specific training on psychopathology in adults with neurodevelopmental disorders [73].

Waris et al. [80] investigated comorbidity between schizophrenia and pervasive developmental disorder (PDD), particularly Asperger's syndrome in adolescents. They found that 44% (n = 18) of the adolescents with schizophrenia fulfilled the diagnosis of PDD in childhood (even if they did not receive this diagnosis at the time). They conclude that patients with psychosis often have a childhood diagnosis of PDD. Palucka, Bradley, and Lunsky [81] highlight the importance of diagnosing autism in individuals with ID before making a diagnosis of psychosis. Such a process could guard against patients being given a diagnosis of psychosis when autism may better fit their presentations. Larson et al. [82] studied 116 adults with autism and psychosis; they found that age of onset had a wide range (e.g. as early as below 12 years of age and in the middle years), progress was insidious in the majority and impaired functioning. Fifty-three percent of the cohort had a diagnosis of psychosis not otherwise specified or schizophrenia with 85% reporting significant affective symptoms (the symptom categories are shown below) (Fig. 20.1).

> Psychotic disorders are frequently reported in people with ASD. These disorders are characterized by a pool of overlapping symptoms, especially negative symptoms, and social interaction/communication difficulties.

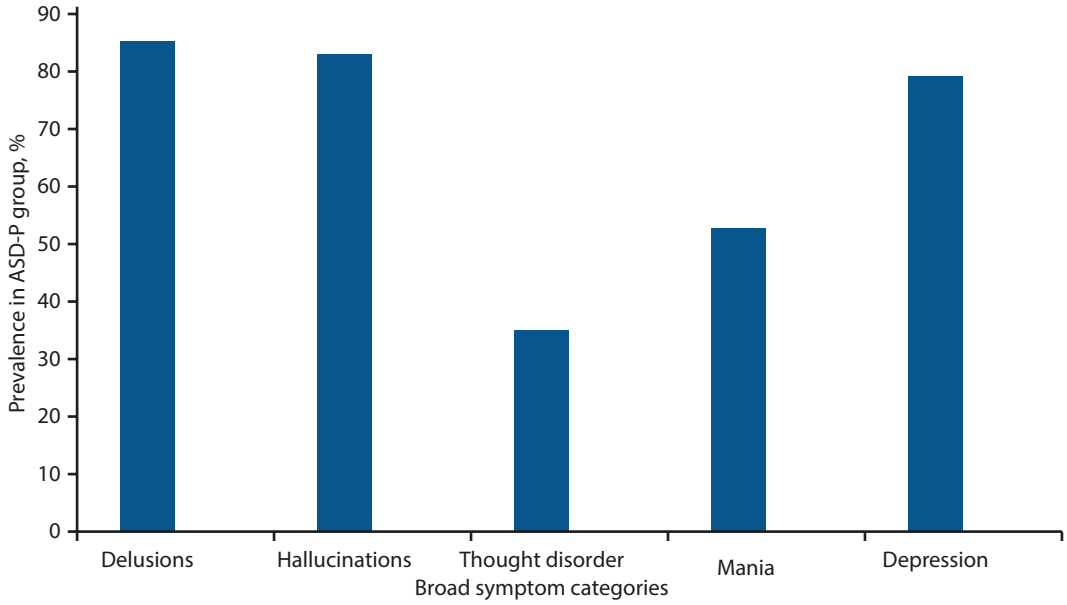

Fig. 20.1 Symptom frequencies in 116 adults with autism and psychosis [82]

20.9 Psychosis in Adults with Severe or Profound ID

Cherry et al. [83] investigated the presentation of schizophrenia in a residential sample of adults with severe ID by comparing their symptoms to symptoms that are seen in populations without ID. They concluded that the symptoms of the group scoring high on the Diagnostic Assessment for the Severely Handicapped (DASH-II) and also had a clinical diagnosis of schizophrenia were clustered around 'reality distortion and disorganization, with less evidence of negative symptoms'. However, it is possible that staff tend to report negative symptoms less as they find them less disturbing than florid symptoms. Furthermore, proxy reports of symptoms of mental disorders in people with lack of verbal ability may generally be inaccurate and further attempts to enhance individual communication is recommended [84]. It may be that interviewing several people with a good knowledge of the individual and paying close attention to changes in usual behaviour may be helpful in identifying negative symptoms that may be less salient and more difficult to identify.

Positive symptoms are generally more easily identifiable and more frequently reported in persons with severe and profound ID, probably due to the difficulties in identifying negative symptoms.

20.10 Psychosis in Children and Adolescents with ID

Psychotic disorders have an earlier onset in persons with ID than in the general population, although evidence is limited [66, 85]. The knowledge on the course of psychotic disorders in children and adolescents with ID is also restricted. Lee et al. [86] examined the development and stability of the symptoms of early-onset schizophrenia (EOS) in 10 children with ID. They found that 8 of the 10 children with a diagnosis of EOS still warranted this diagnosis 2 years later but delusions and visual hallucinations were less prominent. These children had a poor prognosis with particular impact of the disorder on cognitive ability. Of the two children not determined to have EOS at follow-up, one recovered fully and the other suffered from hypothyroidism and Tourette's syndrome. The authors argue that it is important to always gather collateral information and review symptoms at regular intervals.

A Canadian study found that three factors may improve prognosis in youths with ID and psychosis: low rates of alcohol and substance misuse, increased compliance with medication, and good psychosocial supports [66]. More longitudinal data is needed to further substantiate our understanding of outcomes in early onset psychosis in young people with ID.

Psychotic disorders seem to have an earlier onset in persons with ID than in the general population. The knowledge on course and prognosis is limited, although low rates of alcohol and substance misuse, increased compliance with medication, and good psychosocial supports have been associated with better outcomes in youths with ID.

20.11 Principles of Treatment and Management

It is evident that psychosis is a long term condition and a major cause of impairment in functioning. Prompt diagnosis and treatment are essential in managing the consequences on the individual and his/her family and social networks. Generally, medication (antipsychotics alone or in combination with mood stabilizers) and psychosocial interventions to combat persistent symptoms, lack of motivation, relapse, unemployment, and loneliness are likely to achieve best results.

Patients who are acutely unwell may require home treatment of admission to an inpatient facility. There are now fewer specialist inpatient units for people with ID and mental disorders, and therefore, it is essential that the community ID services advocate for

the individual's inclusion within generic mental health services. Risk assessment for self-harm, neglect, and harm to others is paramount and at all times the management plan must be recovery orientated with ensuring that the patient is given opportunities to express their interests and wishes. Treatment options including medications must be discussed with the patient and their family and paid carers and the patient's capacity to agree must be clearly documented.

In the UK (England and Wales) the National Institute of Health and Care Excellence and the Scottish Intercollegiate Guidelines Network (SIGN) (Scotland) have developed guidelines to drive improvements in the management of, among others, mental disorders including psychosis [87]. It is also expected that generic services will make reasonable adjustments when treating people with ID to include easy read information, longer appointments, increase in support, and advocacy to manage complex meetings and reviews. It is essential for clinicians to look for extrapyramidal and other side effects relating to antipsychotics and to ensure that patients have good access to health checks. Use of scales such as the Matson Evaluation of Drug Side-Effects Scale (MEDS) [88] or the Health of the Nation Outcome Scale-Learning Disability (HONOS-LD) [89] could be a significant aid in clinical practice.

Although there are several psychological and social interventions in the general population, there has not been an equivalent growth in people with ID. It is argued that a treatment that is effective or efficacious in people with psychosis without ID should also be available to those with ID. However, there are several issues with this argument: professionals claim to not have experience of treating people with ID, people with ID maybe unable to receive such treatments without adaptations due to cognitive difficulties or lack of knowledge as to what treatments are available. Some examples of treatments that have been delivered to people with ID and psychosis are CBT (positive improvements in symptoms and behaviour) [90] and psychoeducation [91]. Other promising areas of psychological intervention in the general population that could be beneficial in people with ID and psychosis include insight-related work, adherence therapy, family/carer work, and relapse prevention [92].

Hemmings, Underwoord, and Bouras [93] undertook a national Delphi exercise to examine professionals views and create consensus as to what services for people with ID and psychosis should provide. One hundred and thirty-nine items were included and following three rounds, over 80% agreement was reached on 18 of the routine service components, nine of the service user characteristics, and five of the more intensive service components. The authors suggest that certain elements of good care for people with ID and psychosis such as monitoring of mental state, crisis management, and support plans are already available within existing community ID services and their organizational context.

Whilst in the UK, services are more in tune with the care of people with ID and mental disorders, many parts of the world do not have specialist services or resources for this population group. It is also known that people with ID face disparities in healthcare and unequal access; therefore, global initiatives such as the WHO Mental Health GAP Action Programme (mhGAP) [94] are pivotal in improving awareness and reducing stigma of ID and mental illness.

Psychotic disorders require prompt, adequate, and long-term pharmacological and psychosocial interventions to manage the consequences on the individual, his/her family, and social networks. Although several psychological and social interventions have been developed for the general population, there has not been an equivalent growth for people with ID. Some examples of treatments that have been delivered to people with ID and psychosis are CBT and psychoeducation. Promising areas include insight related work, adherence therapy, family/carer work, and relapse prevention.

20.12 Psychopharmacology

High level evidence on the efficacy of antipsychotics in people with ID/low-functioning ASD (LF-ASD) and psychotic disorders is lacking. Most studies on psychotic disorders considered ID and LF-ASD as an exclusion criterion and specific research were hard to realize due to feasibility issues, including participant's compliance and approval by ethical committees.

Classical neuroleptics, also called first-generation antipsychotics, such as chlorpromazine, haloperidol, levomepromazine, clothiapine, and tiapride, seem to have a troublesome side effect profile, which includes a negative impact on cognitive functions and motor skills [95, 96]. Atypical neuroleptics, also called new-generation antipsychotics, such as risperidone, aripiprazole, olanzapine, quetiapine, and clozapine, show a better tolerability and safety profile with a mid and long-term efficacy comparable to that of the old-generation compounds or even higher. Most frequently reported side effects result to be weight gain and metabolic alteration, followed by sedation and more rarely extrapyramidal signs [97]. Risperidone may also determine neurological side effects, such as extrapyramidal symptoms (dyskinesia, akathisia, and parkinsonism), autonomic symptoms, and sedation [96–99]. Seizures, tachycardia, arrhythmias, and agranulocytosis are less common but more serious and require close monitoring, especially with clozapine [100].

Many combinations of antipsychotics may amplify side effects and should be carried out with great care [101].

The scientific literature suggests that new-generation antipsychotics, including clozapine, are frequently prescribed to people with ID and psychosis, although there is no systematic evidence to support its use in this population [102, 103]. Risperidone has been the most studied in aggressive behaviour, self-harm, and stereotyped behaviours, whether these behaviours were considered as determined by underlying psychotic disorders or not. Given the lack of evidence, clinicians are suggested to have an open dialogue with people with ID/LF-ASD and their caregivers as part of the discussion about informed consent for this treatment.

- High level evidence on the efficacy of antipsychotics in people with ID/LF-ASD and psychotic disorders is lacking.
- Adverse effects may be more likely than in the general population, thus potential sensitivities, monitoring issues, and medical comorbidities must be considered.
- Clozapine may be used with confirmed cases of treatment-resistant psychosis. This requires further precautions such as the patient's ability to co-operate with blood tests and other monitoring, and the consideration of epilepsy or elevated baseline cardiometabolic risk profile.

Tip

Future research should provide a more detailed picture of psychotic disorders in persons with ID and low-functioning ASD, especially in relation to age of onset, clinical presentations, disease progression, and aetiology. Also pharmacological and non-pharmacological interventions need a great deal of development supported by good level evidence.

Key Points

- Psychosis is a serious mental disorder with a chronic course associated with increase in all-cause mortality and a considerable burden on caregivers.
- The pathogenesis of psychotic disorders is currently supposed to be multifactorial and to include neurodegenerative, neurodevelopmental, and genetic causes.
- Psychotic disorders are frequently reported in persons with ID and/or ASD and are reported to have an earlier onset in persons with ID than in the general population.

- Psychotic disorders are characterized by a wide variety of conditions that share a cluster of clinical signs and symptoms that can be difficult to identify in people with ID and/or low-functioning ASD, especially in the case of severe communication difficulties. The identification of psychotic disorders in persons with ID and/or low-functioning ASD requires good diagnostic skills and the use of specific diagnostic manuals such as DC-LD and DM-ID 2.
- Patients with psychotic disorders require prompt, adequate and long-term pharmacological and psychosocial interventions to manage the consequences of the disorder on the individual, his/her family, and social networks.
- Although several psychological and social interventions have been developed for the general population, there has not been an equivalent growth for people with ID.
- High level evidence on the efficacy of antipsychotics in people with ID/low-functioning ASD and psychotic disorders is lacking.
- Adverse effects may be more likely than in the general population, thus potential sensitivities, monitoring issues, and medical comorbidities must be considered.

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Mood Disorders

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Contents

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Learning Objectives

To understand the challenges to proper diagnosis of mood disorders in persons with autism spectrum disorder (ASD) and intellectual disability (ID).

To recognize the signs and symptoms of mood disorders in persons with ASD and ID an distinguish them from behavioral problems.

To learn the treatment approaches to mood disorders in persons with ASD and ID.

21.1 Introduction

Mood disorders are common in the general population. Mood disorders include depressive disorders, and bipolar disorders historically referred to as "manic-depressive illness". These conditions cause great suffering for patients and for their families. For individuals with autism spectrum disorder (ASD) and intellectual disability (ID) the challenge of proper diagnosis as well as effective treatment presents a considerable problem. Many symptoms of mood disorders required for diagnosis depend on adequate verbal communication skills, so limitations in self-reflection and verbal reports of symptoms can make diagnosis difficult. Observable symptoms (e.g. being up all night with little need for sleep) are easily noted, but may be caused by other conditions and therefore may not be related to a mood disorder. On the other hand, many mood disorder symptoms may be mistaken for a "behaviour problems" in this population, resulting in a behavioural approach without proper psychiatric care. In this chapter, the most salient information on diagnosis and treatment of people with ID and ASD (ID/ASD) with mood disorders will be reviewed.

21.2 Diagnostic Peculiarities and Behavioural Equivalents

Before the 1980s most psychiatrists acknowledged only "behaviour" problems in people with ID/ASD. Sovner and Hurley [1] researched published case reports for patients with ID, finding 25 reports for patients with ID meeting DSM-III criteria for depression and/or mania, including nine in-patients who had ID/ASD. These reports underscored the difficulties in diagnosis. Often, the chief complaint is a challenging behaviour presented by a caregiver. This alone will invariably lead diagnosis and treatment in the wrong direction. Due to ID, the patient has difficulty answering questions in the interview. The person may also have "unusual symptoms" for someone in the general population, but these may be perfectly normal when compared to others with similar levels of ID. Lastly, the onset of a significant psychiatric illness may cause the person great distress, significantly reducing daily ability to function.

In 1986, Sovner and Hurley [2] proposed four categories of difficulty in psychiatric diagnosis for people with ID. These categories help in understanding the patient who cannot speak eloquently for themselves and supply realistic answers during the psychiatric diagnostic interview. Often, behavioural and/or unusual symptoms have led to overdiagnosis of a schizophreniform disorder rather than recognition of the more common mood disorders. The first category, *baseline exaggeration*, refers to an increase in challenging behaviour frequency and/or intensity during the course of a mental illness. For example, an individual may have a low rate of mild self-injurious behaviour, but during a time of stress or acute psychiatric disorder the frequency and intensity of this behaviour escalates to a dangerous level, prompting a mental health appointment. For some people with ID/ASD, self-injury often increases and causes significant medical risk.

The second, *intellectual distortion,* finds that the individual cannot accurately understand the questions posed by the evaluator nor assemble the correct information to respond. For example, when asked if he or she "hears voices", the individual might respond affirmatively without fully comprehending the implication in the question: the clinician

meant abnormal phantom voices, whereas the individual was thinking of ordinary voices, including the clinician's, that he or she was able to hear. This example may seem understandable, but many responses are clearly inadequate or incorrect. Much as when interviewing children, no answer can be taken at face value.

The third, *psychosocial masking,* focuses on the effects of developmental delay. The individual might present significant symptomatology that occurs within a developmental framework and would be common in a young child. For example, a manic individual with moderate intellectual disability might believe that he or she can drive a car, a skill well beyond such an individual's capability, as a manifestation of grandiosity.

Lastly, *cognitive disintegration* is a situation in which the person has become grossly disorganized, even psychotic, due to the lack of "cognitive reserve" available to cope with the illness. For example, in a major depressive episode, individuals with ID are more likely than neurotypical individuals to be severely affected and suffer mood-congruent hallucinations and delusions. Further, additional behavioural repertoires or thought patterns might be due to, or consistent with, other aspects of developmental delay. That is, they would be considered typical in a young child but would appear bizarre and abnormal in an adult.

For people with ID/ASD, symptom presentation is challenging to interpret. The idea of "behavioural equivalents" of symptoms for people with ID/ASD has been controversial (see ▶ Chap. 5). However, we believe that the format is useful in integrating the illness with symptoms of two other separate disorders that interact with the mood disorder (i.e. ID and ASD) as well as in organizing thoughts about a particular case. The DSM does not directly deal with behavioural symptoms regarding people with ID/ASD [3]. Pary and colleagues proposed a system of "equivalents" in symptoms for depression and bipolar disorder including a comparison of patients with ID and those with ID/ASD [4] (see ◘ Tables 21.1 and 21.2).

Baudewijns, Bertelli, and collaborators [7] have recently found that in persons with ID and ASD major depressive disorder tends to present with a range of aggressive behaviours (mostly self-oriented), which seems to be further differentiable by predominance of emotional symptoms (irritability, anxiety and sadness) or physical symptoms (problems with energy, eating, sleeping, and stress) [7].

With the presence of "behavioural symptoms" it is imperative that the clinician carefully consider how mood difficulties may be causing behavioural problems.

For individual with ID and ASD a proper diagnosis of mood disorders as well as an effective treatment presents considerable challenges. Baseline exaggeration, intellectual distortion, psychosocial masking, and cognitive disintegration are just some of the phenomena that can make the diagnostic process difficult in this population.

21.3 Prevalence

21.3.1 Depression

Depression takes a great toll on the suffering person, and it affects family and friends as well. When ill, physical health is affected. People with depression have alterations in sleep and appetite. They may feel a lack of energy. They also feel inadequate and can distort interactions and self-assessments, with loss of self-worth. Treatment, however, is fairly straightforward. There are many medications that can help. Psychotherapy is always recommended and there are many forms of psychotherapy to choose from. In addition, people with seasonal affective disorder (SAD) become depressed when the winter season is arriving. For this condition, the use of a Bright Light Therapy typically offers

Table 21.1 DSM-IV and DSM-5 criteria and symptoms for major depressive episode with proposed ID and ASD equivalents [4]

DSM criteria	ID equivalents	ASD equivalents
1. Depressed mood or irritable mood	Apathetic, sad or angry facial expression; lack of emotional reactivity; upset; crying; tantrums; verbal and physical aggression carefully judged for context	Irritability; crying; tantrums; verbal and physical aggression carefully judged for context
2. Markedly diminished interest or pleasure in most activities	Withdrawal; lack of reinforcers; refusal to participate in leisure activities or work; change in ability to watch TV or listen to music	Withdrawal; diminished or suddenly exclusive pursuit of fascinations and obsessions, rituals and compulsions
3. Significant weight loss; decrease or increase in appetite	Tantrums at meals; stealing food; refusing activities; hoarding food in room	Food refusal; restriction of intake to a few ritualized items; indiscriminate hyperphagia
4. Insomnia or hypersomnia	May or may not be able to self-report sleep problems; if living with others, may report being up at night; others may note going to bed quite late; if living in staffed situation, staff may note being up at night; any change in sleeping habits; tantrums or activity during sleeping hours; noted sleeping or napping during the day	May or may not be able to self- report sleep problems; if living with others, may report being up at night; others may note going to bed quite late; if living in staffed situation, staff may note being up at night; any change in sleeping habits; tantrums or activity during sleeping hours; noted sleeping or napping during the day
5. Psychomotor agitation or retardation	Pacing, hyperactivity; decreased energy, passivity; development of obsessional slowness in activities of daily living; muteness; whispering; monosyllables; increase in SIB or aggression carefully judged for context	Decreased energy, passivity; development of obsessional slowness in activities of daily living; muteness; whispering; monosyllables; increase in self-injurious behaviour or aggression, rituals and compulsions
6. Fatigue or loss of energy	Appears tired; refuses leisure activities or work; withdraws to room; loss of daily living skills; refusal to do personal care; incontinence due to lack of energy/motivation to go to bathroom; work production decreases; does not want to join activities; just watches TV, sitting for long periods of time	Appears tired; withdraws to room; loss of daily living skills; refusal to do personal care; incontinence due to lack of energy/motivation to go to bathroom
7. Feelings of worthlessness	Statements such as "I'm stupid" or "I'm bad" or "I'm not normal"	Statements such as "I'm stupid" or "I'm bad" or "I'm not normal"
8. Diminished ability to think or concentrate	Poor performance at work; change in leisure habits and hobbies; appears distracted	Poor performance at work; change in leisure habits; appears distracted
9. Recurrent thoughts of death, suicidal Behaviour or statements	Perseveration on the deaths of family members and friends; preoccupation with funerals	Perseveration on the deaths of family members and friends; preoccupation with funerals

Mental Health Aspects of Developmental Disabilities [4]
Legend: The symptoms in the left column are those described in DSM-IV [3] and DSM-5 [5]. In the second column are proposed "equivalents" to symptoms as they might appear in a person with ID as noted in case reports and research articles. The third column addresses symptom presentation in patients with ID/ASD

Table 21.2 DSM-IV and DSM-5 criteria and symptoms for manic episode with proposed ID and ASD equivalents [4]

DSM criteria	Suggested ID equivalents	Suggested ASD equivalents
1. Inflated self-esteem or grandiosity	In relationship to his/her developmental level, person believes and acts as if he/she can do more (e.g. person believes he/she is a teacher, staff member; can drive a car; can repair a broken TV; is better than peers)	In relationship to his/her developmental level, person believes and acts as if he/she can do more (e.g. able to engage in social relations successfully, overestimates likely success or comfort in a social world
2. Deceased need for sleep (e.g. feels rested after only 3 hours of sleep)	Person is up at night; is active and about the room, house; awakens early or does not get to sleep until quite late; appears energetic next day	Person is up at night; is active perhaps with rituals; awakens early or does not get to sleep until quite late; if sleep usually irregular, completely sleepless
3. More talkative than usual or pressure to keep talking	Person talks constantly, often seeking attention; cannot listen to others easily, not really conversing but espousing own thoughts	Increased frequency and/or intensity of perseverative questioning, manneristic speech
4. Flight of ideas or subjective experience that thoughts are racing	Ideas flow because of energy; topics are short, and next topic often unrelated to last; cannot respond easily to topics generated by others	Ideas flow because of energy; topics are short, and next topic often unrelated to last; rapid repetitive speech; increased vocal stereotypy
5. Distractibility (i.e. attention too easily drawn to unimportant or irrelevant external stimuli)	Compared to usual abilities to concentrate on tasks, is distracted by environment, but mainly by own internal energy; ADL tasks and work tasks completed quickly and improperly, skipping from activity to activity	Pronounced inability to concentrate on tasks; distracted by environment; skipping from activity to activity; rituals may become rapid or disorganized
6. Increase in goal-directed activity (either socially, at work, school or sexually) or psychomotor agitation	May work at ADLs or work with great speed, but little attention to detail or work quality; cannot respond to cues to slow or repeat work if sloppy; may create new tasks, take on projects, talk about new jobs and work that is not realistic; may be overactive and/or appear to be in constant motion	Overactivity; increased frequency and/or intensity of ritualistic or compulsive activities
7. Excessive involvement in pleasurable activities that have a high potential for painful consequence (e.g. engaging in unrestrained buying sprees, sexual indiscretions, or foolish business investments)	Increase in obvious sexual interests; more preoccupied with hobbies or general recreational interests; intrusiveness; inhibition; inability to follow previously understood rules and limits	Increase in obvious sexual interests; more preoccupied with obsessions and fascinations; intrusiveness; disinhibitions; inability to follow previously understood rules and limits

Mental Health Aspects of Developmental Disabilities [4]

Legend: The symptoms in the left column are those described in DSM-IV [4] and DSM-5 [6]. In the second column are proposed "equivalents" to symptoms as they might appear in a person with ID as noted in case reports and research articles. The third column addresses symptom presentation in patients with ID/ASD

excellent results. For severe resistant cases of depression, electroconvulsive therapy (ECT) is another option.

Depression is a common psychiatric illness. A study of the general population of the United States utilized the World Health Organization Composite Diagnostic Interview to assess lifetime and 12-month prevalence of DSM-IV axis I disorders. Subjects were individually interviewed by trained staff. The findings for lifetime prevalence of psychiatric disorders were 16.2% of the responders, with 10.4% judged mild; 38.6% moderate; 38.0% severe; 12.9% very severe. Authors noted that only 41.9% of cases were receiving adequate treatment [8]. Premenstrual dysphoric disorder (PMDD) is reported to have a prevalence of 3–8% throughout reproductive age [9]; however, some studies suggest that up to 20% of all women of fertile age have premenstrual complaints that could be regarded as clinically relevant [10].

Smith and colleagues [11] using the UK Biobank of 172,751 individuals found lifetime prevalence rates for a single lifetime episode of major depression of 6.4%, recurrent major depression (moderate) 12.2%, probable major depression (severe) 7.2%, and probable bipolar disorder 1.3%, similar to those found in other population studies.

21.3.2 Prevalence of Depression in People with ID

The prevalence of depression and aging was investigated by Cooper [12] using ICD-10 Kettering/Leicester criteria. A comprehensive psychiatric examination and semi-structured rating scale was used for people with intellectual disability over age 65 ($n = 134$) in a defined geographic area and compared with adults ages 20–65 ($n = 73$). The rate of depression for elderly patients was 6% and 4.1% for younger individuals. In a review of depression and the DC-LD, Smiley and Cooper [13] found that a major problem in the diagnostic formulation is the presence of unusual features appearing during a depressive episode such as lability of mood, onset of challenging behaviours, reduction in speech, social withdrawal, and increase in somatic complaints. Indeed, these symptoms would be unusual for adults in the neurotypical range of intelligence.

A comprehensive set of epidemiological studies of patients with ID were conducted over several years by Cooper and her colleagues [14]. Cases were ascertained in a catchment area through extensive contacts with support agencies and medical personnel resulting in 1023 subjects. Each person was assessed using multiple measures and a standard psychiatric assessment. In addition, four diagnostic approaches were used: clinical judgement; ICD-9; DC-LD; and DSM-IV. In their study of mood disorders, the following results were obtained for unipolar depression episode: clinical judgement 4.1%; ICD-9 3.5%; DC-LD 2.8%; DSM-IV 2.0% [14]. It is important to note that clinical judgement prevailed because so many official criteria depend on self-report that can be unreliable due to intellectual disability. Many patients with intellectual disability could not self-report changes in mood time, feelings of inadequacy, or hopelessness. In addition, if challenging behaviour had developed or increased, the clinical picture was more complicated.

21.3.3 Prevalence of Depression in People with ID/ASD

Melville and colleagues also conducted an epidemiological assessment of the mental health of people with ID and ASD [13]. The larger study identified 77 adults with ASD (mean age was 37.8 years); 59 were men, 18 were women; 14 had mild ID, 14 had moderate ID, 21 severe ID, and 28 profound ID. Controls (158) were matched for sex, level of ID, and Down syndrome diagnosis. Fifty subjects had ASD and were matched to controls. Results were as follows for mood disorders: clinical judgement 5.2%; DC-LD 3.9%; DCR-ICD-10 3.9%; DSM-IV 2.6%. Overall, the incidence of mental ill health was similar to studies on people with only ID. In a second phase of the study a 2-year follow-up was conducted for

98 subjects. The recovery rate over 2 years was less for the subjects with ASD. The most common new disorders were mood disorders. There were no new episodes of mania.

Sterling and colleagues [15] investigated depression using the Autism Diagnostic Interview-Revised [16] and the Autism Diagnostic Observation Schedule (ADOS) [17] a semi-structured observation and interview administered directly to the participants, intelligence test and a psychiatric diagnostic interview. Those subjects with higher intellectual function had more symptoms of depression; 20 (43%) had depressive symptoms. Those with higher cognitive abilities showed less social impairment and were most likely to report depression. Thus people with ID/ASD may have less vulnerability to become depressed, perhaps because they are more unaware of social difficulties, or may be underdiagnosed because they cannot report or display symptoms.

Lainhart and collaborators [18] reviewed all published cases at that time of mood disorders and ASD, with a total of 17 patients. Almost all case reports were individuals with associated ID; 47% of patients were female and 50% had a family history of mood disorders. The chief complaint for 9 of 17 patients was challenging behaviour: aggression, self-injury, destructiveness, and temper outbursts. Repetitive behaviours increased in two cases, and social withdrawal and vegetative symptoms were prominent in two other cases. For all patients, however, vegetative symptoms, such as changes in sleep, were notable. However, no patients with ASD reported a change in self-attitude. For this group, four reported manic episodes only; eight had depression only; four had episodes of mania and depression. The difficulties in diagnosing related to limited self-reflection and expression of feelings are highlighted by reliance on observable behaviour.

Leyfer, Folstein, Bacalaman, and colleagues [19] conducted a study of children on the autism spectrum using the *Autism Comorbidity Interview – Present and Lifetime Version* [*Kiddie Schedule for Affective Disorders and Schizophrenia* (KSADS)] [20] modified for children and adolescents on the autism spectrum. Children were recruited from Salt Lake City and Boston resulting in 109 children ranging in age from 5 to 17 years. The diagnosis of ASD was confirmed by a senior clinician. All met DSM-III criteria as assessed with the Autism Diagnostic Interview Revised [16] and Autism Diagnostic Observation Scale [21]. A total of 109 subjects were recruited. The majority were males (94; 29%); 67.71% had IQ below 70; 57.45% verbal IQ below 70; 49% had nonverbal IQ below 70. Rates of depression were 10.1% and 13.8% for subsyndromal depression; 2.8% met criteria for depression NOS.

Henry and colleagues [22] conducted a study of mood disorder diagnoses in an outpatient sample of youth with ASD. This clinic identified 123 referred children and adolescents meeting DSM-IV-TR diagnostic criteria for autistic disorder. Mood disorder diagnoses and chief complaints along with family mood disorder history were the primary variables analysed. Four subjects (3%) presented with depressed mood. Irritability complaints were frequent ($n = 78$, 63%). Six subjects (5%) received a mood disorder diagnosis (Mood Disorder Not Otherwise Specified). No subjects received a depressive disorder diagnosis. Family history of mood disorders was common.

Regarding premenstrual dysphoric disorder, Obaydi and Puri in a systematic prospective observer-rated study on the prevalence of this disorder in women with ASD found a rate of around nine times higher than that of women of the general population (92% vs 11%) [23].

Depression is a common psychiatric disorder in persons with ID and/or ASD, even if it is not properly recognized. Many patients could not self-report changes in mood onset, feelings of inadequacy or hopelessness, and present unusual features such as onset or increase of challenging behaviours. In most cases the diagnosis is based on clinical judgement.

21.4 Criteria and Clinical Features

21.4.1 Depression

The ICD-10 uses a list of 10 depressive symptoms [24]. Key symptoms are persistent sadness, or low mood and/or loss of interest or pleasure and fatigue or low energy. One of these symptoms must be present most days, most of the time, for at least 2 weeks. In addition, other symptoms include disturbed sleep, poor concentration or indecisiveness; low self-confidence; poor or increased appetite; suicidal thoughts or acts; agitation or slowing of movement; guilt or self-blame.

These 10 symptoms define the degree of the depressive disorder. One to three symptoms present suggests the person does not have a depressive disorder. Four or more symptoms suggest mild depression; five to six symptoms suggest moderate depression; seven or more symptoms suggest severe depression which can be associated with psychotic symptoms. Lastly, the symptoms must be present for a month or more during most of the day.

In the general population, depression is fairly easy to recognize. Patients with ID and ASD cannot easily articulate symptoms or answer questions during the interview. Extra time is needed and third-party information can help establish the development of symptoms.

People with ID/ASD may present with a variety of behavioural symptoms due to the suffering of depression; however, behavioural symptoms are often difficult to interpret (see Table 21.1). Any change in behaviour, however, may be a signal of depression, or other disorder including a medical condition. Key symptoms observed may be changes such as withdrawal, changes in sleep pattern, increase in agitation, and decrease or increases in appetite. These are observable and can be tracked by caregivers. Sleep charts, changes in weight, and measures of participation in daily activities can be reported by others.

Stewart and her colleagues [25] did a literature review of depression in case reports and studies. For 15 case reports, common symptoms were regression in speech, loss of hygiene, loss of bowel and bladder control, increase in stereotypies, and self-injury. A summary of 14 studies at the time on depression and ASD found 10 of 15 studies included subjects who had ID and ASD. Those with ID and ASD had more negative life events, increased anxiety and externalizing behaviour, self-injury. Difficult symptoms associated with ASD such as obsessions may increase during a depression. Impaired verbal and nonverbal communications are barriers to the typical diagnostic interview.

Suicide and suicidal thoughts are most often linked with depression, either during a major depressive episode or in the depressive phase of bipolar disorder. Among people with ID and ASD, there is a low rate of suicide. However, reports of suicide exist. Suicide appears impulsive and generally not planned as it might be in a neurotypical person.

Several studies of suicide among people with ID alone were located. Sternlict and colleagues found a rate of nine suicide attempts per 1000 patients at a large state institution [26]. Methods used included jumping from a high place, use of a sharp instrument, and hanging/strangulation. Hardan and Sahl [27] examined the medical records of all children and adolescents assessed in a special developmental disabilities programme over a 1-year period, abstracting information reporting suicidal ideation, threats, or suicide attempts. They found that 20% of the patients made suicidal attempts. Benson [28] identified 12 patients in a large clinic for persons with ID who had made suicide attempts and ten who had suicidal ideation. In that study, ingestion of medication, cutting with sharp instruments, suffocation, and ingestion of toxic liquids were the most frequently used methods. Walters and colleagues [29] examined 90 consecutive admissions to an inpatient service for children and adolescents with intellectual disability. Ten patients had suicidal behaviour, and 60% of those acts were potentially lethal.

Lastly, Pary and colleagues [30] surveyed suicidal behaviour in persons with Down syndrome compared to a control population of persons with ID of other aetiologies using the Client Development Evaluation Report of

the California Department of Developmental Services. Of 11,277 individuals with Down syndrome, four had reports of suicidal behaviour (0.04%) compared to 1142 reports of suicidal behaviour in a total of 143,143 non-Down syndrome individuals (0.8%).

Suicide is certainly known to occur in people with Asperger's disorder and high-functioning autism. Mayes and her colleagues [31] studied the frequency of suicide ideation and attempts. They compared 791 children with autism, 35 neurotypical depressed children, and 186 neurotypical children. Mothers of children with ASD reported 14% had suicidal ideation or attempts very often, 28 times greater than neurotypical children and less than depressed children (43%). Associated diagnoses were depression and behavioural problems. The majority of children (71%) had four risk factors, such as socio-economic status. Children with high-functioning autism and low-functioning autism did not differ in suicidal ideation or attempts.

In a review of suicide and ASD, Richa and his colleagues [32] found ample evidence from 15 case studies; eight had additional ID. Precipitating events included childhood adversity, stressful life events, bullying, and physical and sexual abuse. In this way, causes of suicide are similar to the general population. Decrease in self-care was quite common. People with ASD may have more risk factors compared to neurotypical people and be more vulnerable to teasing, bullying, or abuse from others. Additional factors such as menstrual cycles can often have effects on women with ASD [33]. Some authors argue that suicide is more common in women with premenstrual dysphoric disorder, as they are more vulnerable to suicidal thoughts, plans, and attempts [34].

Any change in behaviour may be a signal of depression. Changes in sleep pattern, appetite, body weight, and participation in daily activities can represent depressive symptoms. These are observable and can be tracked by third-party information.

Suicide is not common in people with ID and low-functioning ASD.

21.4.2 Bipolar Disorder

Bipolar disorder is a serious and complex illness. Most people understand the concept of "depression" and therefore can easily understand that a severe depression would be immobilizing. It is more difficult to understand the intensity of bipolar illness: a "high" of power and feeling "on top of the world", mood swings, vast changes in typical behaviour, and physical changes such as loss or increase in appetite as well as sleep disorder. In addition, the effect of the mood (whether depressed or manic) is extreme for patients and for anyone with whom they interact. Further complexity is due to the episodic pattern. For some episodes are frequent, for others infrequent, for still others occurrence is very erratic. Because of the seriousness of this illness, people often need to be hospitalized to be safe and receive treatment. In addition, there is often a family history of the illness.

There are multiple conditions that can cause cycles of maladaptive behaviour and thus be confused with bipolar disorder in people who have ID. Pary, Levitas, and Hurley [4] describe various cyclic medical conditions, such as hay fever and inflammatory bowel diseases, which can cause great distress and resulting behavioural difficulties. A case of bipolar disorder was reported in a girl with mild ID and ASD due to menstrual difficulties resulting in acute mania [33].

Clarke and his colleagues [35] reviewed papers published with many historical cases of depression or bipolar disorder and 21 reports had ID/ASD. Clarke reported seven new cases: rapid cycling bipolar disorder; severe depressive episode with psychotic features; severe depressive episode with psychotic features; recurrent depressive disorder; bipolar affective disorder; depressive episodes; one had paranoid schizophrenia.

Matson and Smiroldo studied the mania scale of the Diagnostic Assessment for the Severely Handicapped-II (DASH-II) [36, 37]. Three groups were compared: individuals diagnosed with bipolar disorder who were currently manic, individuals with other Axis I disorders, and those with no Axis I disorders. Key symptoms of mania found were psycho-

motor agitation, decreased sleep, changes in mood, and aggression. In 2007, Matson and his colleagues further studied symptoms of mania and found psychomotor agitation and disturbed sleep were most predictive of bipolar disorder [38].

Hurley and colleagues [39] conducted a study comparing three groups of outpatients in a general psychiatry clinic: 100 neurotypical patients within normal range of intelligence; 100 patients with mild ID; 100 patients with moderate to profound ID. The chief complaint of aggression was the most common for patients with ID: Normal IQ 6; Mild ID 45; Moderate to profound ID 37. Indeed, patients had many other symptoms, but this illustrates that challenging behaviour is often the common symptom found in very distressed people with ID. Further, because challenging behaviour is seen in many conditions, in itself, this symptom has limited value as a specific indicator for a specific diagnosis.

Hurley [40] conducted an outpatient study of patient groups with ID diagnosed with depression, bipolar disorder, anxiety disorders, and patients with no psychiatric disorders seen for disability status review. Bipolar patients were significantly different from depressed patients for elevated mood, acute anger episodes, increase in verbalization, pressure of speech, talk of sexual themes, increase in appetite, and poor concentration. Challenging behaviour was most pronounced in bipolar patients.

Wozniak and collaborators [41] described a series of 66 children and adolescents with ASD. Twenty-one per cent of the children were diagnosed with bipolar disorder. Symptoms of bipolar illness were reported at similar rates for children with and without ASD. Children with mania had a high rate of irritable mood or mixed states. They were also more often described as having a chronic presentation rather than an episodic course.

21.4.3 Prevalence of Bipolar Disorder

Merikangas [42] and her colleagues investigated the 12-month prevalence of bipolar disorder as part of the National Comorbidity Survey Study utilizing the World Health Organization Composite Diagnostic Interview to assess lifetime and 12 month DSM-IV axis I disorders. Lifetime prevalence was 1% for bipolar 1, 1.1% for bipolar 2, and 2.4% for subthreshold cases. Subthreshold cases had significant impairments. Although most patients had lifetime treatment available, it was noted that many were not receiving antimanic medications, especially if seen in general medical settings.

21.4.4 Prevalence of Bipolar Disorder in People with ID

Cooper and her colleagues [14] included bipolar disorder in their epidemiological study. Point prevalence rates for bipolar disorder, currently depressed, were as follows: clinical judgement 0.5%; ICD-10 0.2%; DC-LD 0.3%; DSM-IV 0.1%. Results for mania, currently in episode: clinical judgement 0.6%; DC-LD 0.6%; CDR- ICD-10 0.6%: DSM-IV-TR 0.5%. First episode of mania: clinical judgement 2%; DC-LD 3%; ICD-10 3%; DSM-IV 4%. Bipolar disorder diagnoses occurred at about twice the estimated rate for the general public. For bipolar disorder in the depressed phase, DSM-IV-TR missed half of cases. This is thought to result from reliance on verbal self-report of depressive symptoms, whereas the outward symptoms of mania may be easier to recognize.

21.4.5 Prevalence of Bipolar Disorder in People with ID/ASD

Mellville, Cooper, and colleagues [15] epidemiological study of people with ID and ASD measuring their mental ill health was reported above. Results were as follows for mood disorders: clinical judgement 5.2%; DC-LD 3.9%; DCR-ICD-10 3.9%; DSM-IV. 2.6%. There were no episodes of mania. Given the extensive history and interviews in this study, it may be significant that there was not good

evidence of bipolar disorder as there were no reports of a manic episode.

As previously reported, Leyfer and colleagues [19] conducted a study of children with autism spectrum. The *Kiddie Schedule for Affective Disorders and Schizophrenia* (KSADS) [20] was modified for children and adolescents on the autism spectrum. Less than 2% had a manic episode or symptoms of bipolar disorder, including two with a manic episode; one cyclothymia; two bipolar-1; one bipolar-2; one hypomanic episode; two mixed bipolar episodes. This detailed study included subjects who were within a normal range of intelligence.

In persons with ID bipolar disorder diagnoses occur at about twice the estimated rate for the general population. Key symptoms of mania are psychomotor agitation, decreased sleep, changes in mood, and aggression. Challenging behaviours in general are very common and often much pronounced.

21.4.6 Other Specific Mood Disorders: Criteria and Clinical Features

There is great variability in symptomatology as well as number and length of episodes among patients. ICD-10 [24] lists the following criteria.

21.4.6.1 Mania Without Psychotic Symptoms

In manic episodes without psychotic symptoms some main psychic dimensions are characterized as following.

Affect: Elevated mood; irritability; distance; self-esteem (high); affectability; aggressiveness and hostility

Drive: Increased activity; psychomotor agitation

Vital functions: Reduced sleep needs, increased libido

Formal thinking: Flight of Ideas.

In Confused Mania confusion is caused by an extreme acceleration of thinking and speaking.

21.4.6.2 Bipolar Affective Disorder

In the mixed episodes, the symptoms of mania and depression occur in rapid alternation or are present simultaneously. The inhibition (depression) or excitation (mania) of emotional, cognitive, and psychomotor processes, which move in the same direction, are no longer present in mixed states. Thus, for example, quick thinking and talking (as in mania) can occur simultaneously with anxious-depressive mood. A mixed state is a very diverse, sometimes difficult to diagnose and treat, disorder.

21.4.6.3 Cyclothymia/Cyclothymic Disorder

Cyclothymia or cyclothymic disorder is an attenuated form of bipolar disorder. For these patients, mood variations are often observed over a period of at least 2 years, with the criteria for a depressive- or manic episode not being met in full and often occurring independently of external influences. However, the transition is very fluid, so that a bipolar (ll) disorder can often be detected in people who come to a clinic with a suspected diagnosis of cyclothymia.

21.4.6.4 Severe Depressive Episode with Psychotic Symptoms

The affected patient had at least one clearly diagnosed hypomanic, manic, or mixed affective episode and at least one further affective episode (hypomanic, manic, depressed, or mixed) in the anamnesis; in recent months and presently there is no clear disturbance of mood. Remissions during prophylactic treatment are also encoded here.

21.4.6.5 Moderate Depressive Episode

The affected patient is currently depressed, as in a mild or moderate depressive episode, and had at least one clearly diagnosed hypomanic, manic, or mixed episode in anamnesis.

21.4.6.6 Premenstrual Dysphoric Disorder

Marked affective lability, irritability, depressed mood, and anxiety represent the main symptoms. At least one of these must be present in the week leading up to menstruation together with additional possible symptoms such as decreased interest in typical activities, difficulties in concentrating, poor energy, changes in sleep or eating, feeling overwhelmed or out of control, and physical symptoms (i.e. sensation of bloating, muscle pain, etc.).

This diagnostic category has been introduced in the ICD-10 with the 2017's revision.

In ICD-11 Depressive Disorders and Bipolar Disorders are grouped together as Mood Disorders. Mood disorders are classified based on the sorts of Mood Episodes that occur and their patterns throughout time. The main types of mood episodes include Depressive Episode, Manic Episode, Mixed Episode, and Hypomanic Episode. Mood Episodes do not have their own diagnostic codes since they are not independently diagnosable entities. Mood Episodes, on the other hand, are components of Bipolar and Related Disorders, as well as Depressive Disorders. Bipolar or Related Disorders include Bipolar Type I Disorder, Bipolar Type II Disorder, Cyclothymic Disorder, and Other Specified Bipolar or Related Disorders. Depressive Disorders include Single Episode Depressive Disorder, Recurrent Depressive Disorder, Dysthymic Disorder, and Other Specified Depressive Disorders. Mixed Depressive and Anxiety Disorder is included in the section on Depressive Disorders, although it has certain characteristics in common with Anxiety or Fear-Related Disorders [43].

21.5 Aetiopathogenesis

21.5.1 Depression

Depression is one of the most common mental illnesses in the general population. Human beings experience sadness: a normal reaction to difficulties in life. Often, people report they are "depressed" about something, but these expressions are not the same as a major depressive episode.

While most common in adults, depression can occur in children and teenagers. Depression, especially in midlife or older adults, can co-occur with other serious medical illnesses, such as diabetes, cancer, heart disease, and Parkinson's disease. These conditions are often worse when depression is present. Sometimes medications taken for these physical illnesses cause side effects that cause or contribute to depression.

Causes of depression include genetic predisposition, major life changes, trauma, increase in stress, major loss of supports or friends, physical illness, disability, and medication side effects. In addition, some people are susceptible to seasonal affective disorder, becoming depressed during the fall and winter season.

Depression in people with ID/ASD can be associated with medical conditions. Autism occurs with known medical conditions in at least 10% of cases [44]. These conditions may increase the vulnerability to depression and modify its symptoms. Most notably about 30% of people with ASD have a seizure disorder [45]. Because a seizure disorder is often associated with depression in the general population, people with ASD and a seizure disorder may be more vulnerable to depression [46]. Hormonal dysregulation or sensitivity has been indicated to be involved in the pathogenesis of premenstrual dysphoric disorder, although knowledge on the origin of this condition is only in the early stages [47].

Family genetic factors play an important role in the etiology of depression in the general population, in both children and adults [48, 49]. Children with ASDs who suffer from depression are more likely to have a family history of depression [50]. Since depression does not appear to be part of the broader phenotype of autism [51] it is possible that autism with comorbid depression may form a distinct sub-group.

Life events are also known to be related to depression in the general population.

Ghaziuddin and colleagues [50] compared 11 children with ASD and depression to 11 matched controls without depression and researched unpleasant life events in the prior 12 months using a life events schedule. Nine patients with depression (82%) gave a history of having sustained recent unpleasant life events compared to five (45%) in the nondepressed group. These events consisted of the change in living arrangements, change of education programs, family illness, bereavement, and other losses.

21.5.2 Bipolar Disorder

Bipolar disorder has many possible causes. Genetic inheritance is often cited and found to be involved in many cases. It may also occur after a serious medical illness. Some medical conditions that are associated with ID and/or ASD are also associated with bipolar disorder. For example, bipolar spectrum disorder has been reported by some researchers to occur in 70% of affected individuals with velo-cardio-facial syndrome (VCFS; 22q-, 22q22.1DS) by adolescence, approaching 100% by adulthood [52]. Verhoeven [53] and colleagues identified Phelan-McDermid syndrome in two adult brothers with atypical bipolar disorder. Kerbeshian and colleagues [54] reported atypical bipolar disorder in a 33-year-old female with a deletion of 15q12. She had profound ID, was explosive, had episodes associated with insomnia, cyclical expansive mood. Mood improved markedly with lithium. Sovner and colleagues [55] reported a case of rapid cycling bipolar disorder in a man with ring chromosome 22, and 7 other cases in the literature. The patient responded well to divalproex and fluoxetine. It is currently thought that numerous ID and ASD genetic syndromes contain within their genome deletions genes associated with the endophenotypes of psychiatric disorders, possibly accounting for psychiatric comorbidity in these syndromes [56].

Medical conditions are also associated with the development of bipolar disorder. This was first reported by Krauthammer and Klerman [57] and called "secondary mania". In the general population, conditions associated with mania include degenerative neurological disorders, cerebrovascular disease, metabolic conditions, endocrine conditions, autoimmune conditions, viral or other infections, and certain cancers. There is a report of secondary mania in a patient with ID and polycystic ovary disease [58].

Causes of depression include genetic predisposition, major life changes, trauma, increase in stress, major loss of support or friends, physical illness, disability, and medication side effects. Bipolar disorder seems to be more associated with genetic inheritance and medical conditions.

21.6 Specific Assessment

Due to the complexity of patients with ID/ASD, specific assessments beyond the psychiatric diagnostic interview are essential to understand the patient, as well as monitor treatment. On a simple level, one can ask for any data held by school departments and other social services. Obtaining a properly administered IQ test is essential and hopefully assessments were conducted at least during the early school years. These will give good clinical information as to verbal abilities and other organizational abilities. When starting a new patient, best practice is to make every effort to get full medical records and school records.

Formal assessment using published instruments is helpful to diagnosis and often to assess improvement. For people with ID/ASD assessment of sensory sensitivities and stress is essential. An excellent instrument was developed by Groden and her colleagues, the *Stress Survey Schedule for Individuals with Autism and Other Pervasive Developmental Disabilities* [59]. A carer is interviewed about items commonly experienced as stressful by persons with autism spectrum disorders (e.g. "receiving hugs and affection" or "waiting in line") and rated on a 5-point scale. The items then score to important groupings for plan-

ning purposes: changes; unpleasant, (even positive) sensory-personal, food-related, social-environmental, rituals. A study of informants and self-report of persons showed modest agreement, and it is recommended that self-report is always an important factor [60]. Prior to considering a secondary mental health condition in a person with ID/ASD, a thorough evaluation of stressful events and sensory input is essential to understand complex behavioural reactions. We know that many sensory situations are experienced as aversive by people with ASD. Thus, it is essential to recognize behavioural reactions to sensory events vs. mistaking these behaviours for psychiatric disorders.

Because people with ID and ASD commonly exhibit many behavioural difficulties, assessment of behaviours is also essential. The *Reiss Screen for Maladaptive Behavior (RMB)* [61] contains 38 items rated ideally by two individuals who know the person well and the scores are averaged. This instrument can be used for all levels of ID. Items are rated on a 3-points scale for severity on eight subscales: Aggressive, Autism, Psychosis, Paranoia, Depression and Behavioural signs, Depression and Physical Signs, Dependent Personality Disorder, and Avoidant Personality Disorder. Individual items of interest not included in subscales are drug abuse, overactive, self-injury, sexual problem, suicidal, stealing. The Reiss Screen has been used in a number of studies and is popular because it is easy to complete and score and provides useful information. Although the title suggests it addresses "behaviour", it is rather an excellent instrument to assist in psychiatric diagnosis.

A more thorough and complex set of assessments are also an excellent addition to help with complex cases. The *Psychiatric Assessment Schedules for Adults with Developmental Disabilities* (*PAS-ADD*) [62] includes several measurement options. The *PAS-ADD* clinical interview [63] is the most comprehensive help with establishing a diagnosis using both ICD-10 and DSM IV. The PAS-ADD Checklist [64] is a 25-item questionnaire, designed for use primarily by care staff and families. The Mini PAS-ADD can be used by a wide range of professionals and an suggest a possible psychiatric disorder. This can be completed by interviewing a caregiver or by collecting information through chart reviews. These assessments are highly recommended for complex cases.

The direct assessment of challenging behaviours is also important. The *Aberrant Behavior Checklist* (*ABC*) [65, 66] is an informant rating scale that assesses specific behavioural problems in people with all levels of ID from ages 6–54. It has 58 items distributed on five subscales: Irritability, Lethargy, Stereotypy, Hyperactivity–Noncompliance, and Inappropriate Speech. An increase in behavioural difficulties compared to the past may signal the development of a mood disorder. A Lethargy scale high score may signal depression. An increase in hyperactivity may signal a manic episode. The ABC has excellent psychometric properties and has been used in numerous studies and is an excellent choice for patients with ASD. It can be used multiple times and is therefore useful in following a case for improvement or relapse.

Another useful instrument is the *Diagnostic Assessment for the Severely Handicapped-II (DASH-II)* [36]. The DASH is an "informant interview" scale. Because it does not rely on verbal self-report from the person being assessed, it is very useful for people with severe to profound ID and ASD. Items are rated using a 3-point scale for frequency, duration, and severity. There are 13 subscales and 7 that have direct relevance to psychiatric diagnoses: Anxiety, Depression, Mania, Schizophrenia, Stereotypies/Tics, Organic Syndromes, and Impulse Control Disorder/Miscellaneous. Additional subscales address behaviours of psychiatric importance: Self-Injurious Behaviour, Elimination Disorders, Eating Disorders, Sleep disorders, and Sexual Disorders. This instrument has been translated into Italian, Norwegian, and Spanish. The DASH-II has good psychometric properties.

An excellent instrument to assess depression, the *Mood, Interest & Pleasure Questionnaire (MIPQ),* was developed by Ross and Oliver [67, 68]. This instrument is used with people who have severe and profound ID. It is an informant rating scale with

two subscales: Mood and Interest & Pleasure, rated on a 5-point scale for being observed in the last 2 weeks. Reliability and internal consistency are good and validity was demonstrated with the ABC [65]. This instrument is useful for people with ID in moderate to profound range and also people with ASD.

The *Autism Comorbidity Interview Present and Lifetime Version* is an excellent diagnostic interview discussed above. It is used for children and young adults with ASD who may be suffering from a mood disorder [18].

On a simpler level, caretakers can track mood throughout the day over a period of time, looking for patters. The *Sovner-Hurley Bipolar Mood Tracking Sheet* [69] documents mood changes kept daily by a caregiver. The mood is rated daily so patterns may emerge linking mood to events or days, or to ascertain that a cyclic pattern is developing. Mood is rated 0 to +3 for activation (mania) and 0 to −3 scale for withdrawal daily. A similar tracking sheet, the *Sovner-Hurley Monthly Sleep Chart* [70], tracks sleep through hourly blocks. If there is no one awake at night, the chart can still be used if caretakers become aware at any time that an individual is up and active at night.

There are a number of tools that can be used to investigate the presence of a mood disorder in persons with ID. A good practice before starting the assessment should be to find medical and school records, and evaluate stress and sensory sensitivity.

21.7 Treatment

21.7.1 Depression

21.7.1.1 Psychotherapeutic Approaches

Treatment for depression in people with ID/ASD should ideally be multifaceted. It is ideal to involve the person's full circle of relationships: school, day program, family, teachers, direct support professionals, and psychologists. Medication alone is often not enough.

Increasing supports For every person with ID and ASD, the plan should begin with an increase in supports. The suffering of depression can be ameliorated by therapeutic approaches such as increasing social contact, pleasurable activities, and positive feedback. For people with ID and ASD, supports should be organized and discussed with the person's entire support team. The patient will improve most quickly if the environment and support staff can change to be as "happy" and supportive as possible. Simple interventions provide much promise. For example, offering favourite trips out, favourite foods, walks with some friends, and special music are very easy interventions to organize. Because people with ID and ASD are less able than neurotypical people to help themselves, encouraging a strong and organized support programme is essential. There are many psychotherapeutic approaches to depression, and cognitive behavioral therapy (CBT) is an excellent choice for people with ID. Patients who have bipolar disorder can also benefit from the same approaches, although active direction during a manic episode is seldom useful.

Psychotherapy There is not a large literature on psychotherapy for people with mild ID and ASD. However, there is no reason to question the same efficacy of the approaches to persons with ID and ASD who suffer from depression with as are seen in neurotypical people, modified for level of ID. CBT approaches are usually an excellent choice for patients who are within the mild to moderate range of ID. Psychotherapy should involve a great deal of education for the person and caregivers about depression itself and give hope for recovery. It is important to involve caregivers frequently in person or by telephone contact regarding active suggestions from psychotherapy sessions. For patients with ID and ASD who have bipolar disorder, when not in a manic episode, psychotherapy and supports should be organized as well.

McGillivray and her colleagues [71, 72] demonstrated that people with Asperger's Disorder or High Functioning Autism could benefit from structured CBT in structured group sessions, easing depression. In another

study, people with mild ID and depression were treated with CBT techniques assisted by staff. The programme emphasized the concepts "Think Happy, Feel Happy, Be happy" and used group sessions and typical curriculums for problems associated with depression. People in the programme showed good improvement compared to wait-list controls. There is every reason to assume that CBT approaches could be modified to help people with ID and ASD.

Psychosocial Education Another interesting approach was used by Ingram [73] who intervened with staff using psychosocial education, targeting an individual in their care who had severe ID and severe challenging behaviour. The programme exposed staff to understanding the person's history, challenges, and in some sense, "reinterpreting" their assumptions about the person. The results of attending these workshops were remarkable with dramatic drops in all challenging behaviour (e.g. incidents of physical aggression per daily interval recording when from 19 to 0, and challenging behaviour from 140 to 18). Just like family, educating support caregivers is essential in helping people with ID and ASD to fit in better with home living and day or work programme arrangements.

In persons with ID and/or ASD the treatment of depression and bipolar disorder should be multifaceted and involve the person's full circle of relationships. CBT modified approaches could be an excellent choice for people who are within mild to moderate range of ID. Providing everyone with organized support programme and psychosocial education is also very important.

21.7.1.2 Pharmacotherapy

The literature on psychopharmacotherapy for patients with ASD and mood disorder is almost exclusively geared either to patients with high-functioning autism or to the treatment of behavioural symptoms, as reviewed by Postorino and his colleagues [74], and is therefore not a guide to diagnosis-based treatment in patients with ID/ASD. There is minimal case report literature, almost all single cases; clinical experience is extensive but not reported, since patients in this population rarely meet diagnostic criteria designed for persons with more communicative ability. The pharmacotherapy of both depressive and bipolar disorders is well-known and well-established for neurotypical patients with these disorders, so that while there is no literature support for use of any particular agent, neither is there any support for a belief that they will be ineffective, and clinical experience has shown that, in fact, they are. This discussion will focus on the special considerations particular to patients with these disorders and ID/ASD.

A recent study on older persons with ID from the Swedish national registers indicates a limited use of antidepressants compared to that of other psychoactive drugs, which seems to be related to the tendency of clinicians to under-diagnose depressive disorders in favour of other psychopathological conditions, especially in the presence of problem behaviours [75]. Psychiatrists should consider antidepressants as an option in treating psychiatric disorders rather than routinely prescribe drugs with sedative effects.

Patients with ID/ASD may have quite atypical, and, as of this writing, frequently unknown atypical, neurochemistry. Responses to psychopharmaceuticals are therefore not entirely predictable, and caution should be exercised in dosing. Dosing decisions should not be based on the severity of symptom-associated behaviour, but rather based on consideration of the brain that is producing the symptoms, and by clinical response. The rubric "Start low, go slow" is good advice.

Patients with ASD, in whatever range of cognition they may function, almost always suffer from anxiety, and many have problematic gastrointestinal symptoms. Unlike older (tricyclic) antidepressants, specific serotonin reuptake inhibitors (SSRIs) offer both antidepressant and anti-anxiety effects and have (usually) minimal gastrointestinal effects, making them the logical first choice for pharmacotherapy of depression in this population. All SSRIs take 4–6 weeks for their full effects to unfold; dose increases should therefore take place at 1-month intervals,

with monitoring for activation and increased anxiety. Neuromodulation via SSRIs has also been suggested for the treatment of premenstrual dysphoric disorder, together with ovulation suppression through various contraceptive and hormonal preparations. Unlike traditional depression therapy, the treatment of premenstrual dysphoric disorder does not require SSRIs to be used every day; they can be used cyclically, only during the luteal phase, or also for the duration of the monthly symptoms) [76, 77].

Serotonin/noreprinephrine reuptake inhibitors (SNRIs) may also be effective, but affect two neurotransmitters, making assessment of possible side effects problematic, and perhaps more likely than with SSRIs.

It has long been thought that antidepressant medication may be tapered a year after remission in first episodes, with a 50% recurrence rate in neurotypical populations, a year after remission after a second episode with a 90% recurrence rate; after a third episode recurrence rate after cessation of antidepressant medication is 100%. The recurrence rate in persons with ID, ASDs, and ID/ASD is unknown. Further, one is often unable to determine if a patient has had previous episodes. Tapering of effective antidepressant pharmacotherapy is therefore, unfortunately, very problematic in this population.

21.7.1.3 Mania and Mixed Mood Episodes

Mood stabilizing medications are numerous. The antiepileptic mood stabilizers (valproate, carbamazepine, oxycarbazepine, topiramate, and sometimes lamotrigine) can offer advantages in a population with a high rate of comorbid seizure disorder. Lithium can be difficult to manage in this population; individuals with ID/ASD may have unreliable fluid intake (especially in regimented settings, where they may not have free access to fluids), presenting risk of Lithium toxicity.

All the antiepileptic mood stabilizers, and Lithium, require blood monitoring of various kinds to detect and avoid possible toxicity, which can present problems in patients averse to venipuncture. Supportive measures to reduce anxiety until venipuncture becomes routine may be necessary. This is especially necessary in a patient population which may lack the ability to report side effects. Toxicity may not be apparent until observed by carers in patients lacking the ability to communicate internal experiences. Lamotrigine does not require blood monitoring, but presents the risk of the potentially fatal Stevens-Johnson Syndrome (SJS) in a person who may lack the verbal ability or sensitivity to pain to report the first signs of rash and mucous membrane involvement.

Antipsychotic medications may have to be added to mood stabilizers as adjuncts, or used as mood stabilizers, if the antiepileptic medications and Lithium are ineffective or poorly tolerated. However, these medications present the well-known disadvantages of appetite increase and weight gain, insulin-resistance and hyperlipidemia, in a population already burdened with food rituals and obsessive food preferences, plus the risk of movement disorders. They should not be the medications of first choice to treat mania.

21.7.1.4 Light Therapy

Seasonal affective disorder may be fairly common in the general population and should be considered when helping people with ID/ASD. Changes in a person's behaviour during the fall and remitting in the spring are key symptoms. For seasonal affective disorder, the use of a qualified Bright Light Box can be very effective [78]. Typically, the Bright Light Box is used in the morning daily from late fall and winter months for 30 minutes to an hour. The person sits near it but does not look directly at the light box. Most bright light boxes deliver 10,000 LUX. For people with ID and/or ASD, caregivers typically will be in charge of turning on the box, supporting the person personally while in use, and in limiting time with the light.

Several reports address the use of Bright Light Therapy for people with ID/ASD. Hermans and colleagues [79] treated 14 patients with ID/ASD for depression using Bright Light Therapy. All patients were rated on the depressive mood subscale of the ADAMS (Anxiety, Depression and Mood Scale) [80]. Subjects' levels of ID were: 3 mod-

erate; 3 severe; and 8 profound. Subjects were treated for 2 weeks sitting near the light box for 30–60 minutes. Considerable accommodations were needed to engage the individuals due to their schedules, medical needs, and some difficulty sitting near the light box for a long time. However, most participants showed reduction in symptoms of depression, and six showed remarkable change.

Altabet and colleagues [81] described three people with profound intellectual disability who exhibited signs of depression. Each patient was treated in a special room and a Bright Light Box using 10,000 LUX. Patients were supervised to sit a bit away at an angle to not look at the light. Measurements taken included: ABC Irritability and Lethargy Scale; the DASH depression scale; a sleep record; and a mood chart. Patients were females age 42, female age 61, and female age 68. All improved significantly in standard measures. In all three cases, decreases in depression, irritability, or lethargy attained during treatment remained stable for 3 weeks, but continued to decrease after 8 weeks.

Cooke and Thompson [82] treated two patients with ID using Bright Light Therapy. These patients manifested seasonal cycles in mood and behaviour. The first patient in adolescence displayed significant periods of overactivity, elation, and for example, dancing for hours followed by a period of withdrawal, loss of interest. He was diagnosed with bipolar disorder and treated with medications including lithium. At the age of 40, his depression became more frequent and occurred in winter. He was treated with 10,000 LUX lamp. He became very stable, did not become depressed that winter, and was subsequently maintained on Lithium. The second patient was a 9-year-old boy with severe ID/ASD. At the age of 6, he had very difficult behaviours, which seemed to have a cyclical presentation. At the beginning of October, he was treated with 10,000 LUX lamp. He appeared happier, was much less anxious, and did not have problems of aggression or hyperactivity. Unfortunately, sleep itself did not improve. No structured assessments were used.

Tsiouris [83] used light therapy for four patients. He chose 5000 LUX to assure that there would not be light too intense for patients. One patient had Down syndrome and a seasonal pattern of depression in the fall and winter months. His parents administered the Bright Light Therapy at home which was successful and his fluvoxamine was reduced. The second patient was a female with severe ID, a seizure disorder, traumatic brain injury, and major depression. In childhood, she had a right-side lobectomy for severe aggression. Psychiatric medications alone did not help her depression, which seemed worse in winter months. Bright Light Therapy was used for 30 minutes in the morning. Within 1 week, all symptoms of depression and challenging behaviour were reduced and eliminated. Thereafter, she began Bright Light Therapy each September. The third patient was 45 years old and had severe ID/ASD, Tourette disorder, and was also diagnosed with bipolar II disorder. Her symptoms of depression were reversed with Bright Light Therapy conducted during the winter months. The last patient was a 44-year-old female with ASD, spastic diplegia, and rapid cycling bipolar II disorder. After light therapy, she was much less depressed and treatment continued from September through April annually.

21.7.1.5 Electroconvulsive Therapy (ECT)

It is impossible to review treatment of mood disorders without mention of ECT. There are several reviews of the use of ECT in persons with ID [84–86], and many case reports of its use with patients with ID and mood disorders [87] and many of its use in patients with ASDs and a variety of behavioural issues and diagnoses [88]. Only two case studies of patients with ID/ASD and a mood disorder could be located. Wachtel and colleagues [88] and Siegel and colleagues [89] both describe single cases of patients with ID/ASD and cycling mood disorders refractory to medication and responsive to ECT. There are no controlled studies, and patients all remained on their existing medication regimens to which ECT was added.

Several of these studies, notably Reinblatt and colleagues [84], refer to the difficulties in obtaining informed consent in this popula-

tion, without detailing the reasons. Beyond the obvious issues presented by cognitive limitations, one must consider the scepticism among the general public about ECT, magnified by the scepticism in many parents and guardians of persons with ASDs about psychiatric medication.

Anxiety is bound to be an issue in experiencing ECT for persons who become anxious at far less anxiety-producing medical procedures. The known effects of ECT on memory and cognition may be equally anxiety-producing, and patients with limited or no language would be unable to communicate these adverse experiences. While ECT must remain an alternative where food, fluid, and medication refusal, medication intolerance or ineffectiveness, is an issue, there is insufficient evidence or experience to advocate it as a first-line approach to mood disorder in this population.

Psychiatrists should consider antidepressants as an option in treating psychiatric disorders rather than routinely prescribe drugs with sedative effects.

Patients with ID and/or ASD may have quite atypical neurochemistry, and therefore great caution is required in dosing antidepressant and mood stabilizers. SSRIs represent the first choice for moderate depressive episodes, while antiepileptic mood stabilizers can offer advantages to treat mania.

Several reports address the use of Bright Light Therapy to treat seasonal affective disorder and ECT as an alternative treatment in very specific conditions.

21.8 Prognosis

The prognosis for people with ID and ASD who suffer from mood disorders will be improved by diagnosing the correct condition. In the past, many individuals with ID and ASD were not diagnosed with psychiatric disorders and were treated only for having challenging behaviour. We must be sensitive to the mood of individuals we support and treat. Families and caregivers, who often speak for these individuals in appointments, must be educated so that they do not give prejudiced information. For example, in a staffed home, the carers may have little or no training in mental health issues. Thus, information is framed from a certain point of view, often as a behavioural problem. This makes it essential that formal assessment tools be used so that extensive background information and records are reviewed when we see a patient for the first time. Understanding the person's mood state, psychiatric symptoms, and medical issues is essential [90].

Depression must be treated with kindness and consideration. As previously suggested, family and caregivers can help by increasing pleasurable activities. Just as we might treat someone suffering from cancer with soothing and supportive caring, we must see depression as a serious illness that can be helped by positive environments. Bipolar disorder must be carefully followed and monitored for switches in mood. Medications must be closely monitored with timely laboratory information.

Bipolar disorder requires medication, perhaps multiple medications, and possible hospitalizations. The most effective mediations require laboratory tests, which may be a problem for some individuals. It is important to organize ways to make these tests go smoothly if patients are afraid or object. Sometimes, simple anaesthetic ointment and support may suffice. Ideally, patients with bipolar disorder will have a stable team of support staff, psychotherapists, psychiatrists, and primary care clinicians to care for them.

As of this writing there are no extant studies of the prognosis for treatment of mood disorders in persons with ID/ASD. It is only relatively recently that it was recognized that these disorders could be diagnosed and treated in these populations; studies of prognosis were confined to treatment of behaviours, not comorbid disorders. Case report literature (cited above) is replete with reports of successful treatment of depressive episodes and manic episodes, but studies of long

term antimanic and antidepressant prophylaxis are lacking. If it is ever done, comparison would have to be made with such studies in neurotypical patients with these disorders, with the provision that no expectation should be held that mood stabilization will improve the underlying ASD or ID, only the mood disorder.

Clinical experience so far suggests that prognosis will be comparable. Individual episodes of mania and depression are responsive to antimanic and antidepressant medication much as they are in the neuroptypical population. Recurrence of depression or of manic cycling can be expected if medication is not maintained. The same issues of long-term medication tolerance are encountered as in the neurotypical population (e.g. bone marrow effects, hyponatremia with carbamazepine and valproate; nephrogenic diabetes insipidus and hypothyroidism with Lithium). Breakthrough episodes, especially of bipolar disorder, can be expected to occur, with the necessity for adjunctive medication so familiar in the treatment of neurotypical patients with mood disorders. Whether or not these are more frequent in persons with ASD and ID is at present unknown, but any studies would have to take into account the fact that most of these patients, unlike neurotypical patients with mood disorders, will be in the care of systems that ensure their compliance with their medication regimen, in contrast with the noncompliance often encountered in neurotypical patients. Selection of comparison groups for such studies, and interpretation of results, may prove difficult.

For the moment, lacking any information to the contrary, we must treat with the agents that we have or that become available and do what is clinically indicated to maintain mood stability, much as is done with neurotypical patients with mood disorders.

The prognosis for people with ID and/or ASD who suffer from mood disorders will be improved by diagnosing the correct condition.

Studies of long-term antimanic and antidepressant prophylaxes are lacking, but recurrence of mood disorders can be expected if medication is not maintained.

Tip

Mood disorders in people with ID and ASD deserve increasing research interest and continued acquisition of up-to-date knowledge by clinicians.

Key Points

- Depression and bipolar disorder are common psychiatric disorders in persons with ID and/or ASD, even if they are not properly recognized.
- For individuals with ID and ASD proper diagnosis and effective treatment of mood disorders present considerable challenges.
- In persons with ID and/or ASD changes in sleep pattern, appetite, body weight, and participation in daily activities can represent symptoms of mood disorders.
- Reduction in speech, social withdrawal, and increase in somatic complaints occur more frequently in depression, while psychomotor agitation, decreased sleep, changes in mood, and aggression occur in mania.
- Causes of depression include genetic predisposition, major life changes, trauma, increase in stress, major loss of support or friends, physical illness, disability, and medications, while bipolar disorder seems to be more associated with genetic inheritance and medical conditions.
- In persons with ID and/or ASD the treatment of depression and bipolar disorder should be multifaceted, com-

bining psychotherapy and medications, and including organized support programme and psychosocial education.
- Specific serotonin reuptake inhibitors (SSRIs) represent the first choice for moderate depressive episodes, while antiepileptic mood stabilizers can offer advantages to treat mania.
- In persons with ID and/or ASD the prognosis of mood disorders can be significantly improved by diagnosing the correct condition. Evidence on long-term antimanic and antidepressant prophylaxes are lacking, but recurrence of mood disorders can be expected if medication is withdrawn.

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Anxiety and Stress-Related Disorders in People with Intellectual Disability/Disorders of Intellectual Development

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Contents

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Learning Points

- Anxiety disorders and related disorders are common in persons with intellectual disability/disorders of intellectual development (ID/DID) and frequently missed due to inherent limitations in cognitive function and communication difficulties, described elsewhere in this book.
- The clinical utility of the classificatory systems such as the ICD and the DSM could be adapted and modified as relevant by placing equal emphasis on reliably observed components (signs) as well as symptoms (reported).
- In those with more severe ID/DID, anxiety disorders may present as problem behaviours.
- Accurate diagnosis underpins access to appropriate interventions maximizing optimum quality of life.
- Management includes psychological and pharmacological treatment adapted or extrapolated from the general population since there is overall a dearth of empirical evidence specific to persons with ID/DID.

22.1 Introduction

Anxiety is a ubiquitous emotional state that lies on a continuum between fear-related responses and those characterized by a diffuse sense of dread, unease, and worry/misery. Fear-related anxiety affects cognitive, emotional, and behavioural responses to danger or imminent threat. In fear, the cues are usually external in most anxiety states; they eventually generalize and respond to learned (conditioned) cues. Fear also manifests as freezing and flight (escape and avoidance) or fight behaviours (defensive aggression). These states involve activation of the sympathetic nervous, endocrine (increased cortisol) and neuro-immunological (cytokine release) components of the stress response system [1, 2].

Non-fear-related anxiety arises in response to anticipated threat, worry, and other negative affective states. Physiological arousal and anxiety-related behaviours resemble scaled-down versions of fear-related responses to non-life threatening, diffuse threats. Panic disorder (fear-related anxiety) frequently spawns anticipatory and avoidance behaviours (agoraphobia) to multiple layers of generalized fear-conditioned stimuli. Many of these responses bear only a faint resemblance to the original fear experiences. In summary, anxiety lies along a spectrum of negative affective states that range from acute fear to worries about illness, physical appearance, social acceptance or what might happen in the future.

Anxiety disorders (ADs) can be roughly divided into two overlapping domains: (1) fear-related anxiety which includes phobias, panic disorder (with/without agoraphobia), trauma-related disorder, and social anxiety disorder (performance anxiety) and secondary forms (traumatic brain injury, seizure with fear at onset). Each involves variations on fear response, fear conditioning, avoidance behaviours, and risk for excessive generalization, transformation, and problems with extinction. (2) The second subtype involves excessive worry/distress-related anxiety disorder that includes social anxiety (generalization of social avoidance), generalized anxiety disorder, and related mood-anxiety disorder and dysthymia. In each of these syndromes, the threat is more diffuse, and behavioural responses are mediated by higher levels of cognitive processes, cortical modulation, and executive functions [3–6].

ADs are defined in the DSM-5 [7] as "The apprehensive anticipation of future danger or misfortune accompanied by a feeling of worry, distress, and/or somatic symptoms of tension. The focus of anticipated danger may be internal or external". Anxiety refers to the "anticipation" of future threat while fear is "the emotional response to real or perceived imminent threat". Worry is defined as "apprehensive expectation". The DSM-5 also includes severity as a qualifier for diagnosis in that the anxiety must be of sufficient degree to interfere significantly with the person's social, occupational, or global functioning. In ADs,

the intensity or duration of anxiety is disproportionate to the potential for harm, or in the absence of recognizable threat to the individual. It involves increased levels of arousal, resulting in disorganizing rather than facilitating an individual's performance.

Persons with intellectual disability/disorders of intellectual development (PWID/DID) are a heterogeneous group, which share significant cognitive and communication impairments with physical as well as mental comorbidities and poor social functioning in varying degrees. Consequently, in common with all other psychiatric disorders, ADs are often not diagnosed in this vulnerable population, resulting in poor access to appropriate treatment which further limits quality of life.

Overall, there is a paucity of high-quality evidence specific to AD as is the case in all mental disorders in PWID/DID due to difficulties in carrying out good quality research [8]. Given these limitations, evidence derived from empirical studies in the mainstream population has been adapted, extrapolated, or modified within the context of the individual's psychological, biological, and social circumstances.

This chapter provides an overview of anxiety and related disorders including, generalized anxiety disorder, panic disorder, agoraphobia, specific phobia, social anxiety disorder, separation anxiety disorder, selective mutism, health-related anxiety, and post-traumatic stress disorder (PTSD) as well as obsessive-compulsive disorder (OCD) categorized separately in DSM-5. It covers classification and diagnosis, clinical features, epidemiology, aetiology, pathophysiology, risk factors, treatment, and prognosis within the context of available evidence.

Anxiety disorders refer to negative emotional states ranging from acute fear to worry about something that significantly interferes with the person's global functioning. In persons with ID/DID, anxiety disorders are often not diagnosed, resulting in poor access to appropriate treatment which diminishes quality of life.

22.2 Classification and Diagnosis

The DSM-5 [7] identifies eleven disorders in the anxiety category all of which use the term fear, worry, and anxiety; the exception being selective mutism. It also separates OCD from AD. The ICD-11 [9] describes anxiety and fear-related disorders as being "characterized by excessive fear and anxiety and related behavioural disturbances, with symptoms that are severe enough to result in significant distress or significant impairment in personal, family, social, educational, occupational, or other important areas of functioning. A key differentiating feature amongst the anxiety and fear-related disorders is disorder-specific foci of apprehension, that is, the stimulus or situation that triggers the fear or anxiety. The clinical presentation of anxiety and fear-related disorders typically includes specific associated cognitions that can assist in differentiating amongst the disorders by clarifying the focus of apprehension".

Kogan et al. [10] have outlined the changes in the organization and diagnostic guidelines for anxiety and fear-related disorders proposed by the ICD-11 Working Group on the Classification of Mood and Anxiety Disorders and the rationale and evidence base for the proposals. The ICD-11 [9] contains nine types of conditions defined as follows- (substance/medication-induced AD and anxiety due to another medical condition categorized in DSM-5 have been excluded).

1. Generalized Anxiety Disorder (GAD)

GAD is characterized by marked symptoms of anxiety of least several months duration, for more days than not, manifested by either general apprehension (i.e. "free-floating anxiety") or excessive worry focused on multiple everyday events, most often concerning family, health, finances, and school or work, together with additional symptoms such as muscular tension or motor restlessness, sympathetic autonomic overactivity, subjective experience of nervousness, difficulty maintaining concentration, irritability,

or sleep disturbance. The symptoms result in significant distress or significant impairment in personal, family, social, educational, occupational, or other important areas of functioning.

2. Panic Disorder

Panic disorder is characterized by recurrent unexpected panic attacks that are not restricted to particular stimuli or situations. Panic attacks consist of discrete episodes of intense fear or apprehension accompanied by the rapid and concurrent onset of several characteristic symptoms (e.g. palpitations or increased heart rate, sweating, trembling, shortness of breath, chest pain, dizziness or lightheadedness, chills, hot flushes, fear of imminent death). In addition, panic disorder is characterized by persistent concern about the recurrence or significance of panic attacks, or behaviours intended to avoid their recurrence, that results in significant impairment in personal, family, social, educational, occupational, or other important areas of functioning.

3. Agoraphobia

Agoraphobia is described as marked and excessive fear or anxiety that occurs in response to multiple situations where escape might be difficult or help might not be available, such as using public transportation, being in crowds, being outside the home alone (e.g. in shops, theatres, standing in line). The individual is consistently anxious about these situations due to a fear of specific negative outcomes (e.g. panic attacks, other incapacitating or embarrassing physical symptoms). The situations are actively avoided, entered only under specific circumstances such as in the presence of a trusted companion, or endured with intense fear or anxiety. The symptoms persist for at least several months and are sufficiently severe to result in significant distress or significant impairment in personal, family, social, educational, occupational, or other important areas of functioning [9].

4. Specific Phobia

Specific phobia is characterized by a marked and excessive fear or anxiety that consistently occurs when exposed to one or more specific objects or situations (e.g. proximity to certain animals, flying, heights, closed spaces, sight of blood or injury) and that is out of proportion to actual danger. The phobic objects or situations are avoided or else endured with intense fear or anxiety. Symptoms persist for at least several months and are sufficiently severe to result in significant distress or significant impairment in personal, family, social, educational, occupational, or other important areas of functioning (simple phobia, claustrophobia). Approximately, 75% of individuals diagnosed with specific phobia fear more than one object. When this occurs, more than one diagnosis is given.

5. Social Anxiety Disorder

Social anxiety disorder signifies marked and excessive fear or anxiety that consistently occurs in one or more social situations such as social interactions (e.g. having a conversation), being observed (e.g. eating or drinking), or performing in front of others (e.g. giving a speech). The individual is concerned that he or she will act in a way, or show anxiety symptoms, that will be negatively evaluated by others. The social situations are consistently avoided or else endured with intense fear or anxiety. The symptoms persist for at least several months and are sufficiently severe to result in significant distress or significant impairment in personal, family, social, educational, occupational, or other important areas of functioning.

6. Separation Anxiety Disorder

Separation anxiety disorder is characterized by marked and excessive fear or anxiety about separation from specific attachment figures. In children, separation anxiety typically focuses on caregivers, parents, or other family members; in adults, it is typically a romantic partner or children. Manifestations of separation anxiety may include thoughts of harm or

untoward events befalling the attachment figure, reluctance to go to school or work, recurrent excessive distress upon separation, reluctance or refusal to sleep away from the attachment figure, and recurrent nightmares about separation. The symptoms persist for at least several months and are sufficiently severe to result in significant distress or significant impairment in personal, family, social, educational, occupational, or other important areas of function.

7. Selective Mutism

Selective mutism manifests as a consistent failure to speak in specific social situations in which there is an expectation for speaking (e.g. at school or work) despite speaking in other situations. The disturbance interferes with educational or occupational achievement or with social communication, and the duration of the disturbance is at least 1 month. The failure to speak is not due to the lack of knowledge of, or comfort with, the spoken language required in the social situation. The disturbance is not better accounted for by a communication disorder (e.g. childhood-onset fluency disorder) and does not occur exclusively during the course of autism spectrum disorder, schizophrenia, or another psychotic disorder [7].

8. Other Specified Anxiety or Fear-Related Disorders

Despite not fulfilling all the requisite criteria, for a particular anxiety disorder, if the anxiety symptoms cause significant distress or impairment, the condition may be noted under this category.

9. Anxiety or Fear-Related Disorders, Unspecified

This category is used when anxiety-like symptoms that cause significant distress or impaired functioning is manifest with insufficient information to determine the particular type of anxiety disorder. For example, emergency situations, where a complete history and full psychiatric evaluation are not always feasible.

The DSM-5 [7] classifies ADs into 11 sub-categories, broadly similar to the above with the additional inclusion of substance/medication-induced anxiety disorder and anxiety disorder due to another medical condition. Panic disorder also has a panic attack specifier.

■ Obsessive-Compulsive Disorder (OCD)

NB: OCD is separate from ADs and categorized under obsessive-compulsive and related disorders in DSM-5 [7] and ICD-11 [9].

OCD is characterized by uncontrollable, recurring thoughts (obsessions) and behaviours (compulsions) that the person feels the urge to repeat over and over again which tends to follow a chronic course. Commonly occurring obsessions include fear of contamination, need for order or symmetry, obsessive doubts, excessive conscientiousness (need to do the right thing, fear of committing a transgression, often religious), and unwanted, intrusive sexual/aggressive thoughts. Commonly occurring compulsions consist of hoarding, arranging, and rearranging objects, reassurance seeking, list making, cleaning/washing rituals, counting/repeating actions a certain number of times or until it "feels right" and checking (e.g. locks, cooker, safety of family members).

OCD is a heterogeneous condition [11] and generally follows a chronic long-lasting course, with peaks and troughs, having significant comorbidity with other mental disorders with major depression, anxiety, as well as tic disorder [12, 13]. Lifetime and 12-month prevalence estimates for DSM-IV OCD are reported as 2.3% (0.3) and 1.2% (0.3) [14]. The differential diagnosis of OCD includes obsessive-compulsive personality disorder (OCPD). OCPD typically emerges in early adulthood and refers to individuals who are preoccupied with orderliness, perfectionism, and mental and interpersonal control, at the expense of flexibility, openness, and efficiency.

A small study carried out on patients with mild to profound ID/DID by Vitiello

[15], reported that 3.5% of the sample presented with compulsive behaviour that significantly interfered with their daily functioning. Compulsions occurred in the context of obvious cerebral dysfunction and in the absence of anxiety or "ego-dystonic" qualities. They concluded that the emphasis should be on the behavioural, externally observable components of the disorder rather than the cognitive elements of inner conflicts and anxiety.

OCD also occurs as behavioural phenotypes of specific disorders in PWID/DID such as Down syndrome [16] and Prader-Willi syndrome [17]. The presence of compulsions is helpful in distinguishing this disorder from other ADs as well as depression.

Post-traumatic Stress Disorder (PTSD)

NB: PTSD is categorized separately from ADs in DSM-5 [7], coming under trauma- and stressor-related disorders in DSM-5. The upcoming ICD-11 [9] proposes the inclusion of an additional category of complex post-traumatic stress disorder (CPTSD). This is a condition that results from chronic or long-term exposure to emotional trauma over which a victim has little or no control and from which there is little or no hope of escape, such as in cases of: domestic emotional, physical or sexual abuse, long-term imprisonment and torture, exposure to crisis conditions, etc. [18].

PTSD involves a history of exposure to a traumatic event that results in symptoms from each of four symptom clusters, namely, intrusion, avoidance, negative alterations in cognitions and mood, and alterations in arousal and reactivity. Other requisites for a diagnosis concern symptom duration and non-attribution of symptoms to a substance or comorbid medical condition. The individual initially responds with intense fear, helplessness, or horror to the traumatic exposure, later developing a response characterized by persistently re-experiencing the event, with resultant symptoms of numbness, avoidance, and hyperarousal causing clinically significant distress or functional impairment [19]. Although a substantial minority of PTSD cases remits within months after onset, mean symptom duration is considerably longer than previously recognized [20].

People with disorders of intellectual development (PWDID) appear to have a predisposition to develop PTSD [21] but with differences in manifestation when compared with the general population. These may consist of the expression of symptoms, and the interpretation of distressing experiences, as the manifestation of possible PTSD seems to vary with the level of severity of ID.

Although there is evidence to suggest that PWID/DID are likely to suffer from PTSD, reviews of the evidence base and the potential consequences of this contention are sparse or absent. Some support was found for the notion that PWID/DID have a predisposition to the development of PTSD. Differences in comparison with the general population may consist of the expression of symptoms, and the interpretation of distressing experiences, as the manifestation of possible PTSD seems to vary with the level of ID. In the absence of reliable and valid instruments for assessing PTSD in this, there is no prevalence data on PTSD amongst PWID/DID while Nine papers described treatment of PTSD in people with ID/DID. Interventions reported involve those aimed to establish environmental change, the use of medication, and psychological treatments (i.e. cognitive-behavioural therapy, Eye Movement Desensitization and Reprocessing - EMDR, and psychodynamic-based treatments). Case reports suggest positive treatment effects for various treatment methods. Development of diagnostic instruments for assessment of PTSD symptomatology in this population is required, as it could facilitate further research on its prevalence and treatment.

The DSM-5 classifies anxiety disorders into 11 sub-categories. All disorders are characterized by the presence of excessive fear, worry, and behavioural disturbances for at least several months, sufficiently severe to result in significant distress or impairment in personal, family, social,

educational, occupational, or other important areas of functioning. OCD and PTSD are separated from anxiety disorders, and both conditions occur frequently in persons with ID/DID causing alterations in arousal and behavioural changes.

22.2.1 ICD/DSM Diagnostic Criteria for Anxiety Disorders and PWID/DID

PWID/DID constitute a significantly heterogeneous population where chronological age and developmental level (mental age) are by definition discrepant. The phenomenon described by Reiss [22] as diagnostic overshadowing, where clinicians overlook symptoms of mental illness attributing them to ID/DID, may also contribute to difficulties experienced by clinicians in diagnosing ADs in this population. The presence of a co-occurring autism spectrum disorder (ASD), which is rather frequent in persons with more severe ID, further complicates the situation, compromising traditional diagnostic techniques and processes [23]. When applying ICD or DSM diagnostic criteria for ADs to individuals with PWID/DID, efforts must be made to seek the presence of signs and symptoms using developmentally appropriate methods of investigation and appropriate adaptations that take note of the different levels of ID [24, 25]. Such methods should include the ascertainment of "signs" of disorder that are observable, or those that are reported by parents or others familiar with the person.

All the criteria for ADs in ICD/DSM can be applied validly and reliably in people with mild-to-moderate PWDID [24–27]. A study by Sternlicht [28] on children with moderate ID/DID suggested that the developmental trend of fears that appear in normal children appears in PWID/DID as well, and these fears follow Piaget's level of cognitive development, proceeding from egocentric perceptions of causality to realistic cause and effect thinking. People with more severe degrees of ID/DID experience difficulties in articulating key diagnostic concepts of anxiety particularly with relation to more complex subjective cognitive phenomena and may manifest as problem behaviours [29–31]. However, behaviours may also be a reflection of other triggers in the environment, and it is therefore important to distinguish behaviours due to ADs from behaviours due to other causes. In diagnosing anxiety, Khreim and Mikkelson [32] emphasized the relevance of phenomena such as agitation, screaming, crying, withdrawal, regressive/clingy behaviour, or freezing, which could be interpreted as manifestations of fear in PWID/DID.

In persons with severe ID/DID and/or ASD, diagnosis is therefore reliant on caregiver feedback or completion of questionnaires. Caregivers are asked to interpret the behaviour of the person with ID/DID and/or ASD and accurately report this back. However, there are research findings on caregivers often failing to identify physiological markers in those they care for, making reporting anxiety-related behaviours and therefore diagnosis difficult [33]. The most reliable diagnostic process has been reported to be multimodal; that is, multiple informants, multiple assessment techniques (clinical interview, observation, physiological assessment), in addition to ID/DID and/or ASD appropriate standardized measurement tools [34]. A number of guidelines and diagnostic tools consisting of informant and self-report assessment instruments of psychopathology have been developed specifically for use in PWID/DID involving the DSM/ICD classificatory systems [35, 36]. These include the Psychopathology Instrument for Mentally Retarded Adults (PIMRA) [37] and Diagnostic Assessment for the Severely Handicapped (DASH) [38]. Scale, both based on the DSM-III-R [39] and ICD-10 [40]; the Anxiety, Depression, and Mood Scale (ADAMS) [41] developed using DSM-IV-TR criteria [42]; and the Psychiatric Assessment Schedule for Adults with Developmental Disability (PAS–ADD) [43]. Masi and colleagues [26] evaluated the concurrent validity of different informant and self-report assessment instruments of

psychopathology based on DSM-IV criteria (both general and specific for anxiety and/or depression), in adolescents with ID/DID, and concluded that anxiety measures were positively correlated with PIMRA [37] and Child Behavior Checklist (CBCL) [38] total scores, as well as with the internalizing score of the CBCL. However, the reliability of these instruments was found to be variable. Costello [44] reported that the anxiety element in the PAS-ADD instrument had poorer reliability when compared with the rest of the mental health disorders.

Comprehensive and helpful guidelines have been published in the Diagnostic Manual – Intellectual Disability (DM-ID) [25] and the Diagnostic Criteria for Psychiatric Disorders for use with adults with Learning Disability (DC-LD) [45] to facilitate the diagnostic process in this population. The latter renamed as DC-DID (Diagnostic Criteria for Psychiatric Disorders for use with adults with Disorders of Intellectual Development) is being revised updated within the context of the upcoming ICD-11 [9].

The Glasgow Anxiety Scale for People with an Intellectual Disability (GAS-ID) [46] appears to offer a psychometrically robust and pragmatic (5–10 min) approach to the appraisal of anxiety in people with mild ID/DID.

In the absence of robust empirical evidence, the clinical consensus supports the view that in PWID/DID functioning at lower moderate, severe, and profound levels, the likelihood of eliciting the cognitive elements of ADs is unlikely. Instead, alternative adaptations to the DSM-5 Criteria placing greater emphasis on reported symptoms have been proposed [47].

> The identification of anxiety disorders in people with more severe degrees of ID/DID is particularly difficult and frequently missed by carers and clinicians. Adaptations of the current classification systems (DM-ID, DC-LD) and other psychopathology instruments (PIMRA, DASH, ADAMS, PAS-ADD, CBCL, GAS-ID) could facilitate the diagnostic process in this population.

22.3 Comorbidity

ADs exhibit high levels of lifetime comorbidity with one another to such an extent that it is unlikely for an individual to experience a single anxiety disorder in isolation [48]. They are known to commonly coexist in individuals, both with other ADs and with mental disorders from other groupings, such as affective disorders [49]. The prevalence of having a current comorbid mental disorder is estimated at 50%, with ADs and depressive disorders being the most prevalent comorbid disorders [50, 51]. A population study based on three Swedish national registers over 11 years found that amongst older persons with ID, those with at least one anxiety diagnosis had an occurrence of psychiatric comorbidities approximately 11 times higher than the general population, and the most common comorbidities were specific personality disorders, unspecified nonorganic psychosis, and other mental disorders due to brain damage and dysfunction and to physical disease [52].

Whereas anxiety/anxiety comorbidity occurs earlier in onset and follows a more chronic course, anxiety/depressive comorbidity results in greater functional impairment, even more severity, and a poorer outcome [53]. From a clinical perspective, it is important to diagnose and treat comorbidity amongst ADs since it is associated with higher severity and greater chronicity.

22.4 Epidemiology

The World Health Organisation study of the Life Time prevalence of Mental Health Disorders in a global mental health survey concluded that ADs constitute the most prevalent of mental disorders in the general population [54] and have a significant impact on morbidity, mortality, and global burden of disease [55]. Occurring early in onset, they tend to be chronic conditions with high social and economic costs [55, 56]. Despite the availability of effective psychological and pharmacological treatments, a large proportion of affected individuals do not contact health services, remaining undiagnosed or do not receive correct

treatment [57]. According to large scale epidemiological surveys, ADs are the most prevalent of the psychiatric disorders with a lifetime morbidity risk of 41.7% [58, 59]. Around 80% of consultations for mental health problems in primary care are for anxiety and depression with the highest prevalence occurring during midlife [60]. ADs are more common in women [61]. Prevalence rates have remained unchanged in the past years with variations found in different countries and cultures possibly due to methodological differences rather than culture-specific factors. High comorbidity is found amongst the ADs and between the ADs and other mental disorders, respectively [62].

There have been numerous studies reporting that PWID/DID suffer from ADs [63] at rates at least equal to the general population [64, 65]. ADs have also been reported as one of the most common forms of psychological distress for persons with ID/DID and or ASD [64, 66] with a significantly higher prevalence rate than in typically developing (TD) populations [65]. Individuals with co-occurring ASD in particular are exposed to specific sources of anxiety such as changes in routine, anticipation of change, or environmental stimuli, which are hard to be diagnostically interpreted, as they describe both common experiences in ASD and ADs.

Due to communication and cognitive impairments, ADs are likely to be underreported, especially in persons with severe ID and/or co-occurring ASD [67]. Borthwick-Duffy [68] and Deb and collaborators [64] reported the prevalence of ADs amongst PWID/DID of around 5–10%. Bailey and Andrews [69] concluded that many studies fail to make a definitive diagnosis of AD and report only the prevalence of anxiety symptoms, which range from 6% [70] to 31% [71]. The point prevalence of ADs in PWID/DID is estimated to be 3.8% with GAD being most common (1.7%), followed by agoraphobia (0.7%) [72]. There is evidence to suggest that in persons with ID/IDD, older age, co-occurrence of ASD, and hearing impairments are associated with higher rates of GAD and other ADs [72, 73].

ADs were identified as more prevalent in individuals with ID/DID and co-occurring self-injurious behaviour than in those without such behaviour [43]. A study cohort comparing PWID/DID with the general population revealed significantly higher rates of phobic disorder in the former [64]. Hermans and colleagues [74] reported that older people with ID/DID reported more symptoms of anxiety when compared with older people with average intelligence.

The available evidence from studies carried out on children with ID/DID suggest high levels of ADs vary from 8.7% [75] to 21.98% [76]. The prevalence rate of ADs is also reported to vary according to the different subtypes of ADs [77]. A study by Catani and Sosalla [78] revealed that child abuse in the family to be the only significant independent predictor of PTSD symptom severity in this population of PWID/DID.

The correlation between rates of ADs and chronic physical health conditions has been found to be higher in persons with ID/IDD and ASD than in the general population [73], which might indicate a higher expression of the interconnected nature of mental and physical health.

Anxiety disorders are very common in persons with ID/DID and are often found in comorbidity with other psychiatric disorders, particularly mood disorders. The prevalence rate varies according to the different subtypes of anxiety disorders and depending on the background characteristics (age, level of DID) of the study sample.

22.5 Clinical Features

The clinical features of anxiety include cognitive, physiological, psychological, and behavioural elements. The cognitive components consist of irritability, fearful anticipation, concentration/memory problems, repetitive worrying thoughts, and fear, and when extreme, these may manifest as full-blown panic states. The physiological manifestations include flushing, sweating, pallor, dry mouth, difficulty in swallowing, palpitations, tremor, hyperventilation, pains/tightness in the chest, and other bodily aches and pains such as head/

backache, fatigue, muscle tension, diarrhoea, increased urinary frequency, paraesthesia, heightened startle response, and insomnia. Avoidance is a common behavioural manifestation of anxiety. Chronologically, anxiety can be episodic, continuous, or stress related.

In terms of how young people with mild ID/DID understand anxiety, Wilson and colleagues [79] reported that PWID/DID did not discuss their understanding of mental health issues in abstract terms but tended to use descriptive terms often linked to unpleasant emotional and physical sensation. They used words like "temper", "frightened", "fed up", and "things wrong to me" and described the physiological symptoms of anxiety and depression, such as sweaty palms and hyperventilation. Sometimes they used phrases that seemed to have been picked up from others, like "mood swings" and "in a huff".

While the DSM/ICD diagnostic criteria for anxiety and related disorders could be applied without adaptation to those with mild to moderate levels of impairment, there is no empirical evidence on their application to those with more severe degrees of impairment and/or co-occurrence of ASD [24, 46]. However, available research reports note that in these persons the manifestation of anxiety can be observed rather than subjectively reported and that cognitive symptoms of anxiety are less useful for diagnosing ADs than physiological symptoms [23]. Consequently, the clinical diagnosis relies on observable signs such as dry mouth, increased pulse and heart rate, pallor, flushing, facial expressions, crying, freezing, failure to speak in social occasions, as well as problem behaviours. The DM-ID 2 [25] provides succinct guidelines which elaborates these issues in greater detail.

Further peculiarities of clinical presentation with respect to individuals with ID/DID and ASD compared to those with ID/DID or ASD alone have been identified by research [80–84] and are reported in ◘ Table 22.1.

The clinical features of anxiety include cognitive, physiological, psychological, and behavioural elements. For people with more severe degrees of cognitive and communication impairment, manifestation of anxiety can be observed (i.e. pallor, flushing, facial expressions, crying, freezing, problem behaviours) rather than subjectively reported.

◘ **Table 22.1** Anxiety in ID/DID and ASD compared to ID/DID or ASD alone

Manifestation	ID/DID+ASD	ID/DID alone	ASD alone
Repetitive behaviours	More likely to be a result of anxiety than for sensory seeking purposes More likely during transitions between tasks	More likely to be for sensory seeking purposes than a result of anxiety Less likely during transitions between tasks	More likely to be a result of anxiety than for sensory seeking purposes More likely during transitions between tasks
Sensory experiences	Higher sensitivity and avoidance	Lower sensitivity and avoidance	Higher sensitivity and avoidance
Stress levels	Higher	Lower	Lower
Hyperactivity	Higher	Lower	Lower
Problem behaviours	Higher	Varying	Varying
Communication of symptoms	Lower	Varying	Higher
Cognitive symptoms	Lower	Varying	Higher

Tip

Future research should further explore the correlates and behavioural patterns of anxiety for persons with more severe ID and/or co-occurrence of autism spectrum disorder. The development of an anxiety measure that takes into account the specific behaviours and changes in presentation over time of these persons could result in more reliable identification and diagnosis.

Future studies are also hoped to address the relationship between stress, physical health, and anxiety across the lifespan and particularly during transition phases.

22.6 Aetiology and Risk Factors

In common with many mental disorders, the development of ADs is probably best perceived as an interaction between several different biopsychosocial factors. Currently, the definitive risk factors for ADs remain unclear. However, it is probable that a combination of multiple factors including genetics, environmental, psychological and developmental are likely to be involved [85–87]. For example, ADs tend to run in families, which suggest that a combination of genes and environmental stresses may produce these disorders. The candidate genes relating to ADs are largely the same across diagnoses and tend to be genes whose products regulate the hypothalamic-pituitary-adrenal (HPA) axis and monoaminergic signalling [88]. The impact of individual diagnosis-specific genetic risk factors may vary over time, depending on the developmental stage and previous experience of each subject.

When attempting to identify the genetic contribution towards susceptibility for psychopathology, similarities, however, do not preclude important clinical distinctions between diagnostic classes within ADs or between ADs and major depressive disorders (MDD). Some genetic factors are nonspecific but influence the risk for psychopathology in general; others are diagnosis specific.

There is a large body of evidence which concludes that panic disorder, GAD, phobias, and OCD all have significant familial aggregation and genes largely explain these phenomena. Gottschalk and Domschke [92] concluded from a comprehensive study that GAD is a heritable condition with a moderate genetic risk and heritability of approximately 30%. Within the anxiety spectrum, GAD was seen to be closely related to childhood separation anxiety, social phobia, and panic. A shared genetic origin with other internalizing disorders, especially MDD, became apparent during later developmental stages. Hettema and colleagues [48] reported that in ADs, certain genes predispose to two broad groups of disorders dichotomized as panic-generalized-agoraphobic anxiety versus the specific phobias.

A large population-based study undertaken by Kessler and colleagues [89] concluded that all types of mental disorder, including ADs, reduce in frequency with increasing educational level. Some genetic causes of ID/DID are associated with ADs. For example, Fragile X syndrome is associated with social anxiety disorder; Rubinstein–Taybi and Prader–Willi syndromes are associated with OCD [90]. Williams syndrome appears to be associated with ADs [91] and phobias [92]. Significantly high compulsive behaviour was noted in Cornelia de Lange syndrome [93].

Studies on types of fear reported in persons with ID/DID reported similarities between children and adults of equivalent mental age, highlighting the developmental perspective [28, 94, 95]. For example, individuals with moderate ID/DID experience fears of animals, thunder and ghosts (preoperational thinking), and physical injuries (concrete operational), mirroring normal Piagetian transition in non-learning-disabled children. It is however noteworthy that anxieties and phobias might also occur as transient phenomena in children of average intelligence which is integral to normal early development.

PWID/DID are also more likely to experience child abuse as well as other forms of traumatic or negative events later in life compared to the general population. Bradley and colleagues [96] noted that PWID/DID frequently encounter adversity and stressful life events as well as other trigger risk factors such as poor

coping skills and inadequate social supports which are known to increase vulnerability for anxiety and related disorders.

Multiple factors including genetics, environmental, psychological, and developmental are likely to be involved in the genesis of anxiety disorders. Some genetic syndromes, familial aggregation, low educational level, and stressful life events can increase the vulnerability for anxiety and related disorders.

22.7 Pathophysiology

The symptoms of anxiety and mood disorders are thought to result in part from disruption in the balance of activity in the emotional centres of the brain rather than in the higher cognitive centres [97]. Anxiety is a ubiquitous emotional state that represents an interrelationship between mammalian and primate stress response systems, and more recent expansion of the human neocortex. This expansion signalled the emergence of increasingly complex top-down regulation of limbic-subcortical responses, as well as an enhanced adaptability associated with developing executive functions. These changes which also include a hierarchically organized perceptual, processing, and response system is involved in modulating fear responses, enhanced adaptability, and neurocognitive skills necessary for dealing with uncertainty of increasingly complex social relationships and social/technological ecology [1, 2].

The inherent neuroplasticity of this infrastructure is maintained throughout our prolonged childhood. But development is not a linear process, but one governed by stage-specific time and stage changes in genetic activity, neuronal maturation, and myelination. There are critical periods during this developmental journey that suggest that this developmental flexibility also carries risks, prolonged periods of increased vulnerability, and potential insults that may derail these [2, 98, 99]. This is the essence of neurodevelopmental disorders such as ID/DID. Similar developmental insults (i.e. genetic vulnerability) also derail the developmental trajectory of the stress response system and more complex substrate for ADs. The co-occurrence of ADs with ASD and ID/DID further complicates the atypical neurodevelopmental trajectories of all three conditions and partially explains the nearly twofold increase in the prevalence rates of anxiety and ADs in this population [100–102].

The experience of anxiety undergoes many changes during the life cycle, including developmental processes outlined above. Derailment of this developmental process helps explain why some children may not successfully negotiate and master these challenges [103–105], neurotypical children developmental anxieties related to typical life issues that reflect changing expectations, cognitive development, and mastery of developmental crises. The likelihood of mastering these developmental challenges is profoundly influenced by temperament, patterns of attachment, and the capacity of caregivers to help the child negotiate these challenges.

Pathological anxiety represents an atypical pattern of experiencing and expressing anxiety during development. Left unchecked, pathological anxiety can serve as a source of functional impairment, as well as a potential risk factor for ADs. In genetically and biopsychosocially vulnerable individual, pathological anxiety may also represent either a prodromal phase or a subclinical expression of later developing ADs. These subclinical forms of ADs progress to full syndrome ADs in nearly 40–50% of affected patients [3, 97, 106, 107].

The developmental trajectory of each stage of ADs is complicated by a second pattern of divergence – developmental changes that are homotypic and heterotypic. An example of homotypic development is present when panic attacks (e.g. school phobia) appear during childhood that emerges as panic disorder in late adolescence. The fear-related reactions persist largely unchanged. Childhood school avoidance behaviour without overt fear-related arousal, but associated harm avoidance of behavioural inhibition, may evolve in generalized anxiety disorder, social anxiety (generalized type), or mood disorder. This

form of separation anxiety fits into the heterotypic category scenario [3, 5, 6, 108].

Genetic vulnerability (heritability), puberty, and psychosocial factors (including gene-environment interactions) help shape both homotypic and heterotypic courses [109]. Both pathways are transactional processes that arise from the complex entanglement of anxiety as a neurodevelopmental phenomenon, gene-environment interactions, and psychosocial factors. The presence of ASD, DID, or ASD+DID can strongly influence these developmental trajectories [110–112].

Developmental insults (e.g. heritability, puberty, psychosocial factors) can derail the developmental trajectory of the stress response system, creating a condition of vulnerability for the development of anxiety. The presence of ASD, ID/DID, or ASD+ID/DID can strongly influence these developmental trajectories, increasing the prevalence rate of anxiety disorders in this population.

22.8 Aetiopathogenic Models

Amongst neurotypical patients, a number of functional magnetic resonance imaging (fMRI) studies suggest a relationship between subtypes of anxiety disorders and amygdala hyper-reactivity, reduced integration between prefrontal cortex (PFC), anterior cingulate gyrus, bed nucleus of the stria terminalis, and association cortices with subcortical regions [113, 114].

These studies also suggest the involvement of aberrant processing of fearful faces (inferior temporal cortex), threat perception, vulnerability to fear conditioning and amygdala/hippocampal activity, and dysregulation of frontal-limbic networks that integrate and regulate input and output between these networks [4, 5, 115, 116]. As a general rule, the temporal-parietal and cortical, fronto-limbic, and fronto-subcortical linkages are critical for neuromodulation and top down affect regulation and impulse control as well as other executive functions. There are multiple studies addressing the role of the medial PFC and ventral-medial PFC in fear conditioning and extinction [2, 117].

These circuits are foundational to associative and instrumental conditioning. Applying these findings may enhance our understanding of the neurobiological foundations of fear conditioning, generalization, extinction, and the mechanisms for behavioural and pharmacological interventions [97, 118, 119]. But applying these principles to the pathophysiology of anxiety and ADs in patients with ID/DID will require a deeper understanding of their interrelationships between their underlying neurobiology, these learning models, and atypical cognitive development. One of our greatest challenges is to upgrade our current thinking about the bio-psycho-sociology of specific biomarker and the emergence of neurodevelopmental anxieties, pathological anxiety, and ultimately, ADs. Since ID/DID is a heterogeneous syndrome, this process will require fine tuning subsets of pathological anxiety and ADs. One of the key steps in this process will be the application of specific genetic and other biomarkers, pre-existing medical, neurological, and neurodevelopmental disorders, rather depending on descriptive criteria. Rather than listing biopsychosocial factors, we need to take the next step and explore how and why they influence the developmental trajectory of these endophenotypes. [2, 4, 99, 118, 119]

Recent developments in molecular genetics have suggested complex interaction between decreased activity in serotonin reuptake receptors (SCL6A4, 5-HTTPLR polymorphisms) and reduced activity amongst corticotrophin receptors, dysregulation of corticotrophin releasing factor, and responses to social and environmental stressors [2, 119]. It is unclear how these affect individuals with DI/DID, especially in the context of an increased lifetime risk for psychological, physical, and sexual abuse. For many, the boundary between PTSD and other trauma-related disorders, mood and anxiety disorders, and externalizing disorders is difficult to establish. Epigenetic changes in response to trauma have lifelong implications for anxiety, self-injurious behavior (SIB), aggression, explosive emotional reactivity, and other disruptive/destructive behaviours. These data

may enhance our understanding of trauma-related disorders and the long consequences of trauma on treatment refractory anxiety and mood disorders [120, 121].

Once these changes are underway, epigenetic forces alter the patterns of gene activation that can have lifelong implications. Yet, the effects of these same gene markers can be overridden by a nurturing mother (rat models this involves maternal licking of rat pups). However, those rat pups who do not receive this nurturing are vulnerable to the epigenetic changes that can increase the risk for subsequent traumatization. In humans, similar forms of deprivation can contribute to higher rates of chronic, treatment refractory mood and anxiety disorders [107, 122]. Once these changes are underway, epigenetic forces alter the patterns of gene activation that can have lifelong implications.

In summary, in common with other neurodevelopmental disorders, ID/DID is a complex condition associated with significant clinical diversity. Nevertheless, they do all seem to share a neurophysiological imbalance between dysfunctional excitatory (bottom-up limbic/brain stem) and a dysfunctional inhibitory network (top-down cortico-striatal). This imbalance may explain why panic attacks, phobic reactions, and other fear-related anxiety present as aggression, SIB, or destructive behaviours. For this patient's defensive aggression, SIB and other escape-motivated behaviours reflect deficits in self-regulating negative affective states and a greater vulnerability to automatic/pre-potent fear-conditioned responses. Those prone to approaching negative events and externalizing behaviours may express these in terms of disruptive or aggressive/destructive behaviours. Internalizers may be prone to avoidance and escape behaviours. Both are goal directed in terms of either obtaining relief or escaping affective states. In chronic traumatic or stressful settings, these conditioned avoidance behaviours persist (maintenance-phase) but can morph in vulnerable patients through a combination of sensitization (kindling-like phenomena) and progression to habit-/addiction-like behaviours. Consequently, some individuals are prone to engage in the behaviours independent of their previous trigger stimuli and replace their goal directed behaviours. Once these behaviours become habits, the connection between key stimuli and behavioural responses becomes less clear-cut [120–125].

Research is ongoing into how these variations affect individuals with severe to profound ID/DID, especially in the context of an increased lifetime risk for psychological, physical, and sexual abuse. For many, the boundary between PTSD and other trauma-related disorders, mood and ADs, and externalizing disorders is difficult to establish. Epigenetic changes in response to trauma have lifelong implications for anxiety, SIB, aggression, explosive emotional reactivity, and other disruptive/destructive behaviours [2, 123, 124, 126].

This section on pathophysiology is a synthesis of both conceptual models that moved away from a diagnostic approach that relied primarily on our current descriptive/categorical phenomenological diagnostic criteria. Combining these two models may provide a more comprehensive platform for understanding the relationships between anxiety disorders, ID/DID and ASD. In addition, genetic studies suggest that clinically relevant polymorphisms are associated with secondary and tertiary messenger systems, intracellular ion release and the regulation of synapse formation and stabilization, neuroplasticity and learning, and memory and extinction. Brain maturation involves the maturation, integration, and hierarchical organization of each of these processes [2, 99, 124, 127, 128]. The use of more sophisticated, higher resolution functional neuroimaging and neurophysiological techniques will help map these changes. Combining and integrating these technologies will help us to better understand the pathophysiology of AD, but coping with the heterogeneity associated with ASD/AD remains a work in progress.

The pathophysiology of anxiety disorders in persons with ID/DID remains unclear. Some etiopathogenetic models have been considered, including the interaction between decreased activity in serotonin reuptake receptors and reduced activity amongst corticotrophin receptors, the dysregulation of corticotrophin releasing factor, and the

responses to social and environmental stressors. Epigenetic changes in response to trauma seem to have lifelong implications for anxiety, self-injurious behaviours, aggression, explosive emotional reactivity, and other disruptive/destructive behaviours.

22.9 Neurobiology

A myriad of neuroendocrine, neurotransmitter, and neuroanatomical disruptions in the limbic, brain stem, and higher cortical brain areas appear to be implicated in ADs. The differences remain elusive in view of complexities within this system and the high degree interconnectivity between neurotransmitter- and neuropeptide-containing circuits in limbic, brain stem, and higher cortical brain areas. A primary alteration in brain structure or function or in neurotransmitter signalling may result from environmental experiences and underlying genetic predisposition which can increase the risk for psychopathology [97].

The neurotransmitters that are mainly implicated in the symptoms of anxiety disorders within the central nervous system are noradrenaline, serotonin, dopamine, and gamma-aminobutyric acid (GABA). Others that may possibly play a role include corticotropin-releasing factor and peptides.

Many of the symptoms of the symptoms of ADs are mediated peripherally by the autonomic nervous system, in particular, the sympathetic nervous system [129].

A study by Katerndahl and colleagues [130] using positron emission tomography (PET) scanning demonstrated an increased flow in the right parahippocampal region and reduced serotonin type 1A receptor binding in the anterior and posterior cingulate and raphe of patients with panic disorder. Vythilingam's [131] study using magnetic resonance imaging (MRI) demonstrated smaller temporal lobe volume in patients when compared with healthy subjects with normal hippocampal volumes in both patients and controls. The cerebrospinal fluid (CSF) in studies in humans with panic disorder show elevated levels of hypocretin, thought to play an important role in the pathogenesis of panic in rat models [130].

Obsessive-Compulsive Disorder

The current research and treatment trials suggest that abnormalities in serotonin (5-HT and in some cases, dopamine) neurotransmission in the brain are involved in OCD. The efficacy of serotonin reuptake inhibitors (SSRIs) in the treatment of this condition strongly endorses this contention. Emerging evidence from a genetic perspective support the view that OCD co-varies in conditions such as Tourette's syndrome and multiple chronic tics in an autosomal dominant pattern. Bloch [132] reported that symptoms of OCD in such individuals respond preferentially to a combination therapy of SSRIs and antipsychotics.

Attention has also been focused on glutamatergic abnormalities and possible glutamatergic treatments for OCD. Although modulated by serotonin and other neurotransmitters, the synapses in the cortico-striato-thalamo-cortical circuits thought to be centrally involved in the pathology of OCD principally employ the neurotransmitters glutamate and GABA.

The imaging studies in OCD such as magnetic resonance imaging (MRI) and PET scanning have demonstrated increases in blood flow and metabolic activity in the orbitofrontal cortex, limbic structures, caudate, and thalamus, with a trend towards right-sided predominance in OCD. Some studies report that these areas of overactivity have been shown to normalize following successful treatment with either SSRIs or cognitive-behavioural therapy (CBT) [133]. This supports the hypothesis that symptoms of OCD are a consequence of impaired intracortical inhibition of specific orbitofrontal-subcortical circuitry that mediates strong emotions and the autonomic responses to those emotions. It is noteworthy that cingulotomy, sometimes used for severe and treatment-resistant OCD, interrupts this circuit.

Some neuroendocrine, neuroanatomical, and neurotransmitter alterations appear to be implicated in anxiety disorders. It is well established that abnormalities in serotonin neurotransmission are involved in obses-

sive-compulsive disorder, and the efficacy of serotonin reuptake inhibitors in the treatment of this condition strongly endorses this contention. Attention has also been focused on glutamatergic abnormalities and possible glutamatergic treatments for obsessive-compulsive disorder.

22.10 Management and Treatment

Empirically based studies on treatment of ADs are sparse in PWDID and limited to a handful. However, the full range of strategies utilized in the general population including effective psychological interventions are available [134]. Cognitive behaviour therapy (CBT), dialectical behaviour therapy (DBT), reassurance, and counselling have been successfully used as have anxiety management techniques such as relaxation training, mindfulness, anger management, and self-help techniques [135]. Behaviour therapies such as desensitization and exposure therapy are effective management strategies in OCD and social phobia.

The need for treatment for ADs is influenced by the intensity and duration of illness and the impact of symptoms on everyday life [136]. The NICE Guidelines [137, 138] recommended evidence-based psychological interventions as a first-line intervention with pharmacological therapy as a second choice.

Some studies in the general population support the contention that optimum results are achieved by combination of psychological and pharmacological interventions [139], such as the combination of CBT with SSRIs [140]. Behavioural therapy and cognitive therapy alone or in combination have demonstrated clear evidence of efficacy in the treatment of ADs [140, 141]. The involvement of patients in their treatment by using comprehensible and clear communication is reported to improve outcomes [142].

Psychotherapy, such as exposure therapy, cognitive therapy, and cognitive-behavioural therapy (CBT), has consistent evidence of efficacy in the treatment of ADs [143, 144]. The efficacy of psychological and pharmacological approaches is broadly similar in the acute treatment of ADs.

Evidence based on case studies in people with mild or borderline ID/DID and ADs have demonstrated the effectiveness of CBT [145–147]. However, the core principles of CBT may require modification to meet individual abilities. Overall, currently available empirical evidence with regards the efficacy of CBT for treatment of ADs in persons with ID/DID is weak. A range of psychological interventions [147, 148] including reassurance, counselling, exposure-based approaches, anxiety management such as relaxation training, anger management, and self-help are being found to be effective and useful in this population [134, 147].

The aim of behavioural interventions is to modify any behaviour that is unhelpful or harmful. Commonly used behavioural interventions include relaxation which appears to be effective in reducing anxiety as well as improving cognitive performance amongst those with mild, moderate, and severe DID. Relaxation behavioural training has been found to be effective in the treatment of generalized anxiety in persons with ID/DID. Lindsay et al. studied anxiety treatments for adults who have moderate and severe ID/DID by undertaking a study based on the simplification of a technique called progressive relaxation, which demonstrated that behavioural relaxation training is an effective treatment modality [145, 149, 150].

Studies on psychological interventions of anxiety disorders in persons with ID/DID are scarce and limited. Evidence based on case studies indicates effectiveness of cognitive-behavioural therapy, although it may require variation based on individual skills. Other psychological interventions including reassurance, counselling, relaxation training, anger management, and self-help have been found to be effective in this population.

22.11 Pharmacological Treatment

In the general population, four broad categories of psychotropic medications are used in adults with anxiety syndrome: (1)

antidepressants, (2) benzodiazepines, (3) other medications (e.g. Buspirone), and (4) antipsychotics for severe debilitating symptoms. In GAD, OCD, panic disorder, PTSD, and social phobia, SSRIs (selective serotonin reuptake inhibitors) may be used as first-line treatment. The SNRIs (serotonin noradrenaline reuptake inhibitors) duloxetine and venlafaxine have proven efficacy in short-term and long-term treatment of GAD [151]. Venlafaxine is also efficacious in the acute treatment and relapse prevention of panic disorder [152]. With the exception of social phobia, tricyclics may be used as second-line intervention for all of these conditions. Other pharmacological treatments such as buspirone (GAD, OCD) and antipsychotics (quetiapine or risperidone) as an augmentation for OCD have a weaker evidence base or are less well tolerated.

Despite the widespread belief that antidepressants can lower the seizure threshold, systematic review of data from placebo-controlled trials indicates that the frequency of seizures is significantly lower with psychotropic drugs than with placebo [153].

Benzodiazepines should be considered only for short-term (up to 4 weeks) treatment of acute, disabling anxiety which is causing significant distress, while longer-term strategies are instituted in view of potentially adverse effects. Nevertheless, a minority with intractable anxiety may benefit from long-term treatment, with benzodiazepines which should not be denied [154, 155].

Follow up of anxiety symptom severity after initial treatment is reported to be lower with CBT than with other forms of psychological treatment: but the comparative efficacy of pharmacological and psychological approaches over the long term is not established [156]. It is probable that the combination of pharmacological and psychological treatment is superior to psychological approaches or medication, when either is given alone [136].

In persons with ID/DID and/or ASD, excess prescriptions of anxiolytics, antidepressants, and GABA agonists in comparison with the general population are reported, especially older individuals [157]. Empirical evidence is not available on the pharmacological efficacy of specific anxiolytic medications for discrete types of ADs. As such, current treatment is guided by consensus expert opinion extrapolated from the general population. Anti-anxiety medications should only be undertaken following a comprehensive and meticulous evaluation of the person to exclude medical, psychiatric, behavioural, and environmental precipitants of the anxiety symptom/s. The therapeutic goal for treating ADs is maximum reduction of symptoms or elimination of symptoms as described by the patient and or caregiver or as measured by behavioural monitoring. Whereas in persons with milder degrees of ID/DID the individuals are able to self-report, the clinician is dependent on behavioural symptoms to determine the efficacy of treatment in individuals with more severe degrees of ID/DID. The behavioural monitoring requires consistent measurements over a minimum period of several days of observation.

The pharmacological management of anxiety and related disorders in PWID/DID must be undertaken within the context of a comprehensive physical, psychosocial, and environmental assessment. The treatment aim is targeted at achieving maximum gains with minimum harm the lowest effective dose. PWID/DID need careful and frequent monitoring since they may not be able to report adverse effects. The possibility of drug interactions, lower doses, and the risk of worsening any pre-existing cognitive impairment, particularly from medications with sedative effects, should also be borne in mind [158]. In the patients with ID/DID, consensus expert opinion recommends second-generation, selective serotonin reuptake inhibitor (SSRI) medications as treatment of first choice for ADs. Expert clinical consensus favours the short-term use of anxiolytics in some instances to enable those individuals who are deemed too anxious to engage in psychological interventions to facilitate the process.

▪ OCD

Remission of OCD spontaneously without treatment is unusual. The treatment of choice is CBT modified as relevant with SSRIs as second choice. Most improve significantly

with treatment, but around 15% have symptoms which worsen with functional impairment over time. A minority of approximately 5% reportedly have complete remission of symptoms between episodes of exacerbation. OCD symptoms can worsen with stress; however, stress does not appear to be an aetiological factor.

PTSD

There is robust evidence supporting the efficacy of SSRIs and SNRIs in the acute treatment of PTSD continuing for at least 12 months after response [136]. All PTSD sufferers should be offered a course of trauma-focused psychological treatment (CBT) or eye movement desensitization and reprocessing (EMDR) which is safe and effective [144] preferably on an individual outpatient basis. When evidence-based psychological or pharmacological treatments fail, combination therapy may be considered. There is limited evidence in support of combination therapy with paroxetine and exposure therapy.

Ultimately, the treatment choice should be a consequence of the assessment process and shared decision-making, with a strong emphasis on safety, tolerability, and the patient's preferences within the context of best available evidence.

The pharmacological management of anxiety disorders in persons with ID/DID must be undertaken within the context of a comprehensive physical, psychosocial, and environmental assessment. Selective serotonin reuptake inhibitors may be used as a first-line treatment, while anxiolytics should be used in the short term and only in special cases. Persons with ID/DID need careful and frequent monitoring since they may not be able to report adverse effects.

22.12 Prognosis

In general, maintenance of pharmacological treatment is recommended in anxiety and related disorders where effective, since discontinuation frequently precipitates relapse. Successful completion of CBT may bring about enduring relief following treatment. Evidence also suggests that "extending/top up" CBT may further improve the outcome. Some patients may have treatment-resistant CBT requiring multiple trials of medication. An even smaller proportion with intractable and disabling symptoms may benefit from neurosurgical interventions. OCD sufferers improve significantly with appropriate treatment including CBT and, often, medication.

Tip

Anxiety disorders in people with ID/IDD deserve increasing research interest and continued acquisition of up-to-date knowledge by clinicians, especially in reference to persons with more severe degrees of cognitive and/or communication impairment.

Key Points

- Anxiety disorders in persons with ID/DID are common, but often not diagnosed, resulting in poor access to appropriate treatment which lowers quality of life.
- The identification of anxiety disorders in people with more severe degrees of ID/DID is particularly difficult and frequently missed by carers and clinicians. Adaptations of the current classification systems (DM-ID, DC-LD) and psychopathology assessment tools (PIMRA, DASH, ADAMS, PAS-ADD, CBCL, GAS-ID) can facilitate the diagnostic process in this population.
- The clinical features of anxiety include cognitive, physiological, psychological, and behavioural elements. For people with more severe degrees of communication impairment, manifestation of these features can be observed rather than subjectively reported.
- The management and treatment of anxiety disorders in persons with ID/DID must be undertaken within a multidimensional assessment. Psychological interventions including cognitive-

behavioural therapy and behavioural therapy are been found to be effective in this population. Pharmacological treatment has also proved effective, and SSRIs are recommended as a first-line drug treatment.

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Somatic Symptom and Related Disorders

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Contents

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Learning Objectives

The key objectives of this chapter are to describe:

- The presenting symptoms and phenomenology of specific somatic symptom and related disorders in persons with intellectual disability and autism spectrum disorder with high support needs.
- The differential diagnosis and factors which contribute to the development of these disorders.
- The multimodal approach to their management.

23.1 Introduction

Somatic symptom and related disorders (SSRD) are a group of conditions characterized by persistent, distressing and excessive thoughts, emotions and behaviours regarding somatic symptoms. These somatic symptoms, which are substantially more severe than expected from underlying pathology or have no medical explanation, and the accompanying excessive thoughts, emotions and behaviours impair and disrupt personal functioning and adjustment [1].

In persons with intellectual disability (ID) and low-functioning autism spectrum disorder (ASD), SSRD are often overlooked, especially in primary care and regarded as a medical disorder or as emotional and behavioural difficulties directly due to the ID. Therefore, it is likely that persons with ID and/or ASD who suffer from an SSRD remain undiagnosed and do not receive specific mental health treatment and support.

In persons with neurodevelopmental disorders, mind-body interactions and their influence on mental and physical health seem to be stronger than in the general population, which has been associated with cognitive issues and difficulties in identifying and managing personal emotions and psychological suffering. Features of alexithymia, defined as the inability to differentiate, describe and label one's own emotions [2] and repeatedly associated in the general population with a high risk of SSRD [3, 4], have been found in persons with neurodevelopmental disorders and related to difficulties in identifying and distinguishing between feelings and bodily sensations [5–7]. According to a developmental perspective, these difficulties and the consequent expression of somatic symptoms may be reinforced or weakened by particular life experiences, relationships and psychological assets across childhood, adolescence and early adulthood.

23.2 Somatic Symptom and Related Disorders

SSRD, classified in the DSM-5 [1], are a group of conditions in which the person experiences excessive and persistent somatic symptoms causing distress which adversely affects and disrupts daily life. This group of disorders comprises somatic symptom disorder, illness anxiety disorder, conversion disorder (functional neurological symptom disorder), psychological factors affecting other medical conditions, factitious disorder, other specified somatic symptom and related disorder and unspecified somatic symptom and related disorder.

23.2.1 Prevalence

Somatic symptoms seem to be more frequent in persons with ID than in the general population across the lifespan from early childhood to the elderly. Troublesome somatic symptoms are reported in 12–14% of children and adolescents, decreasing to around 10% in adults [8–12]. These symptoms are more prevalent in females than in males and are associated with increased levels of overall emotional and behavioural disturbance [9–12].

It is well established that children with ASD have a relatively increased risk of gastrointestinal symptoms compared to typically developing (TD) children [13, 14]. A meta-analysis of literature demonstrated an increased odds (4.42 OR) of gastrointestinal symptoms in children with ASD compared to TD children [15]. The likelihood of

gastrointestinal symptoms is also greater in children with ASD compared to those with developmental delay (1.74 OR) [13]. These studies on reported gastrointestinal symptoms have not specifically diagnosed what proportion have SSRD compared to other likely organic causes of symptoms such as ulcerative colitis or gluten enteropathy. There is limited information available on the specific prevalence of SSRD in those with ID who also have ASD. One study of a range of somatic symptoms in children with ASD and ID (N = 118) found an increased likelihood (1.5 OR) of somatic symptoms compared to other children with ID (N = 590) [12]. Clearly further research is required on the phenomenology of SSRD in persons with ID who also have ASD across the lifespan.

23.2.2 Aetiopathogenesis

There is a lack of research regarding the aetiology of somatic symptom disorders in people with ID. To date, a multifactorial origin, biological, psychological and environmental, is the most valued hypothesis, as for most psychiatric disorders in persons with and without neurodevelopmental disorders [16].

23.2.2.1 Biological Factors

The higher rates of chronic physical illness such as gastrointestinal disease and epilepsy in persons with ID [17, 18] and their excessive use of tobacco [19] and caffeine [20] might either create anxiety in relation to somatic symptoms or provide a rewarding experience of care and knowledge of illness, thus predisposing the individual to a somatic symptom disorder. Somatic symptoms are also associated with the behavioural phenotype of some genetic disorders. For example, conversion disorders are associated with Fragile X syndrome and its pre-mutation form [21]. Young people with Williams syndrome are three times more likely than others with ID to have somatic symptoms probably associated with high levels of anxiety [12, 22] and somatic symptoms are commonly associated with Prader–Willi syndrome [23]. Young people with Down syndrome perhaps as a reflection of their generally low level of emotional and behavioural disturbance are four times less likely than others with ID to have somatic symptoms [12, 24].

23.2.2.2 Psychological and Environmental Factors

The increased likelihood that persons with ID will experience adverse life events, stigma, abuse and social adversity might increase the risk of somatic complaints [25], but these symptoms are likely to also be associated with anxiety or depressive illness [8, 26].

Overall emotional and behavioural disturbances have been associated with increased rate of somatic symptoms [9, 11, 12, 19].

According to the authors' clinical experience, the co-occurrence of ID and ASD and the experience of anxiety may increase the probability to develop an SSRD. The inability to express emotions verbally and the impoverished fantasy and imaginal life, which characterize the ASD, are associated with difficulty in distinguishing emotional states from bodily sensations. Furthermore, persons with ASD may experience pain in unusual ways and have considerable difficulties with pain awareness and communication, ending up in a vicious circle that causes great distress [27].

Somatic symptoms seems to be more frequent in persons with ID, and particularly ID comorbid with ASD, than in the general population across the lifespan from early childhood to the elderly. Aetiology is uncertain with multifactorial origin, biological, psychological and environmental, being the most valued hypothesis.

23.3 Somatic Symptom Disorder

23.3.1 Criteria and Clinical Features

Somatic symptom disorder (SSD) presents with persistent, distressing and excessive thoughts, emotions and behaviours regarding somatic symptoms. These somatic symptoms

and the accompanying excessive thoughts, emotions and behaviours impair and disrupt daily life and adjustment. The somatic symptoms may be medically unexplained but might also be associated with a medical condition, but in both instances the critical aspect of the diagnosis is the presence of disproportionate and excessive thoughts, emotions and behaviours regarding these somatic symptoms. Therefore, the task of the clinician, regardless of whether or not the somatic symptoms have a medical explanation, is to undertake a comprehensive biopsychosocial assessment in order to determine if the psychological symptoms are excessive and persistent. Diagnosis therefore requires medical experience or at least clinical consultation. In the DSM-5 [1] field trials, the diagnosis of SSD was found to be highly reliable, identifying more cases than the previous DSM-IV criteria which required that the symptoms had to be medically unexplained [28]. These studies also indicated that the diagnosis of SSD has better predictive validity of psychological functioning at 1 year follow-up than the previous DSM-IV criteria [29, 30]. The DSM-5 [1] criteria also means that chronic pain associated with medical illness and injury is a qualifier of SSD provided that it is associated with excessive and disproportionate thoughts, feelings and behaviours. Therefore, people with chronic pain who, because of a complex interaction of biopsychosocial factors, also experience disabling psychological symptoms can now have this identified and treated as an SSD without the previous stigma which inferred that there was no physical basis to their problems or that their difficulties were a fabrication of their mind.

Diagnosis of SSD requires that the somatic symptoms are distressing and cause significant impairment in daily functioning and adjustment (Criterion A). The number or nature of the somatic symptoms may vary, but symptoms are persistent (Criterion C). Symptoms may be specific such as a chest pain or nonspecific such as fatigue. Individuals with an SSD also experience excessive and persistent thoughts, emotions and behaviours associated with the somatic symptoms (Criterion B). For example, individuals might spend considerable time worrying about the seriousness of their somatic symptoms and are anxiously preoccupied about their health. They often give exaggerated and inconsistent accounts of their illness concerns and frequently seek advice from a range of health and alternative medical treatment practitioners leading to multiple medical and alternative treatments. Normal physical sensations and bodily functions are erroneously thought to be due to physical illness, and physical activity might become more limited due to a fear of injury and time is spent examining the body for abnormalities. The presentation in children is usually less complex with concern focused on one symptom.

Medical illness is the main differential diagnosis, but the presence of excessive and distressing thoughts, feelings and behaviour is necessary to make the diagnosis of SSD in obvious medical conditions such as ulcerative colitis and diabetes. Somatic symptoms can also occur in anxiety disorder, depressive disorder, conversion disorder (functional neurological symptom disorder), delusional disorder and body dysmorphic disorder. A differentiation is made between SSD and obsessive–compulsive disorder in which the compulsive behaviours are an attempt to relieve the anxiety of intrusive, obsessional thoughts. SSD can also be comorbid with other psychopathological conditions, particularly anxiety disorders and depressive disorders, for example, in the elderly. Cultural factors might influence the type of symptoms, understanding of causation and approaches to treatment.

The critical aspect of the diagnosis of somatic symptom disorder is the presence of disproportionate and excessive thoughts, emotions and behaviours regarding persistent somatic symptoms.

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23.3.2 Diagnosis of SSD in Persons with Intellectual Disability

A diagnosis requires that the person is able to talk about their symptoms and reflect on their emotions and thoughts. Therefore, the diagnosis of SSD in a person with moderate or more severe levels of ID who do not have language ability can only be at best inferred through their behaviour. For example, changes in previous behaviour such as posture, the use of limbs and apparent loss of sensation or focus on a specific part of the body might indicate pain or evidence of dysfunction, and other behaviours indicate excessive distress and preoccupation, such as psychomotor agitation, distractibility, irritability or aggressivity. A full adaptation to ID of the DSM-5 diagnostic criteria has recently been produced by Bertelli, Tonge and Brereton [16].

Clearly a careful history and routine observation by caregivers is a necessary component of assessment and management. Persons with ID who possess some verbal skills are more likely to give an inconsistent and medically naive account of their symptoms requiring a supporting history from a caregiver to confirm the diagnosis. Because people with ID are more likely to have more limited access to medical services [31] and general medical practitioners often lack experience in the medical care of people with ID [32], somatic symptoms might be regarded as a medical disorder and any emotional and behavioural difficulties are regarded simply as a consequence of the ID. Therefore, it is likely that persons with ID who suffer from an SSD remain undiagnosed and do not receive specific mental health treatment and support.

The diagnosis of SSD requires that the symptoms impair the person's functioning in daily life. Therefore, for a person with ID, it is necessary to gather information from caregivers, employers and day programme staff to determine that the symptoms have interrupted usual day time activities such as their participation in domestic and community activities.

In people with ID who have limited or no language ability, it is likely to be difficult if not impossible to differentiate the presenting behavioural symptoms from another mental disorder. For example, behavioural symptoms such as psychomotor retardation, loss of interest in usual activities and changes in appetite are useful in the diagnosis of a mood disorder, but these symptoms might also be present in SSD. Somatic symptoms of anxiety including sweating, shaking and agitation can be reliably diagnosed in individuals with ID [33, 34]. Altered sensory responsiveness such as hypo- or hypersensitivity to pain is also experienced in persons with ID who also have ASD [35, 36]. The behavioural phenotypes of Prader–Willi syndrome [23] and velocardiofacial syndrome [37] are likely to include somatization symptoms indicating a genetic predisposition. Therefore, in the absence of the ability of a person with ID to report on their symptoms and emotions and the necessary reliance on the observation of behaviours to infer somatic symptoms and differentiate SSD from other mental disorders or neurodevelopment conditions, making the diagnosis of SSD is difficult if not impossible.

In the diagnosis of SSD, it is necessary to determine that the somatic symptom such as pain is "not intentionally produced or feigned" for some gain such as assuming a sick role, obtaining medical attention or receiving a reward such as avoiding a duty. Therefore, care must be taken in the diagnosis of SSD in a person with ID to be sure that a somatic symptom such as vomiting did not initially occur in a context such as a community residential unit where this symptom has led to repeated secondary gain such as the avoidance of a specific disliked community activity. Traumatic and stressful life events are risk factors for SSD but are also associated with a range of other mental health problems such as anxiety disorders, post-traumatic stress disorder and conversion disorder. Compared to the general community, a person with ID is at increased risk of experiencing stressful and traumatic life events, denigration and physical, emotional or sexual abuse [38] and homelessness [39]. Therefore, it is essential that the clinician gathers historical and current information from the person with ID if possible

and from caregivers regarding their experience of trauma and psychological stress. Environmental changes and unexpected events are also likely to be stressful to a person with ID [38] acting as a precipitating risk factor for SSD. For example, sudden changes in the routine of a day programme or the loss of a regular caregiver can cause stress, highlighting the need for an assessment of the person's historical and current response to change as part of the diagnostic assessment for SSD [40]. In persons with ID, the likelihood of comorbidity of SSD with anxiety and depression reflects findings from the general community in which somatic symptoms and comorbid depression and anxiety are predicted by recent stress, increased use of health services and chronic medical conditions [41].

The diagnosis and treatment of an anxiety or a mood disorder takes precedence over the diagnosis of SSD. In situations of diagnostic uncertainty, particularly in persons with more severe levels of ID and poor communication skills, it is appropriate to make a diagnosis of a mood or anxiety disorder instead of an SSD if the observed behaviours and history of behavioural change suggests a mood or an anxiety disorder. Systematic follow-up of behavioural response to treatment can help to confirm diagnosis or indicate a comorbid SSD.

There are older case study reports in the literature which probably meet criteria for SSD such as a man with moderate ID and somatic symptoms of globus pharyngeus [42]. Carer or parent report checklists of emotional and behavioural symptoms in persons with ID are a reliable and valid means of detecting somatic symptoms. For example, the Psychopathology Inventory for Mentally Retarded Adults (PIMRA) [43] has a Somatoform Disorder Scale [44] and the Developmental Behaviour Checklist (DBC) for children and adolescents [45] or adults [46] records somatic symptoms. However, the likelihood that somatic symptoms recorded by these checklists will be associated with higher rates of other symptoms indicates the likelihood of another diagnosis such as depression or anxiety.

In persons with ID, somatic symptom disorder is often overlooked. Somatic symptom might be regarded as a medical disorder and emotional and behavioural difficulties simply as a consequence of the ID. In those with moderate-to-severe ID, changes in posture, the use of limbs and apparent loss of sensation or focus on a specific part of the body might indicate pain or evidence of dysfunction, and other behavioural alterations might express excessive distress and preoccupation, such as psychomotor agitation, distractibility, irritability or aggressivity.

23.4 Illness Anxiety Disorder

23.4.1 Criteria and Clinical Features

Illness anxiety disorder (IAD) is manifest by functional impairment due to anxiety about health that is not better explained by an anxiety disorder such as generalized anxiety.

The person has fearful preoccupying thoughts that they have a serious illness and misinterpret minor bodily symptoms (Criterion A). Somatic symptoms, if present at all, are mild (Criterion B) and significant anxiety about health is experienced (Criterion C). Concern about illness is a consuming preoccupation to the extent that they repeatedly examine themselves, seek information about their imagined condition and persistently seek advice from family, friends and multiple medical services (Criterion D) which do not reassure them [1].

23.4.2 Diagnosis of Illness Anxiety Disorder in Persons with Intellectual Disability

Persons with ID who are able to communicate are likely to have a naïve understanding of illness and therefore have difficulty realizing that the fear of illness is unjustified. The diagnosis of IAD is usually not possible in those with more severe levels of ID who cannot use language. An episode of distressing illness in a person with ID might sensitize them to an

excessive emotional response to a subsequent reminder of this illness, and if this response persists, the diagnosis of an IAD might be considered. Therefore, a comprehensive history of previous illness and their response assists diagnosis. Two case studies of hypochondriasis (which has been replaced by IAD) in adult men with ID report hypochondriacal delusions [47, 48] which better meet diagnostic criteria for a delusional disorder rather than IAD. IAD is thought to be rare in children and has its onset in early to middle adult life [1].

The main differential diagnosis is a medical condition, although diagnosis of IAD is not excluded if preoccupying anxiety and fear regarding the medical illness are excessive and unreasonable. Individuals with generalized anxiety can have concerns of illness, but they are not persistent and preoccupying. Sufferers of an obsessive–compulsive disorder can have obsessional thoughts about developing an illness but are not focused on anxiety about a current illness. Depressive disorder, SSD and adjustment disorder are other differential diagnoses. Illness delusions can be present in a psychotic disorder but are not credible which differentiates them from IAD. Associated comorbid conditions are not well described but are most probably SSD and personality disorders [1].

One or more episodes of distressing illness might sensitize person with ID to an excessive emotional response to a subsequent reminder of this illness, and if this response persists, the diagnosis of an IAD might be considered. Therefore, a comprehensive history of previous illnesses and their response assists diagnosis.

23.5 Conversion Disorder (Functional Neurological Symptom Disorder)

23.5.1 Criteria and Clinical Features

Conversion disorder (functional neurological symptom disorder) (CD/FNSD) present with at least one pseudo-neurological symptom of abnormal sensory or voluntary motor function (Criterion A) that is inconsistent with a known neurological or medical disorder (Criterion B) [1]. The symptom is not better explained by another medical or mental illness (Criterion C) and causes significant distress or impairment in daily life (Criterion D). CD/FNSD can present with a range of symptoms including abnormal gait, motor weakness, paralysis, altered or absent skin sensitivity, hearing and sight. These abnormalities must be incompatible with the symptoms, signs and investigative findings of known neurological disorders. Symptoms such as depersonalization and amnesia and stressful experiences are common antecedents. It is not necessary to unequivocally determine that the symptom is not feigned for secondary gain, unless it is clearly evident, in which case the alternative diagnosis of factitious disorder would be made.

23.5.2 Diagnosis of Conversion Disorder (Functional Neurological Symptom Disorder) in Persons with Intellectual Disability

A comprehensive developmental and clinical history from parents or carers, neurological examination and indicated investigations are necessary to confirm the diagnosis, particularly in individuals with more severe ID and limited or no language, when the application of modified diagnostic criteria for use in ID such as the Diagnostic Manual-Intellectual Disability (DM-ID-2) [49] are helpful.

Neurological conditions, SSD, factitious disorder, dissociative disorder, body dysmorphic disorder, depressive and panic disorder are differential diagnoses. However, comorbidity occurs with neurological disorders, SSD, anxiety and depressive disorders and personality disorders but less commonly with substance use and psychosis. For people with ID, differentiation of epilepsy from non-epileptic seizures (NES) is a challenge. The risk of having refractory and recurrent epileptic seizures together with NES is increased in

individuals with ID [25, 50, 51]. Psychosocial stress and cultural influences also increase the likelihood of NES [25, 51, 52]. Abnormal activation of frontal inhibitory brain function might also have a genetic basis manifest as an increased risk of conversion disorder and NES in Fragile X syndrome [21]. A study in Bangladesh of 44 children with NES found that 77% had a conversion disorder and 23% of these also had ID [51]. There are some reported case studies of children with ID who might have a CD/FNSD presenting with pseudo-seizures in the context of a normal EEG, but the possibility of secondary gain from attention challenges the diagnosis [53]. The DSM-5 suggests that the prognosis for children is better than for adolescents and adults [1].

A comprehensive developmental and clinical history from parents or carers, neurological examination and indicated investigations are necessary to confirm the diagnosis, particularly in persons with more severe ID and limited or no language.

23.6 Psychological Factors Affecting Other Medical Conditions

23.6.1 Criteria and Clinical Features

Psychological factors affecting other medical conditions (PFAMC) is a disorder where a medical (not mental) illness is present (Criterion A) which is adversely affected by emotional and behavioural factors (Criterion B) that influence the course and/or treatment, that create additional health risks and/or that adversely influence the illness process necessitating treatment and may even be life threatening. These psychological or behavioural factors are not due to another mental illness such as depression, anxiety or Posttraumatic stress disorder (PTSD) [1].

The impact of the psychological factors on the medical condition can vary from a mild increase in medical risk to severe and life threatening such as ignoring the symptoms of an acute bowel obstruction. It can occur at any age and be acute or chronic, related to stress, anxiety, denial, personality, relationship difficulties and cultural context. If the emotional or behavioural problems develop as a consequence of the medical illness, then they are diagnosed as an adjustment disorder. An SSD which has maladaptive thoughts and feelings or an IAD with high illness anxiety and no associated medical illness are differential diagnoses.

23.6.2 Diagnosis of Psychological Factors Affecting Other Medical Conditions in Persons with Intellectual Disability

There is little literature regarding the phenomenology of PFAMC in people with ID. It could be argued that ID itself, particularly when more severe and where there is limited or no language, is a factor that can adversely affect a medical condition. Persons with ID have an increased risk of suffering chronic medical conditions such as osteoporosis and musculoskeletal disorders (MSDs), chronic dental disease and gastrointestinal problems including constipation and reflux and may not be able to adequately report their symptoms [17, 54, 55]. As a consequence, the medical risk is increased. Persons with ASD [1] often have sensory sensitivities which may include reduced sensitivity to pain leading to injuries such as bone fractures or medical conditions such as dental caries or peptic ulcers being unrecognized and deteriorating. Differential diagnoses include a mental illness due to a medical condition, an adjustment disorder where psychological symptoms develop in response to a medical condition, SSD where excessive thoughts and feelings are in response to somatic symptoms and IAD with high anxiety with minimal somatic symptoms. The assessment of PFAMC in children requires a history and observation from parents and teachers.

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23.7 Factitious Disorders

23.7.1 Criteria and Clinical Features

Factitious disorders (FD) are induced falsified symptoms or signs of injury or disease or psychological disorder associated with intent to deceive (Criterion A). The individual presents themselves or is presented by another, as ill, impaired or injured (Criterion B) not necessarily to achieve an obvious reward (Criterion C). The FD is not better explained by a psychotic disorder or mental illness (Criterion D) [1]. There are two types: one imposed on the self and the second imposed on another. A wide range of presentations include producing physical injury, cutting to obtain blood to put in urine or faeces to mimic a disease, talking nonsense, claiming hallucinations, appearing to be forgetful or in tears to indicate a mental illness or claiming symptoms of a medical illness such as stomach pain (which is referred to as Munchausen syndrome after an eighteenth-century German Baron who fabricated stories of his exploits). A factitious disorder imposed on another (Munchausen syndrome by Proxy) occurs when the diagnosed individual intentionally causes illness, for example, by administering insulin, or reports fictitious symptoms in someone they care for such as a child, an elderly person or someone with an ID. The presentation to medical services is often dramatic with an overelaboration and inconsistent history given (pseudologia fantastica). If challenged, they leave but usually travel to present at many other medical facilities.

23.7.2 Diagnosis of Factitious Disorders in Persons with Intellectual Disability

Persons with ID are likely to be medically naïve and produce more obviously fabricated signs or inconsistent symptoms perhaps copied from others or the media. It is essential to the diagnosis to determine that the intentional production of signs and symptoms is in the absence of any obvious reward such as care and attention. Therefore, a clear account from an independent carer of the context and history of the symptoms is necessary to exclude the likelihood of external reward such as avoiding a noisy shopping mall in a person with ID and autism with sound sensitivity and social anxiety, in which case the individual would be malingering. There are reports of FD imposed on the self in persons with mild ID [56, 57], but this is not likely to occur in people with more severe levels of ID as they would not be able to plan and instigate the signs or symptoms. There is a case report of a person who did not have ID who feigned symptoms of ID, depression and psychosis [58]. There is no evidence in the literature that self-injurious behaviour (SIB) occurring in ID has the intent to deceive although some instances might be to gain care and attention [59]. There is no evidence regarding the presentation of self-induced FD in children or adolescents.

> Persons with ID and ID comorbid with ASD with high support need are likely to be medically naïve and produce more obviously fabricated signs or inconsistent symptoms than persons of the general population. Thus, it is essential for the diagnosis to determine that the intentional production of signs and symptoms is in the absence of any obvious reward such as care, attention or avoidance of annoying stimuli.

23.8 Other Specified (or Unspecified) Somatic Symptoms and Related Disorders

23.8.1 Criteria and Clinical Features

These diagnostic categories are used when there are some symptoms of SSRD causing impairment and distress which do not fulfil the criteria for SSRD. A woman who believes she is pregnant when this is not physiologically

so ("pseudocyesis") might be diagnosed in this category and has been described as occurring in women with mild ID. In general, these categories should be avoided, particularly the one on unspecified somatic symptoms, which is suggested by the DSM-5 [1] to be used with caution also towards the general population. However, the lack of a sufficient and comprehensive history and information from carers and limited language ability in persons with ID might make this diagnostic category the only feasible one in a person with more severe levels of ID who have at least sufficient communication ability to indicate somatic symptoms [44].

23.9 Treatment

There is a lack of specific literature on the treatment of SSRD in ID, but the approaches to their treatment in the general population are likely to be relevant provided the cognitive and communication difficulties inherent in ID, the general health and effects of any known genetic causes of the ID, the environment of care and the social, cultural and educational/daily activity context are taken into account.

The first and essential component of management is a comprehensive biopsychosocial assessment including a medical assessment as indicated and symptom history and observations from parents or carers. As far as possible, the patient needs to experience that they are being taken seriously and their distress and symptoms are understood. The social, cultural and environmental context of symptoms must be clear including a history of stressful life events and social and economic adversity. If possible, and using appropriate communication methods such as visual aids, the link between symptoms and psychosocial or medical illness experiences as well as the experience of anxiety should be explained. Reassurance is therapeutic particularly if symptoms are tackled early in the course of the disorder.

Parents and carers need to be included in the treatment and understand the link between symptoms and medical illness and environmental factors. Management skills training, support and respite care services and treatment of any mental health problem in the parent that contributes might be necessary. Consultation with the network and system of services will inform others and support their care of the individual. Any underlying medical problem such as dental pain or gastric reflux requires treatment. Support and changes in the environment that might reduce anxiety, reward adaptive behaviour and improve positive social engagement and daily occupation are therapeutic and help the individual to cope with their situation. Treatment of anxiety using modified cognitive approaches such a picture-based health education, positive behaviour scripts, behavioural strategies using rewards and relaxation exercises adapted to suit the individual's level of understanding may be therapeutic [60, 61]. These strategies require observation and understanding of the context and antecedents and consequences of the symptom. There is a single case report of a man with ID who was successfully treated for symptoms of excessive medical complaints that were consistent with the diagnosis of an FD using non-contingent rewards [62]. Medication should only be used if symptom profiles are consistent with their indicated use, for example, a trial of an antidepressant if there are symptoms of a comorbid depressive illness [61]. For the treatment of anxiety and depression, tricyclics and selective serotonin reuptake inhibitors (SSRIs) might be of therapeutic benefit but using initially low doses as sensitivity to side effects can be prominent [63]. Benzodiazepines should only be used briefly and with caution given tolerance. Propranolol can be useful in highly aroused patients. Analgesics should remain simple and non-addictive, such as paracetamol, given that chronic pain is generally unresponsive to analgesics where the treatment focus needs to be on rehabilitation and behavioural management.

Early intervention is the key because the prognosis for chronic SSD is generally poor, in which case the focus must be on limiting further deterioration and prevention of medical complications, psychiatric comorbidity and avoidable side effects of psychotropic

medication. Response to a management plan is best documented in a systematic way using behavioural checklists sensitive to change and for use in people with ID such as the developmental behaviour checklist for children and adolescents [45] or adults [46].

Tip

Early multimodal intervention has to be favoured, including individual and family psychoeducation, environmental support and changes, tailored cognitive and behavioural strategies. Medication should only be used if symptom profiles are consistent with their indicated use, for example, in case of unmanageable anxiety or depression.

23.10 Case Example

"Sarah" a 26-year-old woman with moderate ID due to Williams syndrome (WS: loquacious but poor comprehension and gross/fine motor skills deficits) fell off a placid horse when on a day programme group activity. A number of young men in the group laughed at her. She suffered bruising and swelling of her right forearm but emergency room examination and detailed X-rays revealed no fractures. A cold compress and sling were applied and she was returned home to the care of her single parent mother. Over the next 6 weeks, she became increasingly fearful and distressed, constantly stating that her arm was "broken" and that it was "so bad" that it would have to be "chopped off". She refused to attend her day programme, isolated herself in her room, her longstanding insomnia (related to her WS) worsened, she cried frequently in distress and refused to use her right arm and hand claiming that her "fingers wouldn't work" necessitating spoon feeding by her increasingly worried mother. Sarah frequently demanded to see her general practitioner or "go back to hospital". General practitioner review and repeated X-rays failed to demonstrate a fracture and the bruising had resolved. A diagnosis of SSD was made taking into account the behavioural phenotype of WS with its risk of phobias and anxiety.

Treatment which was successful included helping the mother develop strategies to manage her daughter's behaviour and manage her own stress and the provision of periods of daily in-home respite care. Sarah received coaching in modified relaxation exercises; a reward programme of her favourite small figurines for a graded removal of the sling and increasing arm and finger movements supported by a detailed picture script; a picture account of the events surrounding the accident, the visit to hospital and the normal X-ray, and recovery of her arm and return to normal activities; a get-well card from her day programme group indicating that they missed her and the promise of tickets to see her favourite pop group when she was better and had her arm and fingers moving again.

Key Points

- Somatic symptoms which adversely impact daily life are common affecting around 10% of adults and 14% of children with ID.
- Somatic symptoms are often a feature of anxiety, depression and acute stress reactions in persons with ID but can also occur independently as the prime symptom of several specific disorders defined in the DSM-5.
- SSD has somatic symptoms associated with persistent and distressing thoughts and emotions related to the symptoms.
- IAD presents as incapacitating anxiety about health concerns and fears that a serious illness will develop.
- CD presents with neurological symptoms that are inconsistent with any neurological disorder.
- PFAMC presents with an abnormal or excessive psychological response to a medical condition that then adversely affects the outcome of that condition.
- FD presents with fabricated symptoms that intend to deceive others.

- Diagnosis of these conditions in individuals with ID becomes more problematic if not impossible as communication skills decrease with more severe levels of ID.
- Behavioural observation, history from parents or carers, medical, cognitive, cause of ID, environmental, social and cultural context assessment are necessary to inform diagnosis and management.
- Early intervention promises the best prognosis.
- Management is usually multimodal including environmental adjustment, modified cognitive (e.g. psychoeducation) and behavioural strategies with examination of antecedents and consequences of the somatic symptom (e.g. relaxation exercises, contingent and non-contingent rewards), anxiety reduction strategies (e.g. reassurance, modified psychological treatments), anxiolytic medication, treatment of any comorbid mental illness, such as depression, parent/carer support, education and skills training, and wider system consultation.
- Systematic follow-up and review of response to treatment are recommended.

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Obsessive-Compulsive and Related Disorders

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Contents

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Leaning Objectives

Recent neuroscientific research in genetics, neurophysiology, functional neuroimaging, and neuropharmacology are broadening our understanding of the complex pathophysiology of OCRD. These new findings are also providing new insights into the complex interrelationships between OCRD, ASD, ID, and other neurodevelopmental disorders. In this chapter, the focus is on the synthesis of new research into OCRD with our growing knowledge about the psychopathology in the context of neurodevelopmental disorders. One approach to this problem involves searching for neurobiological endophenotypes to define various subtypes of OCRD. These subtypes can specify more focused treatment algorithms.

Upon completion of this chapter, the reader should be able to demonstrate the following:

1. Discuss the relationship between OCRD and ASD, ID and other developmental disorders, and the application of this knowledge to treatment planning.
2. An understanding of the basic neuropharmacology of OCD and its psychiatric comorbidities and applying these ideas to treatment decision-making.
3. The use of subtypes of OC-related behaviors to aid the differential diagnosis and treatment of stereotypies, complex tics, self-injury, and ritualized behaviors.
4. A working knowledge of alternative treatments for treatment refractory OCRD.
5. An enhanced ability to recognize, differentially diagnose, and treat patients with OCRD plus ASD and ID.

24.1 Introduction

Obsessive-compulsive disorder (OCD) is a heterogeneous syndrome characterized by (1) intrusive ego-alien thoughts or experiences, (2) varying degrees of insight, and (3) a relatively limited repertoire of avoidance behaviors (compulsions) [1–4]. Behaviorists conceptualize this phenotype in terms of avoidance responses (compulsions) to aversive thoughts and emotional preoccupations (obsessions).

The capacity of these behaviors to reduce anxiety or distress in turn negatively reinforces their continued utility [5–9]. In some patients, the long-term repetition of the stimulus response cycles metamorphose into nongoal-directed behaviors commonly labeled as habits [3, 4, 6].

The publication of the DSM-5 (2013) and the DM-ID-2 (2016) replaced OCD as an anxiety disorder and established a free-standing category of obsessive-compulsive and related disorders (OCRD), which has been adopted also by the last edition of ICD-11 [94]. This step expanded the scope of OCD to include an addition group of complex repetitive cognitive and behavior disorders. These disorders differ from anxiety-driven OCD in terms of motivational state, semiology of repetitive behaviors, and a different set of co-occurring psychiatric disorders [2, 6, 10].

These changes fell short of making significant revisions in the descriptive/categorical approach to diagnosis and shifting the focus of diagnostic criteria to incorporate pathophysiology and aetiopathogenesis. This leaves us with the problems of overlapping of gene markers, biomarkers, clinical symptomatology, and boundaries issues between OCRD, intellectual disability (ID), autism spectrum disorder (ASD), and ASD +ID [6, 10, 95, 96]. The goals for this chapter center on trying to improve our understanding of the effects of such interrelatedness on the predisposition, precipitation, and perpetuation of OCRD. To explore these issues, we need to modify our conceptual model of OCRD that treats OCRD, ASD, and ID as three discrete disorders following separate clinical trajectories and focus instead on them as a mosaic of entangled neurodevelopmental disorders that change throughout the life cycle. As a result, treatment planning and implementation need to focus not only OCRD as a psychiatric disorder but how the presence of ASD and ID influence the interventions. Our goal is to look carefully at the high rates of incomplete treatment responses and tease out how co-occurring ASD and ID influence these outcomes.

This process is a step toward synthesizing a model for personalized treatment and improving treatment outcomes for nearly 30% of patients with OCRD.

24.2 Diagnostic Criteria

The process begins with an exploration of our current descriptive/categorical diagnostic criteria outlined in the DSM-5 [1] and DM-ID-2 [10]. The following criteria are required:

A. Presence of obsession, compulsions, or both Obsessions defined by 1 and 2,
   1. Intrusive, unwanted, and distressing recurrent thoughts, urges, images
   2. Attempt to ignore or suppress or to neutralize them with some other thought or image and Compulsions defined by 1 and 2:
      1. Repetitive behaviors (handwashing, ordering, checking) or mental acts (praying, counting, repeating words silently) that the individual feels is driven to perform to an obsession according to the rules that must be applied rigidly.
      2. The function of these behaviors or mental acts involves eliminating or at least reducing anxiety or distress, preventing some dreaded event or situation; the actions are not connected in any realistic way to what they are intended to neutralize or prevent.

B. Obsessions and compulsions are time consuming (>1 h per day) or cause significant distress or impairment in important areas of functioning.

C. Not attributed to physiological effects of substance of abuse, medication, or other medical conditions.

D. Not better explained by symptom of another mental disorders; this includes other OC-related disorders, paraphilias, substance use disorders, impulse control and conduct disorder, mood or schizophrenia spectrum disorders.

Also in the ICD-11, essential diagnostic features are represented by persistent obsessions and/or compulsions. Obsessions are defined as "repetitive and persistent thoughts, images, or impulses/urges that are experienced as intrusive and unwanted, and are commonly associated with anxiety. The individual typically attempts to ignore or suppress obsessions or to neutralize them by performing compulsions." Compulsions are described as "repetitive behaviours or rituals, including repetitive mental acts, that the individual feels driven to perform in response to an obsession, according to rigid rules, or to achieve a sense of 'completeness'. Examples of overt behaviours include repetitive washing, checking, and ordering of objects. Examples of analogous mental acts include mentally repeating specific phrases in order to prevent negative outcomes, reviewing a memory to make sure that one has caused no harm, and mentally counting objects. Compulsions are either not connected in a realistic way to the feared event or are clearly excessive" [94].

Specifiers

Specifiers are a major part of the DSM-5 criteria for OCRD. They define two phenomenological endophenotypes characterized by the presence or absence of both insight and the presence of tics/tic disorders. According to the ICD-11, "individuals with OCD vary in the degree of insight they have about the accuracy of the beliefs that underlie their obsessive-compulsive symptoms. Although many can acknowledge that their thoughts or behaviours are untrue or excessive, some cannot, and the beliefs of a small minority of individuals with OCD may at times appear to be delusional in the degree of conviction or fixity with which these beliefs are held. Insight may vary substantially even over short periods of time, for example depending on the level of current anxiety or distress, and should be assessed with respect to a time period that is sufficient to allow for such fluctuation (e.g., a few days or a week)." [94]. Hence, insight is a requirement for OCD although developmental studies suggest that children with early-onset OCD frequently lack both self-awareness or the ability to perceive obsessions and compulsions as ego dystonic.

Unfortunately, many adult patients also lack insight or ego dystonicity. This discrepancy creates a potential boundary problem

with delusional thoughts. Resolving this issue requires a more detailed investigation into the tenacity and rigidity of these beliefs, their resistance to contradictory evidence, and relationship to psychosis [1, 6, 10]. Yet these distinctions are not absolute since the co-occurrence of OCD, delusional disorders, and schizophrenia is well established. The presence of ASD creates a similar quandary. It can be difficult to distinguish OCRD from the core features of ASD and ID-repetitive and restrictive interests, cognitive rigidity, intolerance of change, and egocentricity due to subtle deficits in theory of mind skills [1, 6, 10].

The presence of tic disorders creates a second subgroup of OCRD that overlaps both the phenomenology of OCD and the relationship between tics and OCD-related disorders. Both specifiers address basic differences in clinical phenomenology as well as neurobiology and responses to psychotropic medications [6, 11, 97, 98, 99]. These relationships are subject to more careful scrutiny later in this chapter.

There are three basic components to the differential diagnosis of OCRD. These function mainly as rule-outs:

1. The persistence of rituals is well past their normal neurodevelopmental time frame. This refers to childhood preoccupations and ritualized behaviors [1] (see section on OCRD as a neurodevelopmental disorder).
2. OCRD is comorbid with the following neuropsychiatric disorders: anxiety disorders, major depressive disorder, trauma-and-stressor-related disorders, eating disorders, tics, stereotypic movements, psychotic disorders, ASD, and substance use disorder. The neurobiology of these relationship varies in part along the lines of anxiety-related and nonanxiety-related subsets of OCRD.
3. OC-like symptoms are associated with a wide range of medical, neurological, metabolic, genetic, neuroimmunological/autoimmune disorders, traumatic brain injury, and other acquired brain disorders. The secondary forms of OC-like symptoms involve the many interconnections within the corticostriate and corticolimbic neurocircuitries.

OC behaviors in patients with tic disorders frequently have more in common with OCRD, in which intrusive and disturbing experiences differ from the doubts and intolerance of uncertainty and fears of contamination associated with anxiety-related OCD. In many cases, there are no specific obsessions nor compulsions that allow the individual to avoid the adverse outcomes associated with the patient's noncompliance with the rituals. In tic-related OC symptoms, there are differences in the motivational state that are oriented more toward approach/appetitive behaviors in which interference with the ritual leads to an intensification of the drive to complete it. There is more frustration than anxiety.

In addition, "feeling right" rather than less distressed may serve as the terminating event for the repetitive motor or cognitive actions [1, 26].

In addition, there are subtypes of OCRD associated with stimulant abuse, traumatic brain injury, autoimmune (systemic lupus and PANDAS), neurodegenerative and other movement disorders (Parkinson's and Huntington's disease), genetic/metabolic disorders (neuroacanthocytosis and Wilson's disease), more recently co-occurring with abnormal movements associated with anti-neuronal antibody syndromes, and mitochondrial disorders. The semiology of these secondary forms of OC-like behaviors may also lack obsessions, insight, or dystonicity [6, 28, 29, 30].

Patients with a history of traumatic brain injury during infancy display a global pattern of deficits that can resemble ID, but less so for ASD. These adult patients also present with neuropsychological deficits that can complicate the diagnosis of OCRD. Perhaps the most dramatic include environmental dependency (echopraxia, echolalia, and catatonic-like imitative behaviors), perseveration, negative affective reactions to environmental changes (intolerance of change), hyperactivity, irritability and impulsivity, lack of insight, relative intolerance for change, catastrophic anxiety/cognitive disintegration, memory and working memory deficits, and a range of fixed responses and behavior (executive deficits).

Unfortunately, this group may be the most vulnerable to dysfunctional family systems,

ongoing psychological trauma, and disorganized living environments and poorly structured daily routines. Individuals with co-occuring neurodegenerative disorders commonly display neurocognitve decline in conjunction with progressive changes in repetitive behaviors. When the decline is subtle, extended longitudinal follow-up may give the clinician an opportunity to observe progressive ongoing deterioration. Many frontotemporal dementias may present with increases in perseverative behaviors than be overlooked in the face of preexisting patterns of repetitive behaviors. The loss of daily living skills or progressive apathy may require time to emerge [3, 62, 63].

Many early onset psychiatric disorders are associated with neurodevelopmental comorbidities. Both influence the actively developing brain. Like ASD/ID, many of these disorders influence neuroplasticity, neuronal migration, and the development of cortical specialization and coherence. For example, repeated trauma can increase the risk of sensitization and epigenetic changes during critical periods of emerging neurocircuitry. Once these epigenetic changes in promoter regions are methylated and switched off, the likelihood of spontaneously reversals lessens, as does their impact on future interactive specialization and brain maturation [3, 63, 64, 100].

Many "organic forms" of OC behaviors resemble impulsive-compulsive behaviors that include addiction-like behaviors. These include disorders of excessive grooming, repetitive counting, arranging, organizing, need for symmetry and intrusive images, or thoughts involving sex, violence, and destructive behaviors [1, 6]. These appear analogous to another group of automatic (habits) repetitive behaviors that resemble addiction behaviors and tic-related and ASD repetitive behaviors (classified as nonsubstance-related disorders) [1, 4]. These expanded phenocopies interface with personality disorders, bipolar disorder, ASD, other movement disorders, and stereotypic movement disorder (stereotypies and self-injurious behaviors) [6, 11].

Lastly, OCRD also include: type unspecified (subsyndromal), body dysmorphic disorder, hoarding, excoriation (skin picking) disorder, and trichotillomania (hair pulling) in both primary and secondary forms. The secondary disorders include substance-induced and OCRD associated with medical, neurological, immunological, metabolic, and neurodegenerative disorders [1] (Table 24.1).

OCRD also includes other specified OCRD that consist of body dysmorphic disorder (BDD), nail, and cheek biting and differentiate these behaviors from other forms of high-frequency/low-intensity forms of compulsive self-injury. This group also includes obsessional jealousy and several culture-specific disorders. Unspecified OCRD is analogous to a subsyndromal condition that does not meet the criteria threshold for OCRD. Nevertheless, approximately 50% of patients classified as unspecified OCRD will progress to the full syndrome. Each of these subtypes share an increased risk for comorbid mood and anxiety disorders [1, 6, 10].

This section on diagnostic criteria reviews the updated version of the diagnostic criteria spelled out in the DSM-5 and Diagnostic Manual Intellectual Disability-2 (DM-ID-2). The reader should be alert to the implications of a shift from anxiety disorders to a free-standing category, obsessive-compulsive and related disorders. The second change involves the addition of repetitive behaviors that differ from traditional doubting/checking and contamination/cleansing rituals in terms of their relationship to obsessions, motivation, and functional states.

24.3 The Relationship Between OCRD, Repetitive/Stereotypic, and Addiction Behaviors

The DSM-5 and DM-ID-2 classify a variety of repetitive behaviors among the nonsubstance-related disorders or stereotypic movement disorder. As we shall see, complex tics and some forms of stereotypies/self-injury may lie midway between OCD and habits. Habits are automatic behaviors that may no longer appear directly linked to a subjective sense of pleasure or reward [5, 6]. The

Table 24.1 OCRD-related disorders includes

Diagnostic criteria	Body dysmorphic disorder [38–40]	Hoarding disorder [41]	Trichotillomania TTM [44]	Excoriation disorder [101]	Substance/medication-induced [1]	OCRD due to other medical condition [42]
A.	Preoccupation with one or more perceived flaws or defects-not observable by others	Persistent difficulty discarding or parting with possessions, regardless of actual value	Recurrent pulling of one's hair, resulting in hair loss	Recurrent skin picking, causing skin lesions	OCRD-core symptoms	OCRD-core symptoms
B.	Repetitive checking, grooming, skin picking, reassurance seeking, mental acts, comparison with others	Need to save the items and distress associated with discarding them	Repeated attempts to decrease or stop hair pulling	Repeated attempt decreases or stops	Evidence of substance use/abuse temporally associated with symptoms	Evidence of medical/neurological condition directly related to symptoms
C.	Creates significant distress, impairment of social function	Accumulate possessions, congest or clutter living areas, compromises their intended use	Causes significant distress; impairment of social, occupation, or other areas of functioning	Causes significant distress, impairment of social occupational activities	Not better explained by OCRD, delirium, functional impairment	Not better explained by OCRD, delirium
D.	Not better explained by eating disorders	Significant distress. Not attributed to other medical condition; not related to ASD or other psychiatric disorders	Not attributed to medical or other mental conditions	Not due to substance abuse or other medical and mental disorders	Specify: – specific drug; – onset during intoxication, withdrawal; – onset after medication use	Specify type of disorder
E.	Specify: – with muscle dysmorphia; – with fair to good, poor, and absence of delusional beliefs	Specify: – with excessive accumulation; – insight				

boundary between habitual and addiction behaviors can be confusing. Addiction behaviors are repetitive behaviors characterized by increasing urges to carry out a ritual without signicant anxiety or avoidance behaviors. Once the repetitive behavior is completed, the urge temporarily subsides. The initial sense of relief wanes and the urge returns.

Some patients cannot escape the cycle and seem bound to repeating self-defeating or self-destructive behaviors. Several key features of this subgroup arise longstanding re-enactments and repetition:

1. Sensitization of neuronal circuits in a process analogous to kindling of stimulus-induced seizures.
2. Because of the potentially self-defeating properties, some patients experience a sense of guilt or dysphoria associated with gratifying the urge and in some patients, a sense of psychological withdrawal.
3. The cycle of dysphoria and a return of the motivational state contribute to the persistence of these repetitive behaviors. From a functional behavioral model, the original triggers may generalize to include multiple specific and context-related cues as well as internal affective states that can continue in spite of severe adverse consequences [2, 3, 4].

These repetitive behaviors differ from other of nonsubstance-related disorders in terms comorbidity. Many related disorders are less likely to present with comorbid anxiety disorders (and OCD). For example, nonsubstance-related disorders are more likely to occur in the context of dysfunctional impulse control or substance use disorders [6, 10].

Differentiating these subgroups of repetitive behaviors from other stereotypic movement disorders can be challenging especially in patients with co-occurring ID or ASD [6, 10]. This problem seems most obvious for mannerisms and stereotypies associated with the core features of ASD/ID. Similar difficulties arise when the clinician confronts patients with ASD/ID and co-occurring complex tics, hoarding behaviors, and in some cases, perseveration associated with other neurocognitive disorders.

There is a larger subset of repetitive thoughts and behaviors that are germane to our understanding of the relationship between OCRD and autism spectrum disorders (ASD) and intellectual disability (ID). These differences overlap motivational states (goal-directed behaviors) and underlying neurobiology. These subtypes may partly explain the variability in both behavioral and psychopharmacological treatments.

24.4 Boundary Problems Created by Co-occurring Neurodevelopmental Disorders

In many settings, clinicians may overlook the fact that many psychiatric are complex genetic-neurodevelopmental disorders. This combination creates some challenges. For example, familial ASD (multiplex families) is a highly heritable, polygenic disorder. Symptomatic ASD on the other hand is more likely to be associated with multiple genetic, neurological, metabolic, early infectious, and toxin-related conditions. This latter group is also more likely to co-occur with ID. The origins of both subtypes occur during critical developmental periods (embryological and early postnatal insults) that effect very early brain organization and maturation. Many of the genes linked to ASD also affect synaptogenesis, neuroplastic changes in the neuropil, stabilization of these new synaptic connections, and interconnections between newly developing cortical-subcortical/limbic and striatal networks. These changes seem to guide future attachment, temperament, neurocognitive, emotional, and social development [7, 61, 65, 67].

However, most psychiatric disorders emerge during later stages of brain development. For example, trauma-related insults during early childhood disrupt the entrainment of the stress-response system as well as later episodes of neuroplasticity, neurogenesis, myelination, interactive specialization, and the development of hierarchically organized frontal-striatal and fronto-limbic

neuronal networks. During early childhood, these insults may not result in ASD or ID but may adversely affect the emergence of affect regulation, attention, executive functions, and other higher cortical functions [16, 37, 61, 73, 74]. A close look at the neuropsychology and neurobiology of OCRD reveal a mean age of onset around age 10, as well as neurocognitive deficits in spatial orientation, information processing, and decision-making and executive functions (including working memory) [6, 11, 15, 102].

Neurotypical children express transient rituals, repetitive behaviors, and developmental, age-dependent magical thought. The typology of early OCRD differs from the typical childhood worries and rituals. However, some children fail to resolve these worries but never develop OCRD. Children with mild ID are likely to fit into this group. Recent evidence suggests that the childhood precursors of OCRD usually differ from the beginning. The worries and behaviors among children with OCD belong to a more restricted repertoire of concerns and behaviors. In addition, children with early onset OCRD may lack insight, display more compulsions than obsessions, and may present with comorbid externalizing and internalizing disorders [6]. Neurotypical and children with OC symptoms differ from those with ASD in terms of complexity, persistent of need for sameness/reduced tolerance of change and limits on the richness of imagination, social communication, and shared, social expression (imaginative social play or age-related fiction) [6, 11, 15].

As alluded to earlier, there is a subgroup of preschool males who present with transient motor and/or vocal tics alongside ASD, ID, and other neurodevelopmental disorders [6, 18, 19]. These young males present OC symptoms characterized by fewer obsessions, limited insight, and a cluster of arranging, ordering, counting, and symmetry rituals. These OC behaviors give way to tic disorders in between 20% and 40% of the original cohort. Others may present with both tics and OC symptoms at the outset. This group will also experience fewer obsessions, less anxiety, fewer avoidance behaviors, and later onset problems associated with changing typologies of tics. A smaller percentage develops Tourette disorder (TD) in which both motor and vocal tics are present.

Within the first 5 years after TD onset, nearly 50% develop intrusive thoughts, urges, and sensory experiences that are relieved by ticqueing. These children may also develop sequences of multiple tics that terminate when the urge or sensation abates ("feels right"). At this stage, the boundary between tics and OCRD becomes more difficult to establish. Careful factor analytics offer some help. These studies suggest that complex tics differ from compulsive behaviors in anxiety-mediated OCD. In contrast to fears of contamination/cleaning and doubting/checking, tic-related, repetitive behaviors include touching, arranging, organizing, counting, and need for symmetry. Tic-related OCRD also display different patterns of impulse regulation and comorbidity with other neuropsychiatric disorders [11, 20–22].

Similar patterns of tic-related repetitive behaviors also occur in patients with ASD and ID. The higher prevalence rates for tic disorders in ASD and ID than in the general population underscore the significance of these data [11]. The entanglement of boundaries between OCRD, tic disorders, and ASD/ID phenotype confounds attempts to track their clinical trajectories. In addition, the severity of OCRD and TD tend to worsen during periods of increased stress, and wax and wane over time. This inherent variability challenges attempts to monitor diagnostic fidelity and treatment efficacy. To do so, requires ongoing assessment. However, it can be difficult to sort out these variables, and this process may take repeated examinations over an extended period of time [23–28].

In general, childhood onset OCRD with and without tics may differ in terms of intrusive sensory experiences, sexual, and violent images, as well as urges and cravings. Embedded in this group, however, are patients with OC-like repetitive behaviors that are difficult to characterize. This in part is due to a relative absence of readily discernable patterns of contingency-driven behaviors (avoidant/escape behaviors), less overt anxiety, and tendency toward negative affective or behavioral

displays in response to limits. These problems appear more habit-related behaviors, and tend to occur without readily discernable reinforcers [4, 6, 11]. Unfortunately, the enhanced hedonic drive associated with these behaviors can give way to an addiction-like pattern that continues in spite of significant negative consequences (e.g., some forms of severe self-injury). Their waxing and waning course may also increase the chances of a bipolar disorder diagnosis [6, 26, 28].

Even with this subtype analyses, there are still important issues that shape the boundaries between OCRD and repetitive/ritualized behaviors. Our next step is to delve into issues linking OCRD to more complex neurodevelopmental disorders such as (Autism, ID, ADHD) and other neuropsychiatric disorders (tic disorders, other medical/neurological syndromes). The central theme in this analysis is to explore the effects atypical development in the mergence and presentation of OCRD.

24.5 Neuroethology of OCRD

Tip

Neuroethological models suggest a relationship between conflict mediated behaviors in animals and many human repetitive behaviors. Think of the neurobiological similarities between OCRD in humans and ritualized behaviors in animals.

Our current understanding of the transformations from stereotypies, into self-defeating, habitual behaviors remains a work in progress. Neuroimaging studies of these habit-related behaviors suggest a pattern of overlapping, dysfunctional fronto-striatal (caudate), ventral striatal, palladial, thalamocortical ensembles substrates [4, 6, 11, 29, 103]. These networks differ from the ancient fear/anxiety-aversion conditioned behaviors that underlie OCD and other anxiety disorders. Studies of the anxiety-conditioned group suggest involvement of the basal-lateral amygdala, basal striatum, hippocampus, limbic and insular, brainstem, and several orbitofrontal regions. This network also underlies trauma- related anxiety disorders [32–34]. The fear pathway is highly conserved in evolutionary terms. However, mammals, and especially primates (especially humans), have a more complicated fear/anxiety network that is interconnected with several late evolving, cortical networks. These pathways involve higher-level processing and interpreting cortical input that plays a role in regulating and planning cognitive and behavioral responses [6, 32, 35].

Humans are also social primates who evolved an elaborate, finely tuned, social brain that remains highly adaptable to social contexts. The prolongation of neuroplasticity, neurogenesis, and ongoing myelination in modern humans permit ongoing modifications of late-developing networks that underlie language, social communication, abstract thought, more complex social behaviors, and a new range of executive functions (interactive specialization). ID and ASD may represent complex patterns of dysfunctional maturation in these developmental lines [6, 32, 35].

OCRD may represent more system-specific malfunctions that release mammalian patterns of ritualized social behaviors. Neuroethologists and evolutionary biologists take this approach. They expand the ideas to humans in order to link OCRD to "fixed action patterns" with species-specific, trigger stimuli ("innate releasing mechanisms") in ambiguous or conflictual, social situations. In humans, the enhanced top-down regulation provided by fronto-cortical expansion overrides automatic responses associated with instinctual behaviors. Metaphorically, OCRD is a dysregulated remnant (disconnection syndrome) that permits the expression of these attenuated species-specific behaviors (fear, fear-conditioned influence). In addition, dominance hierarchies may underlie social anxieties and perhaps some disorders of excessive grooming [6, 36–38, 62].

Although neglected in most diagnostic models, neuroethological principles provide observational social data to the assessment. This information is useful in the study of disorders excessive grooming, ritualized social defeat behaviors associated with OCRD and social anxiety. In animal models, these social

behaviors occur in socially ambiguous situations that relate to conflicts over behavioral expectations, dominance, territorial, and courtship rituals. Of course, these animal models are only analogs of OCRD. Nearly six million years of primate evolution separate us from our closest relative, the chimpanzee. During that time, humans evolved a much more sophisticated brain that exercises greater cortical regulation of rituals and automatic behavior. From an evolutionary biology perspective, OCRD may represent dysfunctional top-down neocortical regulation. Developing a more expansive understanding, its neurophysiology and neuropharmacology can provide additional insights into OCRD [6, 49, 50, 51].

An understanding of neuroethology can provide insights into how we may adapt current findings in the developmental neuroscience to the broader concepts of diagnoses in terms of hypothesis testing. The neuroethological model has obvious limitations but may provide useful tools for understanding OCRD in individuals with severe developmental disorders.

24.6 Neurophysiological Models: Search for Biomarkers and Endophenotypes

For many years, our understanding of OCD focused almost exclusively on psychoanalytic concepts such as ego dystonic, anxiety disorder, and preserved insight. A major change to this approach began with the Research Diagnostic Criteria and the DSM-III. Since that time, the authors of DSM-5 and then the DM-ID-2 expanded this model in two ways. The first involved creating a separate category (OCRD) and the second was to graft other repetitive behavioral disorders onto anxiety-related OCD. This new category includes disorders of dysfunctional grooming behaviors (body dysmorphic disorder, trichotillomania), aberrant attachment behaviors, working memory deficits, intolerance of change/insistence on sameness (hoarding), and a group of minor self-injurious behaviors (skin picking, nail biting) [6, 40, 44, 45].

OCRD excludes nonsubstance abuse disorders such as gambling, compulsive shopping, sexual behaviors, and Internet use. These behaviors resemble addiction behaviors and other habit-related disorders. They differ from other OC behaviors in terms of clinical semiology, patterns of increased arousal (skin conductance), and regional changes in activation related, functional neuroimaging [47–50]. This group also includes patients with complex tic disorders that may wax and wane in repetitive cycles that are occasionally confused with bipolar disorder [4, 6, 11, 21, 46].

The clinical application of these pathophysiological findings leads us to modify assessment and treatment by specifying a mechanism of action (pathophysiology), identification of particular genetic and physiological features (biomarkers) rather than relying exclusively on syndromal psychiatric disorders (Phenomenological phenotypes). There is growing research interest in dissecting syndromes into component parts, and adding biomarkers with pharmacogenomics to diminish this frustration. In addition, applying both drug-specific pharmacokinetics and pharmacodynamics can assist in designing a more personalized plan of care. Accomplishing this goal requires matching more specific medication regimen-based features such as threshold for arousal, intensity of negative affective states, and level of stress tolerance, impulse dyscontrol, and the degree of sympathetic arousal needed to initiate and sustain avoidance behaviors into the treatment of OCRD [51, 52].

In summary, these subgroups of OCRD represent variations in approach/avoidance, learning models (types of conditioned stimuli), vulnerability to overgeneralization of avoidance behaviors, and the metamorphosis of some goal-directed repetitive behaviors into habits [6, 11, 32]. A comparison between contamination-motivated handwashing and trichotillomania or skin picking reinforces these differences. Close factor analysis reveals not only differences in overt behaviors but also variations in genetic risk factors, functional neuroimaging, relationship to ID/ASD, sensitivity and response to selective serotonin reuptake inhibitors (SSRIs), and efficacy of

CBT-exposure-response prevention/habit-reversal therapies [52, 53]. Differentiating subsets of OCRD in this fashion can result in a treatment plan that more closely matches to individual patient needs.

24.7 The Use of Research Domain and Intermediate Endophenoytypes

The heterogeneity of OCRD, ASD, ID, and ASD/ID creates many problems for assessment and treatment. Most of our existing diagnostic protocols depend on descriptive/phenomenology and do not address issues related to aetiopathogenesis. This section provides brief description of the genetic, neurobiological, neuroimaging, and neuropharmacological underpinnings of neurodevelopmental disorders in clinical/research settings.

The following bullets outline an attempt to pull together some of the many data sets from recent neuroscience. This algorithm may be useful in organizing data:

1. Syncretize the expression of clinical subtypes with temperamental style such as harm avoidance, low novelty seeking, adaptability [2, 6] with the complex social ecology of a family, and social environment. These factors influence the age of recognition, nature and severity of symptoms, predictability of clinical course and treatment response, as well as the entangled biopsychosociology of co-occurring OCRD, ID, and ASD.
2. Address clinical subtypes of OCRD in terms of neuropsychological data. These data focus on conflict and error monitoring; response inhibition; integration and regulation of motor output; task switching and reversal, assessment of risk, coping with uncertainty; reward processing and impaired shifting, inhibition of intrusive thoughts, experiences and urges (input gating), impaired decision-making (require too much evidence to act); working memory issues; and other executive functions. Each of these traits is associated in some way with an OCRD phenotype [18, 27, 28, 33].
3. Apply data from neuroimaging studies that help define anatomical and functional changes in the corticostriate and limbic networks. The patterns observed in OCRD suggest a disconnection between the regulatory functions of the medial prefrontal, orbitofrontal, cingulate, and reward pathways in goal-directed behaviors. The transition from goal or contingency directed to habit formation centers on the role of the caudate in this process [17, 25, 90, 91].
4. Address the role of multiple neuropharmacological systems involved (especially relevant for treatment-resistant cases):
   (a) Neurotransmitters, neuropeptides (oxytocin, vasopressin), glutamate, gamma aminobutyric acid (GABA), and patterns of neuronal firing. These may increase limbic activity that disrupts the normal homeostatic mechanisms linked to top-down regulation of excitatory/inhibitory networks [11, 12, 14, 92].
   (b) The foundation for these dysfunctional circuits arises is associated with aberrant brain interconnections related to several gene polymorphisms that involve serotonin reuptake (serotonin-transporter-linked promoter region; 5HTTPLR) [11, 53, 65, 104].
   (c) Catechol-0-methyltransferase (COMT) and the whole dopamine metabolism in the frontal cortex play an indirect role in both the direct and indirect dopamine systems (fronto-striate, substantia nigari/globus pallidus (exterior and interior), subthalamic and thalamic nuclei, and their interaction with pathways in the limbic system. The latter may be significant in OCRD-related tic disorders [11, 67].
   (d) Brain-derived neurotrophic factor (BDNF) that is involved in neurogenesis, synaptogenesis, neurophysiological changes in the cortical-striatal; amygdala-hippocampal, cingulate and supplemental motor regions that influence generalization, and extinction of conditioned responses. SSRIs may liberate promoters that increase the production of this nerve growth factor.

This may apply for neuropsychological, behavioral, and pathophysiological models for OCRD and ASD/ID. More effective treatment agents may provide an important contribution to new treatment models [27, 32].

24.8 Neuropsychological and Neurobehavioral Models

Although relevant, the author did not expand upon OCRD and ASD/ID as disconnection syndromes. ASD and ID present challenges to early prenatal development, especially the organizational phases of the eventual hierarchical organization of the sensory, processing, and integrative motor activity. These organizational disruptions arise from neurogenesis, synaptogenesis, axonal growth, myelination, and the effects of ongoing environmental events on brain maturation. Many of these events occur during critical periods of brain development and maturation in which external and internal insults disrupt sequential gene-environment interactions. These affect both interactive specialization and emerging cortical interconnections (coherence).

Later many of these gene-environmental interactions influence the ongoing transformation of many repetitive behaviors (including OCRD) from goal-directed compulsive behaviors (e.g., OC behaviors that permit escape from aversive stimuli, reducing overarousal or coping with understimulation) into a group of habitual behaviors that represent automatic responses to specific cues. This group encompasses OC symptom related to tic disorders and addiction-like stereotypic repetitive behaviors (severe self-injurious behaviors (SIB) and milder forms of impulsive control disorders). These habit disorders are not directly involved with activation of reward pathways [7, 14, 17, 25]. This group differs from OCD/anxiety-driven disorders in several ways. These include treatment responses to SSRIs and cognitive behavior therapy-exposure and response prevention therapy (CBT-ERP), and greater clinical improvement with habit-reversal therapy (HRT) and augmented pharmacotherapies [2, 21].

Other sequential transformations enhance generalization of cue-stimuli (defective pattern separation) and limit extinction learning as a means of overriding these conditioned behaviors. As in kindling studies in induced seizure activity, repeated activation of these pathways influences long-term potentiation (LTP) and depression (LTD). Glutamate evolved as a key player in synaptic spine changes involved in laying down new learning and memory formation. Extinction also involves new learning in the medial prefrontal regions that help override previous dysfunctional conditioned experiences [32, 58, 61, 62, 64, 72]. The role of glutamate in resistant OCRD is an important step in our understating of ASD, OCD-related disorders, and disorders in which extinction and generalization are major players. Unfortunately, many treatment protocols that address the imbalance between excitatory and inhibitory circuits (dopamine, GABA, and glutamate) are not as helpful as anticipated. This small step opens the doorway for a new array of potential pharmacological treatment approaches. The increased use of repetitive transcranial magnetic stimulation (rTMS) and direct current stimulation protocols may prove helpful. The use of opioid antagonists, partial agonists for glutamate, n-acetyl cysteine, oxytocin, and other neuropeptides may have indirect benefit, especially by restoring homeostasis in excitatory/inhibitory dysfunction and facilitating synaptic plasticity necessary for extinction learning and memory [6, 11, 67, 72].

The relationship between brain dysfunction, neurodevelopment, and psychiatric disorders is a complex process that evolves during childhood. The concept of endophenotypes involves looking for core traits that co-occur with many other behavioral/psychiatric disorders. The application of endophenotypes can narrow the heterogeneity gap while at the same time leads clinicians to consider psychiatric and neurodevelopmental disorders as complex genetic disorders. These complex phenotypes appear to represent final common pathways involving multiple genetic and neurobiological factors (endophenotypes).

The overlap between the syndromes suggests that a high degree transdiagnostic relatedness may partly explain the relatively low rates of remission in many current treatments.

24.9 Gaps Between Diagnosis and Treatment Planning

We will review three approaches to the treatment of OCRD in patients with ID and ASD. These basic therapies are psychopharmacological, behavioral, and neuromodulation therapies. Behavioral, cognitive-behavioral, habit-reversal, and pharmacotherapies are more familiar to most clinicians. Neuromodulation therapies include deep brain stimulation, transcranial direct current and magnetic stimulation, and neurosurgical tractotomies. Intervention studies remind clinicians that treatment response rates for most therapies hover around 60–70%, whereas remission rates are in 30% range. Among the incomplete nonresponders, 20% have partial or marginal improvement whereas around 10% continue to worsen. We need novel approaches to close this treatment-nonresponse gap. Currently our best tools are polypharmacy (augmentation strategies), combinations of pharmacological and behavioral therapies interventions, alternative therapies/neuromodulation, and combinations of all three. Unfortunately, the response rates to combined therapies are not dramatically better than monotherapies [8, 16, 19, 37].

Until recently, demonstrating treatment nonresponse was the best way to access neuromodulation therapies. As a result, it is not surprising, however, that a significant number of patients also fail these new treatments. For a select few, deep brain stimulation (DBS; stereoscopic implantations of electrodes) and tractotomies (sectioning key circuits) are useful neurosurgical interventions. Although effective in many cases, these procedures carry significant risks for adverse neurosurgical outcomes [55]. Unfortunately, few patients with ASD or ID receive these neuromodulatory treatments.

As alluded earlier, the treatment approaches to subtypes of OCRD differ based on the following:

1. The presence of anxiety-related with obsessions-compulsions versus habit-related, nonanxiety forms of repetitive behaviors.
2. OC behaviors associated with tic disorders.
3. Secondary OC behaviors related to neurobiological, genetic, metabolic, and neurological disorders.
4. Comorbidity with ADHD (hyperactivity, impulsivity), presence of chronic pain, level of ID, and the presence of other neuropsychiatric disorders.

Each of these subtypes differs in terms of their sensitivity to available therapies. For example, the response rates of CBT-ERP vary based on severity of OC symptoms, co-occurrence with neurodevelopmental disorders like ID, age of onset, and the presence of comorbid major depressive/anxiety disorder and schizophrenia spectrum disorders [52]. Anxiety-related OCRD are most responsive to SSRIs and CBT-ERP. Neuromodulation therapies appear more for patients with complicated OCRD. Nevertheless, there is a rub. The improvement in OCRD relates to improvement in depressive disorders rather than selective benefits for repetitive thoughts and behaviors. Patients with OCRD plus Tourette syndrome are less likely to benefit from SSRIs and/or CBT-ERP but more likely to improve with neuromodulation, neuroleptic/benzodiazepine/SSRIs combinations and habit-reversal therapy (HRT) [35, 52, 55]. The Tic-related group also creates diagnostic problems due to their confusing mixture of sensory and premonitory tics and differentiating involuntary from voluntary movements.

There is gender difference in phenotypic expression of both OCRD and tic disorders. Perhaps the most confusing findings arise from population genetics studies, which demonstrate evidence for a greater level of genetic loading among symptomatic females with TD, but also a female gender bias toward phenotypic expression of OCD rather than tics. The presence of both OC behaviors and Tourette's

disorder appears to represent a more severe form of both disorders regardless of gender. Modifications in treatment are also frequently necessary, but there are limited data about gender differences in response to specific therapies [6, 11, 12, 21]. The prevalence rates of tic disorders tend to be much higher in ASD and ID patients than in neurotypicals [6, 11]. As in other neurodevelopmental disorders, there tends to be a male bias toward ASD without ID, TD, and tic-related compulsions. Each may bias the treatment of OCRD toward combination therapies.

High rates of repetitive behavior such as self-injurious behaviors in patients with ASD occur in the presence of ADHD-like overactivity/impulse dyscontrol. Other factors include the relationship of self-injurious behaviors (SIB), ASD, presence of severe ID, and severity of repetitive-restrictive behaviors. In patients with severe ASD/ID, medical issues (pain) and gastrointestinal distress also increase rates of SIB. This population is less well studied, but it remains uncertain if HRT or CBT-ERP are truly ineffective or is there a selection bias toward behavioral modification techniques or pharmacotherapies overshadow consideration of these therapies. Current data suggest that their cognitive demands pose a barrier to even modified therapies [6, 10].

A second source of diagnostic relates to uncertainties in differential diagnosis. Relative absence of insight and the intrusive nature of aversive images, urges, and emotional/affective states suggest the presence of OCRD + tics. However, by adulthood, tics may no longer be observable or confused with tardive dyskinesia or masked by long-term use of neuroleptic. Many complications are the result of the widespread use of second- and third-generation neuroleptics for aggressive behavior, rage outbursts, property destruction, and SIB. In addition, the presence of trauma-related alterations in stress-response networks and the entanglement of ASD, severe ID, and tics + hyperactivity/impulsivity confound the clinician trying to differentiate OCRD from other forms of repetitive and self-injurious behavior. Each of these conditions can accentuate surges in these target symptoms during periods of psychosocial stress (an example of baseline exaggeration; for a description of the concept of baseline exaggeration see ▶ Chap. 5) [6, 10, 55–58]. In this context, matching treatment options with data from a behavioral analysis or detailed diagnostic workup and diagnosis can be a very challenging task [57, 58].

Additional problems arise with the inclusion of skin picking (e.g., Prader-Willi syndrome), body dysmorphic disorder (with SIB), hoarding (in ASD), trichotillomania (high-frequency/low-intensity, mild SIB), or partial expression of OCRD (specified and unspecified OCRD). Many of these behaviors resemble nonsyndromal self-injury, self-stimulation, and repetitive behaviors associated with ASD and ID [1, 2, 6, 59]. Perhaps the best sources of data are a psychiatric assessment and comprehensive functional behavioral analyses combined with the use of standardized rating instruments. The functional behavioral analysis addresses setting events, antecedents, operationalized description of repetitive behaviors, and a sense of what trigger and maintain these challenging behaviors. There are drawbacks. Most relate to difficulties integrating these divergent treatment models into a comprehensive and coherent intervention.

In summary, diagnoses are hypotheses based on "symptoms" and findings from various examinations [11, 60]. However, OCRD is a complex group of disorders with a wide range of behavioral manifestations. It is also comorbid with other behavioral and psychiatric disorders. Yet connecting the dots between psychiatric assessment and behavioral data can be a challenge. Although discussed elsewhere in this chapter, ethological data can be very helpful at this point. This ethological approach adds social or group observational data to the assessment. For many patients with severe ID/ASD, behaviors are the means of communicating distress, expressing emotional states, dealing with disrupted attachments, and even territorial or dominance issues within stable social groups. Unfortunately, systematic ethological assessment is not a routine part of most evaluations. In the future, clinicians may combine ethological data with information about tempera-

ment variability, specific biologic markers, and available biomarkers for social behavior to enhance treatment planning, implementation, and monitoring [6, 50].

24.10 Cognitive-Behavioral, Exposure-Response Prevention, and Habit-Reversal Therapies

The introduction of cognitive behavioral, exposure-response prevention, and habit-reversal therapies revolutionized the psychotherapy of OCRD. In many clinical settings, CBT-ERP is a frontline treatment choice based on high rates of clinical improvement and a low risk of adverse effects [2, 4, 15, 105]. CBT addresses the top-down regulation of automatic thoughts that serve as setting events and limit potentially effective solutions. The basic technique focuses on overriding and replacing these automatic thoughts and the anxiety associated with new forms of thinking and behaving. The patient rehearses and then challenges the cognitive distortions (inaccurate risk assessment) and substitutes more adaptive cognitions and behaviors, as well as means of defusing and de-catastrophizing uncertainty rather than simply avoiding anxiety. CBT relies upon the integrity of frontal and cortical networks that regulate subcortical and limbic input in a top-down fashion. The role of language and thought process in behavioral regulation is to override associatively conditioned automatic linking of negative affective states to previously neutral experiences [3, 5, 64, 65].

ERP on the other hand focuses on the extinction of chains of these conditioned responses [2, 80, 90]. The basic components of ERP arise from a mixture of associative and instrumental learning. The mechanics of ERP involve breaking the linkages between unconditioned (aversive or distressing affective state) and generalizable conditioned stimuli (CS-neutral stimuli) that trigger a pattern of escape/avoidance behaviors (compulsions) that provide temporary relief. OCD represents a narrow group of triggers and repetitive behaviors. For example, performing a handwashing ritual (specific compulsive behavior) provides temporary relief from anxiety-provoking stimuli (dirt and fear of contamination) [8, 52].

The therapeutic technique of CBT-ERP focuses on a form of exposure based on a graduated schedule of affective intensity associated with systematic desensitization. This process includes hierarchically organizing threat cues or context-specific antecedents; then preventing the overlearned pattern of responding. OCD has a limited number of categories (doubt/checking; contamination/cleaning), whereas the "related disorders" have a broader sweep of triggers and symptoms. The ERP therapist tries to help regulate setting events (anxiety or threat level) and establish some degree of stimulus control. Once these techniques are in place, the therapist gradually increases the intensity of the exposure phase while desensitizing the patient to graduated levels of uncertainty and anxiety [52, 62, 66, 80, 90].

In some treatment-resistant patients, the combination of psychotropic medications with CBT-ERP provides additional therapeutic benefit. Combined therapy is useful for response maintenance, although many patients are able to tolerate a reduction and possible discontinuation of their psychotropic medications. Ongoing CBT-ERP can also be an effective relapse prevention technique. Thus far, adapted forms of CBT-ERP are effective for selected patients with mild ID/ASD but uncertainties about the risk of relapse, value of booster sessions, or the continued benefit over time remain [8, 16, 52].

Nevertheless, CBT-ERP is not a panacea. Data suggest that nearly 30% of patients do not achieve long-term positive outcomes. Remission is less frequent. CBT-ERP is constantly readapting to cope with treatment nonresponders. One approach addresses the differences between focuses on "in therapy" versus "out of therapy" responses working on the patient's compulsive use of exposure and response prevention but never focusing on the underlying anxieties associated with uncertainty and unpredictability [80, 90]. Interestingly, these response data are similar

outcomes for pharmacotherapy, but they may not represent the same 30% [2, 66]. Many of the same negative outcome predictors apply to both treatments, but neuroimaging suggest that there may be biological differences among CBT-ERP nonresponders. These include early onset OCD (children under age 10), chronic, severe OCD, high rates of psychiatric comorbidity (including tic disorders), significant psychosocial/family dysfunction, and in some impulsive/compulsive behaviors, habitual behaviors, and fronto-striatal disconnections that resemble some addiction-like behaviors [52, 63]. Other markers for nonresponse include comorbid psychiatric and neuropsychiatric conditions, poor insight, greater symptom severity, and specific clinical subtypes of OCRD (hoarding, sexual and religious preoccupation, and in some cases co-occurring tic and other movement disorders) [2, 8, 16, 52].

Habits, addiction, and tic-related repetitive subtypes are generally more resistant to CBT-ERP. Many of these patients do not respond to with anxiety but struggle instead with urges or intrusive images that crescendo until satisfied (or feel right). There are also differences in the typology of their repetitive behaviors, patterns of neuropsychological deficits, and basic neurobiology/neuropharmacology. This group is also more etiologically diverse. The focus of HRT is on recognition of the motivational factors associated with tic-related, compulsive behaviors combined with attempts to find alternative activities that may relieve the urges associated with tics and socially unacceptable behaviors [2, 10, 22, 58, 62].

At a deeper level, nonresponse to CBT-ERP parallels unsuccessful pharmacotherapies. Combined CBT-ERP and SSRIs may compensate for incomplete changes in neurophysiology and imbalanced excitatory/inhibitory brain functions. Functional neuroimaging (fMRI) suggests a failure to override a metabolically hyperactive caudate and medial prefrontal cortex. Correcting these faulty circuits may aid in behavioral and extinction protocols. Our understanding of the regulation of neuroplasticity is limited, but the role of several polymorphisms of the genes that regulate the production of brain-derived nerve growth factor, FMR1 protein, and metabotropic glutamate receptors and micro-RNA control of synaptic protein transcription are offering tantalizing clues for possible pharmacological interventions in this process. These may alter the course of learning, extinction, and deficits in neuroplasticity associated with OCRD and other neurodevelopmental disorders such as ID and ASD. Integrating these with CBT-ERP and HRT may be a useful strategy in patients with early-onset, chronic, and recurring OCRD [62–68].

CBT-ERP and HRT require modifications for patients with ASD or ID. There is little evidence that these techniques are applicable for patients with severe-to-profound ID. Children with ASD and ID are more vulnerable to the effects of trauma-related stresses (toxic childhood) and ongoing family dysfunction on psychopathology. Disentangling the effects of dysfunctional, family system dynamics on epigenetic changes is a subject of intense interest. Reversing these changes may require manipulating epigenetic tools (acetylation, blocking de-acetylation, blocking methylation, and enhancing the methylation to reverse some of the damage done during critical periods in development and restore neuroplasticity) [35, 58, 62, 64, 65, 68].

> The use of manualized behavioral psychotherapies is a very effective intervention for OCRD. For many patients, these interventions are better tolerated and more effective that other biological interventions. The do require some modifications for individuals with ASD and ID.

24.11 Psychopharmacology

This discussion will shy away from dealing with specific drugs, doses, variability in metabolism, or a restatement of evidenced evidence-based data on efficacy. Instead, the emphasis is on patterns of response-partial response, nonresponse, and or continued worsening in spite of our best efforts. The philosophy behind this approach centers on the premise that in an era of evidenced-based medicine, we may be able to learn more about

OCRD among patients with ASD/ID from nonresponders than from responders. This decision, in part, was an outgrowth of an observation that the psychopharmacological treatment of OCRD appears in a state of flux as several new trends are emerging. The use of pharmacogenomic profiles is one of these. There are multiple cytochrome P450 polymorphisms that influence drug metabolism. Some psychotropics are metabolized by multiple P450 pathways, glucouronidases, enolases, or are renally excreted largely unchanged. In addition to these enzymatic steps, the degree of protein binding, p-glycoprotein impact on the blood brain and cellular extrusion also influence pharmacokinetic variability [2, 32, 37].

Additional pharmacogenomic studies focus on pharmacodynamics, mostly mechanism of action and what drugs actually do within the cell. Ironically in recent years, there is growing interest not just on neurons, but drug effects on astrocytes, microglia (inflammatory peptides), local rRNA translation, and gene expression [32, 46]. Additional studies focus on differences on second and third messenger networks, differences between specific neurotransmitter or channel sites (ionotropic or metabotropic receptors), autoreceptor modulations, allosteric modulation, intracellular effects on calcium release and activity, phosphoinositol pathways, and gene activation that underlie drug response. These downstream effects constitute elements of the pharmacodynamics of various classes of psychotropics [32, 46].

Recent evidence suggests that OCRD shares at least some neurobiological, neuropsychological, neurophysiological, and genetic markers with a larger group of spectrum of clinical phenotypes. These include ADHD, ASD, ID, and tic disorders [2, 12, 32, 56, 67]. It can be difficult to disentangle these relationships based solely on descriptive/categorical phenomenology. For example, OCRD comprises at least five subgroups: anxiety-related OCD, nonanxiety-related compulsive behaviors, OC behaviors with tics, addiction-like behaviors, and OC behaviors related to other medical, neurological, metabolic, and neurodegenerative disorders [20, 42, 43, 67].

Although psychotropic drugs can influence the neurobiology of developmental disorders at many levels, mental health clinicians and psychopharmacologists tend to focus on diagnosing and treating syndromes (psychiatric disorders). The measurement of treatment response is largely based on symptomatic improvement. In this chapter, we face a different set of needs. As clinicians, we focus on a broad spectrum of repetitive behaviors and OCRD and monitor changes in these states. However, even our most effective treatments do not eliminate ASD or ID. Most controlled studies compare treatment with nontreatment (placebo). Lest we forget, ASD and ID consist of multiple subtypes. Perhaps it would be more useful to compare changes in specific temperamental or neurophysiological states in patients with ASD or ID with and without OCRD (control population). But even this level of analysis leaves us to cope with a heterogeneous sample. ASD and ID are complex genetic disorders with both Mendelian and polygenic patterns of inheritance and multiple etiologies. The most reasonable approach may be to accept that at this point we cannot eliminate the effects of ASD or ID on OCRD, but we can narrow our focus and address shared biomarkers and combine phenomenological and neurobiological endophenotypes [32, 50, 70].

One example of such an approach involves addressing the effects of psychotropic drugs on observable phenomena like temperament differences in neurophysiological arousal, behavioral inhibition, harm avoidance, neuroticism and new neuropharmacological studies of fear/anxiety conditioning, sensitization, generalization, and resistance to extinction learning. Each of these traits may help tease out a specific treatment approach to the different subgroups of OCRD. This is a slightly more refined treatment model than just focusing on OCRD. This approach is a step toward a behavioral pharmacology of intermediate endophenotypes rather than focusing treatment exclusively on DSM-5/DM-ID-2 diagnostic categories [16, 70, 75].

More powerful functional neuroimaging and electrophysiological tools are slowly exposing drug-induced changes in specific

neuronal ensembles, sensory processing networks, and downstream effects associated with default mode, selective attention, and the interconnections between innumerable brain networks (coherence). These new developments shed additional light on aberrant cognitions and behaviors associated with ASD/ID and are likely to influence the course of research on future treatments for OCRD.

At a deeper level, such analyses define differences in temperamental vulnerabilities such as threshold intensity and extent of fear/anxiety conditioning, excessive generalization, and deficits in extinction. These intermediate endophenotypes arise from underlying developmental neurobiological, neurophysiological, differences in functional neuroimaging, and behavioral neuropharmacological strata. The most complicated step in this process may be addressing the role of biomarkers shared by subgroups of patients with OCRD and subtypes of ASD/ID. This would be a step in the direction of more personalized care, but unfortunately, we must leave most of these complex issues unexplored and definitely unresolved.

Realistically this level of analysis is beyond the scope of practice for most practitioners. We rely on evidenced-based studies or best practices models to provide guidance and reassurance. Unfortunately, many of our current evidenced-based treatments do not require this level of data-driven diagnostic precision. Many of our current psychotropics were developed to affect a relatively small collection of neurotransmitter systems. The mechanism of action generally includes reuptake blockade to increase neurotransmitter bioavailability. More recently, an expanded understanding of their mechanisms of action includes up/down regulation of autoreceptors, and postsynaptic receptors, allosteric modulation by means of neuropeptides, nitric oxide (NO)/carbon monoxide (CO), and cytokine activity. On the horizon, there are immune modulators, anti-inflammatory biologicals, and treatments designed to modulate or reverse epigenetic changes; metabotropic glutamate receptors that help regulate local protein transcription [53, 60, 65].

Most clinicians successfully grapple with the pharmacological treatment of OCRD at the syndromal level with or without comorbidities. For the most part, these pharmacological strategies are effective for a large number of patients. Many patients are resistant to available evidence-based and best practices treatment recommendations. The underresponding patients either only partially respond, or worsen in spite of treatment. This effectiveness gap in neurotypical populations suggests the need to look nonresponders at different perspectives. This strategy is especially true for patients with OCRD+ ASD+ ID. For example, many clinic referrals are not limited to primary OCRD [69, 71, 76, 77]. Most patients present with coexisting static encephalopathy, epilepsy, or medical conditions that influence their mental health. The process of teasing out OCRD is not always straightforward. For many, side effects from medical treatments can exacerbate baseline repetitive or OC behaviors. Resolving these usually requires close collaboration with medical colleagues in an attempt to avoid misattributing these changes in symptoms to ineffective treatment of OCRD. In other situations, the OCRD is misdiagnosed and the decision to treat with an SSRI and/or a neuroleptic may increase the likelihood of treatment nonresponse, or worsening of the original, presenting symptoms [2, 6, 11, 21, 29].

Is improvement with SSRIs related to a specific anti-obsessional effect or a reduction in comorbid anxiety or depression or adverse effects from antipsychotics, mood stabilizers, or SSRI dosing patterns? We could answer "yes" to each. Comparison studies of pharmacotherapies suggest interesting differences between antidepressant and anti-OCRD effects. These differences include a longer latency of treatment response in OCD; higher dosing needs for OCD; different responses to tryptophan depletion; and a more selective response to SSRIs than to noradrenergic specific tricyclic antidepressants. The tricyclic clomipramine is effective for OCD (stronger serotonin reuptake inhibition) but its metabolite is not (inhibits norepinephrine reuptake). The data about the relationship between OCRD and depression, bipolar disorder, and schizophrenia are equally nuanced and warrant close attention [6, 12, 14, 41].

Many prescribers do not factor in the prolonged latency of response or dosing needs for patients with OCRD who also have ASD/ID. This problem has two faces: the dosing requirements for OCRD and the heightened sensitivities to side effects among patients with ASD/ID. The latter problem relates to the complex neuropharmacology and apparent pharmacodynamic variability associated with ASD and ID. In addition, SSRIs do more than block serotonin reuptake. They display a prolonged latency of response that is due in part to a delayed effect on brain-derived neurotrophic factor synthesis, release and activation, related effects on synaptic plasticity, receptor up/down regulation, reduction in cytokines and inflammatory peptides, and increased neuro-steroid activity. Many of these complex biological responses are not specific to OCRD, but may represent shared traits with other mood-anxiety disorders, as well as temperamental traits like irritability, pain responses and some forms of affective aggression. These multiple pathways are sensitive to SSRIs/selective norepinephrine reuptake inhibitors (SNRIs) and support the generalilzed nature of SSRIs response [34, 51, 53, 69, 70, 106].

A third factor involves a complex response to tryptophan depletion (lowering serotonin levels). The effects are most dramatic in patients who are SSRIs responders. Patient response to serotonin-depletion differs between patients with major depression, ASD, and anxiety-related OCRD. Those with OCD require months to relapse if at all. Patients with depression experience a rapid downturn in mood and hedonic drive. Those with ASD also experience a rapid increase in irritability and other challenging behaviors. This pattern of serotonin effects suggests that ASD and mood disorders are sensitive to this technique while patients with anxiety-related OCRD are not. It may also indicate a subtle difference between OCD (anxiety disorder) and the other subtypes of OCRD that may be relevant to treatment response. The symptoms that seem most resistant to SSRIs include a need for symmetry, arranging, organizing, and counting with obsessions or overt anxiety. Many of the compulsive behavior resemble complex tics and some symptoms of ASD [37, 69, 71, 75, 76].

Many patients with OCRD remain are at increased risk for developing other psychiatric disorders [20–22, 32]. Many also present with early-onset OCRD and co-occurring ADHD-like symptoms. For these patients, SSRIs can be less than optimal. For children with OCRD, minor tics, and ADHD, many clinicians avoid the use of stimulants due to uncertainties about the safety of stimulants, the risk of "causing" or exacerbating tics, or worsening repetitive behaviors. These worries may be unfounded for many patients, but multiple alternatives are available [2, 21]. These include alpha agonists, N-acetyl cysteine, omega-3 fatty acids, second-generation antipsychotic drugs plus SSRIs, and stimulants for younger children. Older children and adolescents are treated with SSRIs/SNRIs and, if necessary, adjunctive buspirone, benzodiazepines (clonazepam in particular), and, in recent years, partial glutamate agonists, antagonists, anti-inflammatory agents, probiotics, and neuromodulation [2, 6, 11, 21].

The response to alternative SSRIs, SNRIs, and clomipramine is more variable in the population with coexisting tic disorders. This group may require augmentation in order to better stabilize their obsessive-compulsive features. The need for augmentation with first- or second-generation antipsychotic drugs, clonazepam, and in some cases N-acetyl cysteine suggests that the relationship between serotonin, dopamine, excitatory amino acids (glutamate), and other neurotransmitter networks is far more complex than originally proposed. Older studies suggest differences in cerebrospinal fluid (CSF) levels of vasopressin and males with tic disorders and oxytocin in females with OCD. The broad range of targets for these two hormones suggests a more complex problem [12, 32]. In addition, some patients worsen on naltrexone, whereas those with related symptoms (trichotillomania) may improve. Cannabinoids may improve tics but have little or no benefit for OCRD. In the

last 10 years, the introduction of glutamate antagonists, opiate antagonists, both benzodiazepine type A and B receptor agonists, N-acetylcysteine, and inositol have demonstrated limited efficacy [32, 51, 53].

Of course, the heterogeneity of ASD and ID challenges the validity of this pharmacological model for OCRD. Many patients are receiving complex multidrug regimens that target OCRD, but on closer inspection, they do not meet the diagnostic criteria for OCRD. In addition, many of these patients have their repetitive-restrictive behaviors (rituals, stereotypies and self-injury) misattributed to OCRD. This challenge is particularly difficult in patients with severe ASD/ID. There is a tendency to label repetitive behaviors as treatment-resistant OCRD rather than focusing on the presence of co-occurring tic, neurogenetic, and other neuropsychiatric disorders [37, 69, 71].

As noted earlier, many patients will experience symptoms of OCRD for years before seeking treatment. The emergence of depression, however, prompts the decision to seek treatment [2, 6]. In many, the successful treatment of depressive symptoms leads to an improvement in OC symptoms but usually not remission. As we shall see later, studies of neuromodulatory treatments suggest that their antidepressant effects may actually mediate their positive effects on treatment-resistant OCRD.

Tip

Neuropharmacology involves genetics (pharmacogenetics), complex mechanisms of action (pharmacodynamics), and modifications of multiple neuronal systems that are more complex than simple drug effects on specific neurotransmitters. Treatment response involves many biopsychosocial factors as well. The entanglement between neuropharmacological and biopsychosocial factors are major contributors to clinically observed low remission rates and high rates of treatment unresponsiveness.

24.12 Rationale for Combined Therapies

There is ample clinical evidence that SSRIs are helpful for 60–70% of individuals with OCRD. The remaining 30–40% points to several gaps in our treatment models [37, 69, 71]. Treatment response depends upon an appropriate dose and sufficient patience to endure the long latency of treatment response. This type of nonresponse is frequently due to disregarding these caveats. However, nonresponse is also traceable to pharmacokinetic differences in rate of absorption and the functional dynamics of presynaptic drug transport receptors (5-HT transporter protein).

Pharmacogenomic studies suggest that variability in drug response is also due to the gene effects on the regulation of postsynaptic receptor activity, intracellular activity such as effects on second messenger systems, and gene regulation of neuroplastic changes and neuronal interconnectivity [6, 28, 73–75, 77]. In addition, we may focus our attention solely on serotonin and overlook the fact that it may not be the only neurotransmitter involved. There are multiple systems interfacing the dysregulated serotonin system we observe clinically such as dopaminergic, glutamatergic, neuropeptidergic, neuroendocrine, neuroimmune (anti-neuronal antibodies) and inflammatory (cytokine), and diffusible neuromodulators. It turns out that the bigger challenges may be determining which interface is involved, matching a particular interface with our OCRD subtypes, and then describing how ASD/ID influences both [3, 6, 16, 51, 74, 77].

In neurotypical patients, treatment resistance in OCRD relates to the presence of comorbid tic disorders [21, 77]; co-occurring impulse control disorders and addiction behaviors [1, 4]; trauma-related, mood and anxiety disorders; and other neuropsychiatric disorders. These insights may not clearly apply to OCRD co-occurring with ASD, ID, and ASD+ID. All too frequently, we lump these heterogeneous phenotypes and not dissect ASD or ID into its many subtypes. For example, clinicians may have difficulty differ-

entiating OC behaviors from complex rituals, stereotypies, self- injury, and other repetitive behaviors associated with ASD and ID [53, 75, 78]. The limited effectiveness of pharmacotherapy is frequently the result of mismatching psychotropic drug to problem behavior. A second problem area is due to a lower threshold for troublesome side effects. For example, akathisia is a sense of inner restlessness and dysphoria that accompany a dysphoric affective state that can exacerbate insomnia, stereotypies, aggression, self-injury, and other psychiatric symptoms. It is generally associated with antipsychotic-related extrapyramidal side effects but can also occur in response to higher doses of SSRIs. Unless clearly recognized and appropriately treated, akathisia becomes a contributor to high dose polypharmacy [69, 71].

Other medical conditions are frequently associated with OCRD and other disorders of repetitive behavior. This list is a familiar one in this chapter and includes a variety of medical/neurological disorders and trauma and stress-related disorders. OC-like symptoms in these cases may not respond to standard anti-OCRD treatments. Polypharmacy may also contribute to delirium and further complicate treatment [32, 37, 55, 59, 67, 79].

In keeping with the philosophy of this chapter, adopting a behavioral pharmacological approach can help focus the assessment on the search for a neuropharmacological basis for the intermediate endophenotypes of temperament. This approach is also a step towards linking sympathetic/parasympathetic imbalance, and disruption of excitatory/inhibitory networks with the neuropharmacological foundations of learning theory. This linkage appears to underlie not only conditioned response but also their generalization of conditioned stimuli-response learning, neuroplastic changes associated with sensitization of specific experiences, and the stability of memory storage and retrieval of this information. Each of these phenomena contribute to the extinction of maladaptive conditioned learning and the capacity for new learning of alternative patterns of behavior [6, 46, 64, 66, 77]. These techniques would match temperamental traits routinely with selective or combined pharmacotherapy and behavioral or CBT-ERP [5, 17, 28, 80, 81]. One intriguing strategy involves pretherapy treatment with glutamate partial agonist (d-cycloserine) to enhance memory during extinction training [2, 6, 18, 25, 26, 32]. The combination of novel pharmacotherapies for addictions with habit-reversal therapy depends on a different set of neuropsychopharmacological and behavioral strategies. Many of these interventions are useful in the treatment of chronic substance use disorders. But neither augmentation strategies are panaceas. They do however open the doorway to novel treatment ideas and future research [52, 63].

In summary, the definition of treatment resistance remains a fluid one. In recent years, combination therapies and nonpharmacological interventions are gaining ground.

This is especially true in light of the 30% of patients who may fail combined pharmacotherapy and cognitive-behavioral, exposure-response/habit-reversal therapies. There is growing evidence from functional neuroimaging to suggest not only brain changes arising from psychotherapies that overlap drug effects. These studies also demonstrate that CBT-ERP/HRT nonresponders fail to show similar reversals in the Caudate/ventral orbitofrontal cortex dysfunctional activity [21, 22, 32, 52, 55, 62]. For some nonresponders, there is growing use of neuromodulation (transcranial magnetic and electrical stimulation), deep brain stimulations, and neurosurgical interventions for severe, treatment refractory OCRD [25, 54].

There are many factors related to incomplete treatment response to standard psychotherapies and pharmacotherapies. Keep in mind that response takes proper dosing, sufficient time, and methods of monitoring clinical response. In some situations, treatment augmentation and combined psychotherapies can be helpful.

24.13 Neuromodulation and Neurophysiology

Neurobiological research into OCRD consistently points to dysfunctional connections between supplemental motor, medial prefron-

tal; orbitofrontal, caudate dorsal and ventral striatum, putamen, globus pallidus, subthalamic nucleus, thalamus, greater amygdala and hippocampus, and temporal-parietal association cortices [11, 14, 25, 54, 82, 83].

The behavioral expression of functional neuroanatomical connections is influenced by co-occurring ASD and other neurodevelopmental disorders, Tourette syndrome, schizophrenia spectrum, mood, ID, and most neuropsychiatric disorders. Our ability to measure brain activity with neurophysiological probes, magneto-encephalography (MEG), and diffusion tensor imaging (white matter tracts) allows researchers and clinicians to better define the relationship with higher cortical functions. Functional neuroimaging adds to our capacity to study both resting and activation states. These studies expand our knowledge base in areas such as a lack of coherence between functional circuits, dysregulation of default mode cognition, attention and sensory processing and integration, and cognitive-behavioral processing and motor expression [2, 6, 15, 36, 58, 84, 93].

Transcranial direct current stimulation and transcranial magnetic stimulation are noninvasive technologies that use electromagnetic current and fields to not only mapping neuronal activity within these circuits but as effective treatment strategy for mood disorders and anxiety disorders [25, 26, 54]. Transcranial direct current stimulation (tDCS) delivers a low-intensity electrical current that influences both immediate and distant neuronal activity. It affects the interconnections within and between neuronal populations. tDCS may influence the synchronous firing rates of neurons within a circuit. There are short- and long-range effects that may induce neuroplastic changes over time [54, 89].

The treatment effects on the neurophysiology of brain functioning with seizure induction (ECT) and without (tDCS) suggest that tDCS may become a reliable treatment for disorders like OCRD, and perhaps augmentation of other interventions for patients with ASD and other neurodevelopmental disorders [24, 36, 46, 58, 68, 81, 85, 107].

TMS uses magnetic fields to alter regional cortical activity. Low frequency (1–2 Hz) stimulation has an inhibitory effect on overexcited neuronal activity. These changes raise the threshold for activation of various neuronal pathways and reduce their rates of firing. Higher frequencies (10 Hz) may stimulate similar pathways, especially those in the premotor cortex. The clinical effects of both may depend on lateralization, selection of neuroanatomical sites, nature of the circuits under study, and the relationship between over and reduced activity on the circuits involved [84, 85, 86].

For rTMS, lower frequency probes provide additional information about thalamocortical and corticostriatal and limbic regulation. Many of these inputs are excitatory but they synapse onto inhibitory subcortical substrates and fine-tune sensory gating, attention, emotional perception, and responses, prepotent responses. OCRD, tic disorders, traumatic experiences, and primary psychiatric disorders result from various disconnections within these circuits. The high-low frequency probes are useful for regional mapping studies as well as treatment of mood/anxiety and tic disorders, and reduce auditory hallucinations in some patients with treatment refractory schizophrenia [54, 84, 85, 86, 108]. tDCS may influence the synchronous firing rates of neurons within a circuit. There are short- and long-range effects that may induce neuroplastic changes over time [54, 89].

Deep brain stimulation (DBS) involves neurosurgical implantation of electrodes that deliver programmed electrical activity directly within fronto-striatal networks. Although DBS is frequently more effective for treatment refractory OCRD, it does carry surgical risks [82, 87]. Currently clinical research suggests that electrode place in the brain circuits connecting the ventral striatum, palladium, and corticostriate pathways. The exact mechanism of action for DBS remains unsettled, but in all likelihood, it influences the imbalance between excitatory and inhibitory activity in several corticostriate pathways. For many patients, ablation neurosurgical intervention can be most effective when directed toward disconnecting dysfunctional circuits between the ventral basal ganglia, limbic, and other cortical regions [54, 85, 86, 87, 88, 89].

However, there are limitations to these strategies. The risk treatment refractory OCRD emerges in context of comorbid mood/anxiety disorders, early age of onset, chronic illness, comorbid tics, ASD/ID and other neurodevelopmental disorders. Some of these can decrease the efficacy of rTMS [16, 26, 37]. For example, ECT can be an extremely effective treatment for severe depression but has little or no efficacy for patients with OCRD. The response rates for ECT, however, vary relative to the number of previously unsuccessful medication trials. This also may apply to rTMS and suggests a change in the neurophysiology of depression with repeated episodes, early onset, and long duration of the disorder.

Nevertheless, rTMS does have a positive effect on depression—especially newer strategies that combine high and low frequencies to reflect bifrontal activation. Improvement in OCRD may be indirect, reminding clinicians that many demoralized patients with refractory OCRD go on to develop depressive disorders. rTMS may have a direct effect on mood and an indirect one on the level of functional impairment in which many patients feel less overwhelmed by their OCD. The problem of comorbidity means that we still must disentangle the positive effects of rTMS and ECT on comorbid mood and anxiety disorders from their effects on OCRD [35, 36, 46, 54, 82, 84, 92].

To date it is unclear if tDCS or DBS differs in this regard. This may require additional studies about age of onset, duration of symptoms, and past treatment failures; alternative placement arrays, combination of high and low frequencies for direct current and magnetic field probes, or more research into better electromagnetic access to critical nodes within and between these complex circuitries [19, 37, 53, 65].

Unfortunately, patients with treatment-resistant OCRD, ASD, and ID are not participants in many of these studies. In ASD, the aberrant functional neuroanatomical and imbalanced neurophysiological-neuropharmacological networks relate to a risk increase of stereotypies, rituals, and other repetitive and restrictive patterns of cognition, emotions, and behaviors [1, 13, 37, 59]. The insistence on sameness and reactions to change may underlie some of these rituals. The boundaries between these repetitive behaviors and OCRD can be confusing [55–57]. Many ritualized behaviors in ASD seem to become habitual over and more difficult to pinpoint specific triggers, typologies, or reinforcing properties. In this sense, patients with ASD may have more in common with tic disorders, the related disorders spectrum of OCRD, hoarding, and impulsive-stereotypic movement disorders than from current knowledge [13, 17, 23, 62]. Clinicians are less apt to find classic OCD (obsessions, contamination cleaning or avoiding, and doubting-checking symptoms) in patients with ID/ASD [6, 37, 59]. Our understanding of OCRD in patients with ID/ASD may require a dissection of habit-related (reactions to social ambiguity, unexpected noises, broken routines, or environmental changes) from those anxieties related to anxiety/misery/worry dimensions, dysphoric affective responses, high harm avoidance [6, 15, 17, 80, 62].

In both ASD and ID, the use of these exploratory neuromodulation may be helpful in dissecting aberrant interconnections between localized and distal neurocircuitries that underlie ASD and ASD + ID. These studies can provide additional data on the patterns of atypical neuronal development and maturation that most closely link to ASD and OCRD. In addition, they may provide a better understanding of gender dimorphism in ASD, tic disorders and mood/anxiety disorders. As a rule, males are more vulnerable to primary ASD (polygenic inheritance, higher familial occurrence), ADHD, and many other neurodevelopmental disorders and tic disorders. The male:female ratio is significantly larger than those found in late onset OCRD are. A similar shift in gender ratios occur in secondary ASD (associated with neurological and metabolic disorders) [2, 21, 23, 37]. These studies will hopefully provide a better understanding of the role played by serotonin, glutamate, dopamine, and oxytocin/vasopressin and may offer new insights into different patterns of drug response [32, 37, 92].

As noted earlier, rTMS and tDCS technologies are very useful for mapping out the neu-

rophysiology and functional neuroanatomy of OCRD. More detailed functional probing and mapping will continue to provide insights into the neurocircuitry of fronto-striatal ensembles and their role in top-down regulation and overly zealous subcortical/limbic activation [3, 7, 24, 36]. The second positive impact involves developing deeper insights into the interconnections between excitatory/inhibitory circuitries at a local level of neuronal activity and their regional influences on distal activation and coherence. These distal connections may heighten our understanding of top-down regulation of sympathetic/parasympathetic, neuroendocrine, and neuroinflammatory functions [21, 24, 26, 27]. Lastly, these technologies may help refine our understanding of the neurodevelopmental underpinnings of ASD and ID on the emergence of coherence, default mode cognitions and neurocognitive and neurobehavioral substrates.

Mapping the neurophysiological differences with OCRD can be combined with studies of aberrant patterns of white matter tracts (diffusion tensor imaging; DTI) to gain a deeper understanding of excitatory/inhibitory balance, top-down regulation, and long-term interconnections in ASD that may differ from OCRD [23, 25, 26, 37, 92]. The impact of ID on these neuropsychological and cognitive traits of ASD and OCRD may improve the quality of current treatment approaches. In many cases, such combined studies may help reduce considerable phenomenological and pathophysiological heterogeneity that make studies so difficult to extrapolate and interpret. These new methodologies may help to better define phenotypic variability within ID (including specific behavioral phenotypes) that may help differentiate stereotypies and some treatment-resistant forms of self-injury [10].

More recently, alternative biological treatments using direct brain stimulation are becoming more acceptable for individuals with high levels or treatment refractory OCRD. In general, there is limited application of these technologies for individuals with co-occurring ID and ASD. Less-invasive strategies such as various iterations of rTMS are gaining interest.

24.14 Summary and Conclusions: How Pathophysiology Guides Treatment

The DSM-5 and the DM-ID-2 bring together obsessive-compulsive disorder and other repetitive behavioral syndromes into obsessive-compulsive and related disorders (OCRD). In this chapter, the author expanded beyond diagnostic criteria, and focused instead on deconstructing OCRD. The goal of this deconstruction project was to sort out as much diversity as possible and create a framework for understanding the complex interrelationships between OCRD and ASD and ID. The mechanics of this process involved dissecting specific temperamental features and behavior clusters down to their basic pathophysiology. This included their basic neuropharmacology, neurophysiology, and ultimately gene regulation and expression.

Throughout, the explicit goal was to augment the categorical, descriptive definition of OCRD with data from the neurosciences on biological and neuropharmacological endophenotypes. In doing so, the search was on for interconnections between OCRD, mood and anxiety disorders, impulse control and addiction-related disorders, and ASD/ID. A second goal focused on the exploration of behavioral (learning models), neuroethological, and behavioral psychopharmacological subtypes of OCRD. Many behavioral programs focus on challenging behaviors rather than specific syndromes. Functional behavioral data guide an analysis of variables associated with the precipitation, maintenance, and treatment of repetitive behaviors and OCRD as a subset of ID/ASD. From this perspective, clinicians are tasked with understanding mutually interacting systems rather than three distinct, co-occurring syndromes. The level of syndromal and biological entanglement may push researchers and clinicians to consider strategies that focus on the behavioral pharmacology of OCRD subtypes within the framework of ASD and ID.

The last segment of this chapter concentrated on three treatment models: exposure-response prevention/CBT and habit-reversal

therapy, psychopharmacology, and neuromodulatory/neurosurgical interventions. A review of psychopharmacological interventions suggests that our best treatment approaches fall short of clinical remission for as many as 70% of those treated for OCRD [2, 7, 51, 66]. A similar observation holds for CBT/ERP, habit-reversal therapy (HRT), and combinations of CBT/ERP and psychotropic medications. Neuromodulatory and neurosurgical treatment currently apply to the most severe, treatment-resistant forms of OCRD. This is unfortunate since direct current neurostimulation, rTMS, and even neurosurgical interventions may be more effective earlier in the treatment selection process. The problem with each of these new technologies involves how to apply them to patients with ASD and ID.

In the final analysis, clinicians must deal with the sense of frustration with our current treatment approaches to OCRD. Although we are making significant gains, persistently high rates of treatment resistance still plagues clinicians. Part of the problem lies in the often overwhelming heterogeneity of OCRD patients with ASD and ID. We are dealing with three entangled neurodevelopmental disorders that present as final common pathways to three species of complex genetic disorders. They converge along many nodal points and breakthrough as subtypes along a OCRD-ASD/ID continuum. Further developments in the neurosciences may eventually help us improve outcomes by designing more personalized treatment approaches.

Of course, the ideas expressed in this chapter will need additional revision as new data emerges. The application of pathophysiological model will be contingent on these data, but there are new models such as Research Domain Criteria (RDoC) [47] and the Intermediate Phenotyping [49] that will play a major role in this transformation. New technologies will help to look beyond our current fixation on specific neurotransmitters and mechanism of drug action. However, there are dangers in any paradigm shift, especially when we reduce complex patterns of human behavior and emotional life down to a molecular level.

However, these are not simple challenges. These many models addressed in this chapter make this abundantly clear. It is easy to become discouraged or get bogged down in the details and lose ourselves in the forest while concentrating on the microscopic structure of molecular biology. Any hope of success hinges on our capacity to translate the neuroscience explosion into manageable chunks, which are applicable to clinical problems. Hopefully, this chapter is at least one step in that direction, and a bequeathal for our students, residents, and trainees who will struggle after us.

To date, we have no cures or magical silver bullets, but as new discoveries trickle down from the neurosciences, new diagnostic and treatment models will follow. The brain and disorders of brain functioning share more in common with a complex ecosystem than discrete lesions or single neurotransmitters.

Key Points

- Obsessive-compulsive disorder is a complex polygenic and heterogeneous neuropsychiatric disorder.
- The DSM-5 and DM-ID-2 recategorized OCD from an anxiety disorder to obsessive-compulsive and related disorders. This step expanded OCD to include other patterns of repetitive behaviors.
- OCD frequently co-occurs with anxiety and mood disorders. Psychotic disorders, ADHD and other impulse control disorders, stereotypic and tic disorders, and neurodevelopmental disorders such as autism spectrum and intellectual developmental disorder.
- The neurobiology of OCRD involves elements of the fronto-striato-thalamic networks that are involved in sensory gating, and top-down regulation of multiple reciprocating subcortical connections.

- The boundaries between the core features of OCD overlap ASD, tic disorders, and other movement disorders, as well as other neuropsychiatric disorders, OCD, and other.
- Manualized psychotherapies (cognitive-behavioral), exposure-response prevention, and habit-reversal therapies are considered frontline treatment in many settings. The presence of ASD and ID require modifications.
- Primary psychopharmacological treatments focus on modifying serotonin networks. The presence of other comorbid conditions impacts the nature and clinical course of OCD as well as the effectiveness of these treatments.
- Both psychotherapy and pharmacotherapy lead to significant improvement in the majority of patients. The co-occurrence of ASD and ID can be effective, but may require modifications of these techniques.
- Remission is rather uncommon even with combination therapies. A substantial minority does not respond and requires more complicated treatment protocols.
- Treatment-resistant, severe OCD may require transcranial direct current or magnetic stimulation, as well as neurosurgical interventions such as lesioning connecting tracts or implantation for direct brain stimulation.

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Feeding, Eating and Weight Disorders

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Contents

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Learning Objectives

- To understand why feeding and eating disorders (FEEDs) are among the least studied psychiatric disorders in persons with ID/ASD.
- To define what FEEDs are and outline their main psychopathological, physical and nutritional features in adults with ID and ASD.
- To clarify the prevalence, types and aetiological associations of the different FEEDs in adults with ID and ASD.
- To discuss screening, diagnosis, clinical assessment and management of FEEDs in adults with ID and ASD.
- To consider the personal psychological meaning of FEEDs for individual adults with ID and ASD.
- To suggest future clinical, research and service development directions.
- To understand implications of FEEDs for individuals with ID, their families/carers and care service providers.

25.1 Introduction

Eating disorders (EDs) in the 'mainstream' (non-ID) population are serious frequently persistent and complex mental disorders that usually involve young adolescent and adult females. The burden of such 'mainstream' EDs (anorexia nervosa, bulimia nervosa and binge eating disorder) for healthcare systems is high. The prevalence of such mainstream EDs (especially binge eating disorder) seems to have increased in the last decades [1, 2]. Little is known about these 'mainstream EDs' when they occur among adults with intellectual disability (ID). This has been partly explained by the obviously lower levels of communication and conceptualizing skills demonstrated by most adults with ID, making it challenging to adequately clinically assess and diagnose such complex EDs in them [3].

Moreover, little attention was paid to these diagnostic issues by clinicians and researchers, with few contributions evaluating the complex relationships between feeding and eating disorders (FEEDs) and ID [4]. Therefore, more studies were and still are needed to further clarify the prevalence, clinical and service impacts and personal meaning of FEEDs in adults with ID [5] and autism spectrum disorder (ASD) [6].

Little is known about clinical and service impact, and personal meaning of FEEDs in adults with ID/ASD.

25.2 Recent Classifications of FEEDs

ICD-10 [7] and ICD-10 MR [8] will still be in widespread use for several years awaiting full role out and adoption of the new online ICD-11 beta version available for field trials since 2015 [9]. In the UK, DC-LD [10] is still widely used despite the availability of DSM-5 [11] and DM-ID [12]. Evaluation of DM-ID [13] is awaited before adoption can be considered.

Table 25.1 compares the DC-LD, DSM-5, DM-ID and current field trial ICD-11 diagnostic classifications for FEEDs. The FEEDs have included: classical 'mainstream' eating disorders best known in adolescent/young adult females without ID, that is, anorexia nervosa, bulimia nervosa and binge eating disorder; feeding disorders most commonly seen in developmentally less mature children and adults with ID and ASD, that is, pica, rumination/regurgitation disorder, food faddiness/refusal aka avoidant and restrictive food intake disorder (ARFID); previous psychogenic stress related loss of appetite, overeating and vomiting EDs; some less common EDs such as night bingeing syndrome and newer EDs disorders including orthorexia [15–17] and muscle dystopic syndrome ('bigorexia') [18, 19].

Mainstream EDs are severe and chronic syndromes that in most cases involve young women affecting their personality development and their physical health [20]. EDs are sustained by socio-cultural, psychological and biological factors [21].

Table 25.1 Comparison of ICD-10, DC-LD, ICD-11, DSM-5 and DM-ID-2 diagnostic classifications for FEEDs

Eating disorder	ICD-10	DC-LD	ICD-11	DSM-5	DM-ID-2
Pica	Not present as a specific category	Pica	Pica	Pica	Pica
	Coded as a category of 'Other behavioural and emotional disorders with onset usually occurring in childhood and adolescence' or included within 'Other eating disorders' for adults	–	–	Different codes for children and adults, within the same category	Excluding people with severe or profound ID just being unable to differentiate between prepared and unprepared, raw or discarded food
Avoidant/restrictive food intake disorder	Not present as a specific category	Food faddiness/food refusal disorder	Avoidant-restrictive food intake disorder	Avoidant/restrictive food intake disorder	Avoidant/restrictive food intake disorder
	Partially included in 'Feeding disorder of infancy and childhood'	–	–	Specifier for 'in remission'	Including atypical manifestations such as food refusal, turning away of the head or keeping lips sealed while being fed, vomiting during or after the meal. Excluding inadequate diet or insufficient care
Rumination disorder	Not present as a specific category	Food rumination/ regurgitation disorder	Rumination-regurgitation disorder	Rumination disorder	Rumination disorder

	Rumination disorder of infancy, within 'Feeding disorder of infancy and childhood', part of the chapter 'Other behavioural and emotional disorders with onset usually occurring in childhood and adolescence'	'Excessive chewing/ spitting out food disorder' as a separate category	Does not include 'Adult rumination syndrome' (under chapter 13 - Diseases of the digestive system) and 'Nausea or vomiting' (under chapter 21 - Symptoms, signs or clinical findings, not elsewhere classified)	Specifier for 'in remission'	Excluding inadequate diet or insufficient care
Anorexia nervosa	Anorexia nervosa	Anorexia nervosa	Anorexia nervosa	Anorexia nervosa	Anorexia nervosa
	'Atypical anorexia nervosa' as a separate category	–	Specifiers for 'with significantly low body weight'; 'with dangerously low body weight'; 'in recovery with normal body weight'; 'other specified'; and 'unspecified'	Subtypes: binge-eating/purging; restricting. Specifiers for 'in partial' or 'in complete remission'. Specifiers for severity	Including additional observations from caregivers and family members for persons with moderate to profound ID
Psychogenic loss of appetite disorder	Not present as a specific category	Psychogenic loss of appetite disorder	Not present	Not present	Not present
	Included in 'Other Specified Feeding or Eating Disorder'	Specifiers for 'currently in episode' or 'currently in remission'	–	–	–
Psychogenic vomiting disorder	Vomiting associated with other psychological disturbances	Psychogenic vomiting disorder	Not present	Not present	Not present
	–	Specifiers for 'currently in episode' or 'currently in remission'	–	–	–

(continued)

Table 25.1 (continued)

Eating disorder	ICD-10	DC-LD	ICD-11	DSM-5	DM-ID-2
Bulimia nervosa	Bulimia nervosa	Bulimia nervosa	Bulimia nervosa	Bulimia nervosa	Bulimia nervosa
	Atypical bulimia nervosa as a separate category	–	–	Specifiers for 'in partial' or 'in complete remission'. Specifiers for severity	–
Binge-eating disorder	Overeating associated with other psychological disturbances	'Binge-eating disorder' and 'psychogenic overeating disorder'	Binge-eating disorder	Binge-eating disorder	Binge-eating disorder
	Includes: overeating due to stressful events; Psychogenic overeating	'Psychogenic over-eating disorder' has specifiers for 'currently in episode' or 'currently in remission'	–	Specifiers for 'in partial' or 'in complete remission'. Specifiers for severity	–
Other Specified Feeding or Eating Disorder	Other eating disorders	Not present	Other specified Feeding or eating disorders	Other specified feeding or eating disorder	Other specified feeding or eating disorders
	Includes: Pica in adults; psychogenic loss of appetite	–	–	–	–
Unspecified feeding or eating disorder	Eating disorder, unspecified	Not present	Feeding or eating disorders, unspecified	Unspecified feeding or eating disorder	Unspecified feeding or eating disorder
	–	–	–	–	–

25.2.1 DSM-5 Criteria for Feeding and Eating Disorders

In the DSM-5, EDs are in a new chapter called *Feeding and Eating Disorders*. This category encompasses different psychopathological conditions such as anorexia nervosa (AN), bulimia nervosa (BN), binge eating disorder (BED), and 3 FEEDs disorders previously described in the *Feeding and Eating Disorders of Infancy or Early Childhood* chapter in DSM-IV-TR, that is pica, rumination disorder (RD), avoidant/restrictive food intake disorder (ARFID) [11].

Feeding disorders are defined as a set of symptoms and behaviours related to an inadequate quantity of food intake that does not satisfy nutritional needs and causes quantitative and/or qualitative alterations of nutritional status. The main difference between feeding disorders and EDs is the absence of body shape and weight concerns or disturbance of body image as a main feature in the development of feeding disorders. Consequently, in these psychiatric conditions the eating behaviour is not enacted with the aim to control body weight or shape.

According to the DSM-5 criteria, AN is characterized by a persistent restriction of food intake leading to a very low body weight; an intense fear of gaining weight or of becoming fat, or a persistent behaviour that interferes with weight gain even though the body weight is significantly low; an altered experience of body weight and shape, an excessive influence of body shape and weight on self-evaluation, a persistent lack of recognition of the severity of the current underweight. In DSM-5 classification, two subtypes of AN are included: restricting type, based on a persistent restriction of food intake in order to control the body weight and shape; binge-purging type, characterized by restriction of food intake associated with episodes of binge eating and compensatory behaviours [11].

The diagnosis of BN is made when patient's recurrent episodes of binge eating are associated with the use of inappropriate compensatory behaviour to avoid weight gain, and their self-evaluation is influenced by body shape and weight.

An episode of binge eating has two main features: eating, in a defined period of time (e.g. 2 h), an amount of food significantly larger than most people would eat in the same period and in similar circumstances and a sense of lack of control over eating during the binge eating episode. People with BN use recurrent compensatory behaviours to prevent weight gain such as self-induced vomiting, use of laxatives, diuretics or other medications, fasting or excessive exercise. Furthermore, in BN binge eating episodes and compensatory behaviours must occur at least once a week for 3 months, and not exclusively during an episode of AN.

Unlike DSM-IV [22] and DSM-IV-TR [23], DSM-5 officially recognizes binge-eating disorder (BED) as a formal diagnosis. BED is characterized by the presence of binge eating episodes that must be associated with at least three of the following features: eating more rapidly than normal; eating until feeling uncomfortably full; eating a large amount of food when not hungry; eating alone because of feeling embarrassed by how much one is eating; feeling disgusted with oneself, depressed or guilty after the episode.

Unlike BN, in BED the binge eating episode is not associated with any compensatory behaviour and this is the reason why patients with BED are often overweight or obese. As in BD, a diagnosis of BED is made when the episodes of binge eating have a frequency of at least once a week for the last 3 months [11].

AN, BN and BED share the concern about body shape and weight, which leads these patients to dysfunctional eating behaviours in order to try to modify or control their body. In the same chapter of these eating disorders, the DSM-5 also includes pica, rumination and ARFID, which are distinguished from EDs mainly by the absence of fear of gaining weight or concern about body shape/weight.

The term pica originates from the specific Latin epithet of the Eurasian magpie 'corvus pica', introduced by Linnaeus in the second half of the eighteenth century [24] in reference to bird characterized by appetite for a diversity of objects, including inedible objects [25]. The DSM-5 describes pica as a persistent craving and eating of non-food substances. This behaviour has to be judged as inappropriate to the developmental level of the person (includ-

ing persons with ID/ASD), not part of a cultural practice, and severe enough to deserve specific clinical attention when occurring during the course of other health or clinical conditions such as pregnancy [26, 27], psychotic disorders [28, 29], obsessive-compulsive disorder [30, 31], some forms of masked depression [32] or ID and ASD themselves. In order to fulfil the DSM-5 criteria, this behaviour must last longer than a month. Various substances may be craved, including cigarette butts, clay, raw starch, paint, paper, string, clothes, dirt, dust, plastic, hair, metal, rocks, foliage and faeces. Ingestion of clay (geophagia), raw starch (amylophagia), faeces (coprophagia) and ice (pagophagia) are reported to be the most common types of pica [33–35].

Rumination disorder (RD) is defined as the presence for at least 1 month of repeated regurgitation of food that may be re-chewed, re-swallowed or spit-out, not due to a medical condition nor in the presence of other EDs. When RD occurs in patients with other mental disorder, it has to be severe enough to warrant clinical attention.

Finally, avoidant/restrictive food intake disorder (ARFID) is described as a persistent inability to fulfil appropriate nutritional needs, associated with one of the following: nutritional deficiency, loss of weight, dependence on oral nutritional supplements or enteral feeding and inference with psychosocial functioning. This behaviour is not associated with lack of available food and is not part of a cultural practice. Furthermore, it does not occur in the presence of other EDs and is not associated with disturbance of experience of body shape and weight. When ARFID manifests in patients with other medical conditions, it has to be more severe than the eating symptoms typically associated with that medical condition, and, therefore, warrants clinical attention [11].

25.3 Prevalence of Feeding and Eating Disorders

The lifetime prevalence of EDs is estimated to be up to 4% for AN, 2% for BN and 2% for BED [36]. In recent decades EDs seem to have become more common, except for BN. The reason for this increase remains unclear, although it is suggested that it may be due to better detection by clinicians [2]. As for feeding disorders, there is a lack of prevalence data among general population and also among people with ID/ASD. Indeed, the prevalence of pica in the general population is unclear and clinical studies are lacking also among people without ID or ASD. In individuals with ID, prevalence of pica seems to increase with the severity of the ID [11]. The prevalence of RD is 'unclear' according to the DSM-5 criteria. Moreover, diagnosis of RD cannot be made in the presence of another ED, but there is evidence of the presence of rumination behaviour in individuals with other EDs [37]. To date, ARFID has not been the subject of any large-scale epidemiological study, therefore, the prevalence of ARFID among the general population remains unknown [38], and the same applies to its prevalence among individuals with ID/ASD.

Studies have documented higher incidence of eating and feeding problems in people with ID and ASD [39, 40] than in the general population. A range of 3–42% of adults with ID living in congregate residential settings and 1–19% of those living more independently in the community have diagnosable EDs [4, 10]. More generally, a variety of problems with eating have been estimated to prevail in about one third of people with ID [41, 42] and up to 80% in those with more severe impairment, problem behaviours and co-occurring ASD [41–45].

Nevertheless, as for other psychiatric disorders, the real prevalence of FEEDs in people with ID is probably underestimated because most of these patients do not present symptoms as described in the DSM and ICD diagnostic criteria for the general population and their psychopathological co-occurrence is not identified (see Chap. 9).

A variety of problems with eating have been estimated to prevail in about one-third of people with ID and even more in those with higher severity of cognitive impairment, problem behaviours and co-occurring ASD.

25.4 Aetiopathogenesis of FEEDs

Thousands of studies have attempted to specify exactly what causes eating and feeding disorders. The current consensual approach to integrating the various factors that contribute to these conditions is the 'biopsychosocial' model, which has the advantage of taking into account several factors that are associated with an increased risk, ranging from the broadly cultural to the narrowly biological, and stopping along the way for relational, personality and other psychological factors [46, 47].

As from the DSM-5 criteria reported above, all EDs are characterized by a loss of physiological meaning of nutrition. Food loses its value of nourishment and pleasure and it assumes a negative connotation. Consequently, in EDs we find that feeling of hunger and satiety are no longer determinant upon the power supply, that appears qualitatively and quantitatively based on arbitrary parameters such as rigid rules or concept of control. All EDs are defined by pathological behaviours such as dieting, vomiting, avoiding specific foods, binge eating combined in different patterns and resulting in specific medical complications including weight loss, obesity, heart or kidneys failures and blood changes. Moreover, EDs are frequently associated with several psychiatric comorbidities, such as anxiety and mood disorders, or personality disorders, which makes the long-term outcome poor [48]. Longitudinal studies indicate that most patients migrate among diagnoses over time, mostly among AN, BN and BED [20, 49] without a substantial change in fundamental psychopathological features [48, 50, 51]. This evidence points towards a common psychopathological core of EDs.

Because of the tendency to migrate among ED diagnoses and the relevant overlap between ED symptoms and different medical and psychiatric conditions (e.g. obesity, obsessive compulsive disorder, autism spectrum disorders, depression, substance abuse, personality disorders), many researchers focused on the different behavioural patterns, rather than the full syndrome [52].

This tendency leads to create a new clinical perspective which is expressed in the concept of 'spectrum'. As for ASD and other clinical conditions within psychiatry, the term 'spectrum' has been used to refer to etiologically related clinical features; eating disorders spectrum involves core, atypical and subclinical symptoms and signs, personality and behavioural traits that belong to a group of psychiatric syndromes related to eating disorders. The 'spectrum approach' may be useful to identify subjects with vulnerability to EDs that do not meet criteria for a clear diagnosis, or individuals with prodromes to a full-blown disorder or with sequelae of a previous disorder [53].

In the DSM-5, EDs are defined as 'characterized by persistent disturbance of eating or eating-related behavior that results in the altered consumption or absorption of food and that significantly impairs health or psychosocial functioning' [11]. Considering the three main ED diagnoses, AN, BN and BED, the dysfunction of eating behaviours can be considered as the clinical expression of specific cognitive and emotional disturbance [20, 54, 55], including the overestimation of the body shape and weight concerns, and personal identity [56].

Persons with EDs have a tendency to overvalue their body shape and weight [20], and to evaluate themselves on the basis of their ability to control their body weight and shape, instead of valuing themselves on the basis of work performances or relationships, as people without EDs are supposed to do. Consequently, we may better describe EDs as a psychiatric condition based on a disturbance of subjective perception of body and personal identity; in particular, it has been suggested that the main characteristic of EDs patients is suffering from a specific disorder of lived altered reality contributing to an abnormal constitution of one's identity [57].

To better understand the overvaluation of body shape and weight in EDs patients, it is important to introduce the concept of 'lived body'. This concept comes from phenomenology that distinguishes between lived body (or body subject) and physical body (or body

object) [58]. 'Lived body' is the primitive experience of body in the first-person perspective, the cenesthetic self-awareness of one's own body not mediated by reflection [57, 59]. On other hand, the physical body, or body object, is the body studied by natural sciences, as anatomy and physiology, that can be manipulated (e.g. by surgery), in the third-person perspective [58, 59].

The lived body is the centre of three main dimensions of experience [60]: the experience of oneself (self-awareness); the object-experience; and the experience of other people. First of all, the primitive self-awareness is based on personal experiences, such as emotions or perceptions, as well as on actions and thoughts. It is not a conceptual representation, neither an object-awareness coming from the observation of oneself. We can use the term 'ipseity', created by Henry [61], to explain this primitive form of self-awareness that is immediate, non-observational and pre-reflexive. Ipseity has two main features: self-ownership, which is the awareness that I am the one who is living an experience, and self-agency, which is the consciousness that I am the one who is taking an action. These two characteristics allow the differentiation between me and the object I perceive, and between my representation of the object and the object itself. It is the lived body that provides the 'ipseity', the immediate self-awareness that allows the engagement with the environment [59].

Secondly, according to Husserl, lived body is the protagonist of the making experience of objects just as I perceive them intuitively. Consequently, a modification of the lived body causes an alteration in the perception of the external world. The lived body is at the same time the responsible of perceptions and the place of their integration. We can consider it as the instrument that allows the relationship with the world itself, notably by perceiving objects as a part of a context in which the individual is engaged. In this view, human beings understand environment as long as they live it, thanks to the constant projection of their body into it, and their knowledge of objects does not require a merely theoretical cognitive act, but rather a concept which manipulates things and allows the use of them [62].

Moreover, the lived body has been proposed to be the centre of intersubjectivity, that is, the experience of other people, based on the immediate and pre-cognitive perception of the linkage between my own and other's body, by transferring the corporeal schema to another human being. This process, which allows to recognize others as similar to myself, is defined 'intercorporeality' and is considered the basis of intersubjectivity [63]. Intercorporeality is the fundamental of all interactions based on behaviour and it is present ahead of explicit communication. Proprioception seems to be involved in this process by promoting the understanding of other persons' perspectives through immediate body-to-body attunement.

An interesting integration to Husserl's dimensions of corporeality (body subject and body object) has been introduced by Sartre [64], who suggested that individuals can know their own body also from the observation of it by another person ('lived body-for-other'). According to this view, the realization that one's own body can be looked at by another person would favour its perception as an object and the extremization of this process would lead to a progressive loss of the autonomous consciousness of one's own body, up to the belief that it can be modified by the gaze of others.

Persons with EDs often have difficulties in distinguishing their emotions [65–68] as well as in having a first-perspective perception of their own body and a sense of themselves as embodied agents, which implicates unstable sense of identity and awareness of feelings, that is, what one likes or dislikes [69]. People with EDs feel a sense of extraneity from their own body and they often try to get back in touch with their physical dimension through starvation or other objectified aspects [57]. They experience their own body as an object for other people's senses and feelings, rather than the result of their own somato-kinaesthetic perceptions and emotions, and tend to construct their self-identity basing on the way they feel looked at by others (see Fig. 25.1).

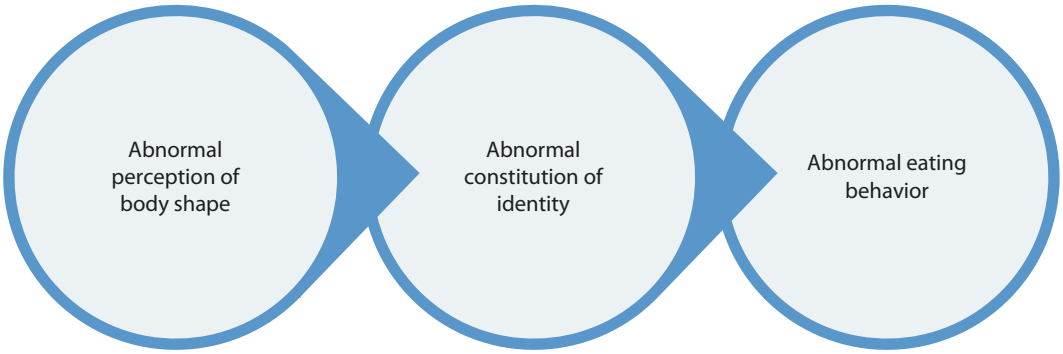

Fig. 25.1 The relationship between body image, self-identity and eating disorder

Individuals with EDs experience a disturbance of the body image that causes an alteration of the process of shaping their personal identity. In EDs patients, the identity disorder is considered as a maintaining factor of the eating disorder mostly through two main features: perfectionism and low self-esteem [70]. In addition to the control of body shape and weight, patients with EDs show perfectionism in almost all the areas where performance is required (work, sport), and are concerned with achieving unattainable ideals or unrealistic goals, which often lead to an aggravation of underlying low self-esteem.

These hypotheses are supported by the data collected by using a self-reported questionnaire called IDEA (IDentity and EAting disorders), aimed at identifying psychological vulnerability factors in persons with abnormal eating habits [69]. This questionnaire assumes that most pathological eating behaviours and features are a consequence of the severity of abnormal bodily experiences and identity disorders, and it is focused on the phenomenological areas explained above: (1) feeling oneself through the gaze of the other, (2) defining oneself through the evaluation of the other, (3) feeling oneself through objective measures, (4) feeling extraneous from one's own body, (5) feeling oneself through starvation, (6) defining one's identity through one's own body, (7) feeling oneself through physical activity and fatigue [69].

The IDEA was tested in a population of individuals who did not suffer from EDs, in which it resulted to be able to identify vulnerability factors in persons with abnormal eating patterns and significantly higher identity alterations in those with clinically evident EDs than in those without a full-blown pattern of symptoms [57]. When used among obese patients, the IDEA test confirmed that abnormal body experience may represent a key risk factor for developing an eating disorder [71].

The IDEA test explores mainly two phenomena: abnormal corporeality and difficulties in defining self-identity. These results support previous clinical impressions that specific dysfunction of lived corporeality, namely experiencing one's own body as an object being looked at by another (rather than from a first-person perspective), and of personal identity, namely defining one's own self by the way one feels being looked at by others and through one's ability to control one's shape and weight (rather than through other kinds of personal quality), are supposedly core features of ED psychopathology.

The main difference between eating and feeding disorders is the absence of body shape concern and/or weight phobia in individuals with feeding disorders.

25.4.1 Physical, Sexual Abuse and Neglect in Individuals with Intellectual Disability: Potential Risk Factors for Eating Disorders

Emotional and physical abuse represents important risk factor for the development of EDs, especially when occurring during child-

hood. In reference to the general population recent meta-analyses [72, 73] show a strong association between childhood abuse (i.e. physical, sexual and emotional abuse and neglect) and prevalence, severity and early onset of EDs, especially BN, BED and AN binge purging subtype [73]. Persons with ID and/or ASD are significantly more exposed to different kinds of abuse than peers with other types of disability and peers without disability, with prevalence rates ranging from 7% to 34% [74–81]. More specifically, sexual abuse is reported to occur in 14–32% of children with ID and 7–34% of adults, physical abuse in 22–26.8% of children, and emotional abuse in 17.5–26.7% of children [74, 75, 80]. Victims of sexual abuse have been found to be mostly females, aged between 11 and 20 years, and living with their parents [82]. The abuse usually takes place in victim's house and is perpetrated by someone known by the victim [82].

Causes of maltreatment are complex, involving stressors of various kinds that interact with family, parent and child characteristics. Environmental stressors include poverty, educational deprivation, low socio-economic status and unemployment. Family and parent factors include social isolation, problems in attachment or interaction between parent and child, parental substance abuse, mental illness or neurodevelopmental disability, and parents' own experience of childhood abuse. Increased parental stress and a lack of social supports are associated with increased aversive, coercive and inconsistent discipline. Child factors include temperament, personality, physical characteristics, co-occurrence of attention deficit-hyperactivity disorder, problem behaviours, age-inappropriate sexual behaviour as well as obviously emotional and cognitive immaturity [80, 83–85].

Within child maltreatment, sexual abuse represents the main risk factor for the development of all EDs [86–88], especially BN [88]. In some cases, this relationship between a history of sexual abuse and EDs, especially BN, seems to be mediated by dissociative experiences, which in fact are reported to be more common among bulimic women overall and particularly in those who experienced sexual abuse during childhood. Some authors have argued that dissociation would allow the bulimic to shift (more or less episodically) to a lower level of consciousness in order to be able to binge and purge, while other authors suggested that binge-eating behaviour (with or without purging) might develop as a means of inducing dissociation or reduced levels of self-awareness [89–92].

However, dissociation is widespread across many different psychiatric disorders, not exclusively in EDs [93] and is challenging to identify in persons with ID and low-functioning ASD, mostly due to their insight and communication difficulties. Furthermore, in these persons neither the role of dissociation in the development of binge eating behaviour nor BN itself appear to be measurable.

In addition to abuse, the scientific literature indicates that many other negative life events are associated with the onset of EDs across the life span [73, 94]. In the general population some eating behaviours have been found to be instrumental in shunning or avoiding trauma-related emotions or thoughts [94], and it is probable that this type of reaction to trauma also happens in people with ID/ASD. Addressing this issue in future research could significantly improve our understanding of psychological mechanism underlying FEEDs in individuals with ID/ASD, and improve standards of care both for eating symptoms and trauma-related manifestations.

Emotional and physical abuse represents important risk factor for the development of EDs, especially when occurring during childhood, and persons with ID and/or ASD are significantly more exposed to different kinds of abuse than peers with other types of disability and peers without disability.

Tip

The role of dissociation in determining specific presentation of EDs in persons with ID/ASD and the pathogenic contribution of negative life events deserve high attention in future research and clinical practice.

25.4.2 Aetiology and Pathogenesis of Specific FEEDs in Persons with ID and Low-Functioning ASD

25.4.2.1 Anorexia Nervosa

In adults with ID, anorexia nervosa has been found to be associated with previous dieting, identity problems, sexual issues, bereavement, regression, family conflicts and history of psychopathology in family members [4].

Scientific literature includes reports of anorexia nervosa in persons with Down syndrome [95–98], also in association with depression [99–101] as well as in persons with Prader-Willi syndrome [102–104] and to a lesser extent in those with phenylketonuria [105].

25.4.2.2 Bulimia Nervosa and Binge-Eating Disorder

Very little is known on possible causes of bulimia nervosa and binge-eating disorder in persons with ID and or low functioning-ASD. Hyperphagia, including gorging, pica, food hoarding, and polydipsia, has been found to associate with distress in persons with hyperphagic short stature syndrome, a condition sharing many signs and symptoms with Prader-Willi syndrome but having slightly lower IQ reduction, normal or low body mass index and different aetiology [106, 107].

25.4.2.3 Avoidant/Restrictive Food Intake Disorder

Feeding difficulties including apparent lack of interest in food and food refusal have been reported in persons with Down syndrome [108], Joubert syndrome, Lesch-Nyhan syndrome, Smith-Lemli-Opitz syndrome, Sotos syndrome and Willem Bueren syndrome, mainly during childhood. Similar feeding difficulties also occur in Prader-Willi syndrome and Costello syndrome but only in the very first months of life [109–111].

Some features associated with ID and ASD such as the difficulty to identify and express one's feelings or specific sensory sensitivities may contribute to restrictive eating.

Persons who received supplementary feeding in the form of beverages or smoothies for a long time often show food refusal and gagging when only or mainly textured food is offered, especially during childhood.

Other factors associated with persistent lack of interest in food and food refusal are experiences of abuse or neglect as well as high level of anxiety, expressed emotion and psychopathology among family members and/or other significant caregivers [112].

25.4.2.4 Rumination Disorder

Within persons with ID, RD occurs more frequently in males, in those with more severe IQ impairment and in case of co-occurrence of ASD, anxiety disorders and other FEEDs [113]. No specific genetic syndromes have been associated with RD.

Persons with ID may begin to engage in ruminating behaviour due to an exacerbation of dysphagia, gastroesophageal reflux disease or pyloric stenosis. Other gastric sensory and motor dysfunction may also represent contributing factors [114]. As for other FEEDs, high level of anxiety, expressed emotion and psychopathology among family members and/or other significant caregivers may represent contributing or precipitating factors [113].

25.4.2.5 Pica

In persons with ID, prevalence rate of pica has been reported to be higher in persons with more severe IQ impairment, co-occurrence of ASD, depressive disorder, ADHD, anxiety disorders and personality disorders as well as in males, children, adolescents and younger adults, and those with weight problems [4, 25, 35, 110].

High occurrence of pica in persons with Prader-Willi syndrome [115, 116] and Kleine-Levin syndrome [117] has been reported.

Some researchers have associated pica with problems with impulse control and the broad spectrum of the obsessive-compulsive disorder. In some cases, this association was supported by the finding of decreased serotonin metabolite concentration in cerebral spinal fluid and frontal lobe hypoactivity on

SPECT (single photon emission computed tomography) [4, 30, 118–120].

Pica is sometimes found in conjunction with vitamin, minerals or oligoelements deficiencies, although the direction of this relationship is not well understood. Some researchers have suggested that pica may represent an instinctive attempt to replenish low micronutrients levels from non-food substances, which could be effective in some cases and non-adaptive in others. Conversely, other researchers suggested that micronutrients deficiencies could be a consequence of pica, as pica materials may limit absorption and metabolism of specific minerals or oligoelements [121]. In general, there is no clear evidence of correlation between low level of specific nutrients and the onset of pica as well as the type of non-food ingested [25, 121].

Addiction to specific substances may play a role in specific forms of pica, such as nicotine addiction in cigarette butts pica [4].

Psychosocial theories on pica have described an association with poverty, environmental deprivation, family stress and small social support network [4, 122, 123].

25.5 Diagnosing Issues in Persons with ID and Low Functioning ASD

As explained above, the psychopathological core of EDs, especially AN, BN and BED, implicates disturbance of body image and body perceptions, low self-esteem, difficulties in self-definition and identity impairment, which adults with ID especially of severe degree are unlikely to be able to articulate [4]. Consequently, the clinical evaluation of the psychopathological core of these disorders remains challenging.

Some have tried to investigate body image among people with ID [124, 125]. One of these studies underlines that people with ID tend to underestimate their body weight [125]. In this study, researchers use the 'Stunkard Figure Rating Scale' (SFRS, [126]) to assess body dissatisfaction and body perception. The SFRS scale is based on drawings of nine male and nine female figures ranging in size from underweight to obese; participants have to indicate the figure associated to overweight, underweight and healthy weight by their point of view. They are also required to indicate which figure they see as more similar to their own body [125].

The results of this study [125] suggest that most individuals with ID have a correct general conceptualization of appropriate weight, but they do not apply this concept to themselves. This could be one of the reasons of the high prevalence of overweight (BMI 25.1–30 kg/m^2) and obesity (BMI > 30 kg/m^2) among these persons, with rate ranges of 28–71% and 17–43%, respectively [127]. Similar rates have been found in adolescents with ID, with overweight varying between 11% and 24.5% and obesity between 7% and 36% [128, 129]. It is possible that the underestimation of body weight underlying these alterations is based on a wrong transposition of what observed on other persons to themselves as well as to inability to contextualize general rules and concepts [125].

Others studies addressed the relationship between body perception and EDs in persons with ID [124, 125]. A recent review suggests that both social context or experiences and social identity may contribute to a negative body image and subsequent ED [124]. Persons with ID were found to present higher vulnerability for AN and higher association with relational factors than those described for the general population. For example, Dymek and le Grange described a girl with mild ID and AN, who used her weight loss as a 'strategy' for gaining attention from her caregivers [130]. Difficulties in interpreting beliefs, attitudes and behaviours of other persons, especially peers and significantly others, may represent and adjunctive vulnerability factor. Cottrell and Crisp reported on a person with Down syndrome and moderate ID who developed AN after having long heard her caregivers to associate weight losing and physical attractiveness [96].

Studying and managing EDs in persons with ID and low-functioning ASD is challenging not only because of the difficulty in identifying alterations of body image, self-

Table 25.2 The multiaxial structure of DC-LD

Axis I	Axis II	Axis III	Appendix
Severity of intellectual disability	*Cause of intellectual disability*	*Presence of additional psychiatric disorder*	*Appendix I:* Learning disability syndromes and behavioural phenotypes
		Pervasive developmental disorder (Axis III level A)	*Appendix II:* Other associated medical condition
		Psychiatric illness (Axis III level B)	*Appendix III:* Factors influencing health status and contact with health services
		Personality disorder (Axis III level C)	
		Problem behaviours (Axis III level D)	
		Other disorders (Axis III level E)	

Cooper et al. [14]

esteem, identity or other core aspects of EDs, but more generally for the atypical presentation and course of the full range of symptoms and the peculiarities of risk and pathogenic factors [131]. As mentioned, diagnostic criteria developed for the general population, including DSM and ICD criteria, need to be adapted to be used with persons with ID and/or low-functioning ASD. Even the dimensional approach, as proposed by the latest edition of the DSM, has limitations, as it does not allow the establishment of abnormal eating behaviour and what associated alterations of weight and mental or social functioning should be considered as constituents of a core feeding or eating disorder [4].

25.5.1 Specific Diagnostic Approaches for FEEDs in People with ID

To overcome these diagnostic problems, adaptations of the diagnostic criteria for the general population to persons with ID were developed. The adaptation to ICD-10 criteria was produced by the Royal College of Psychiatrists in 2001 with the name of 'Diagnostic Criteria for Psychiatric Disorders for Use with Adults with Learning Disabilities/Mental Retardation – DC-LD' [10], while those to DSM-IV-TR and DSM-5 were realized by the National Association on Dual Diagnosis (NADD, United States of America) with the name of Diagnostic Manual – Intellectual Disability (DM-ID [12]) and Diagnostic Manual – Intellectual Disability 2 (DM-ID 2 [13]), respectively (see ▶ Chap. 5).

The DC-LD are based on a multiaxial and hierarchical differential diagnostic approach that allows to clinically describe problem behaviours in adults with ID and to classify them as a manifestation of ID itself, co-occurrence of ASD, psychiatric disorder, personality disorder or none of these (see Table 25.2) [14]. The application of this hierarchical and multiaxial approach to eating behaviour makes the differential diagnosis between abnormal eating behaviours and EDs possible, and allows multiple diagnoses [5].

DC-LD also allows the evaluation of symptoms and signs of EDs in persons with ID basing on the information provided by their caregivers and other significant proxies (see Fig. 25.2). DC-LD includes data about type, frequency and duration of abnormal

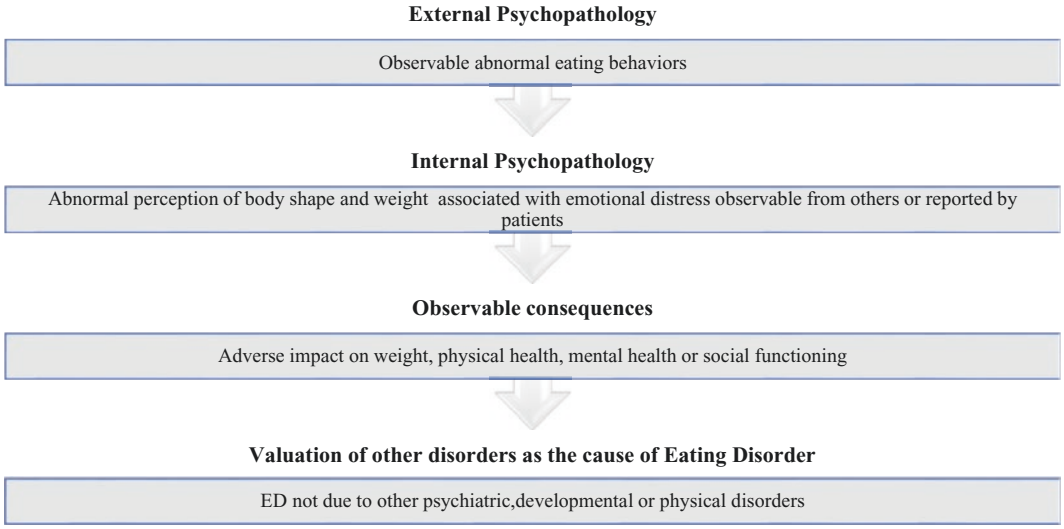

Fig. 25.2 The DC-LD model for eating disorders in adults with ID. (From Gravestock [5])

eating behaviours as well as their impact on weight, and physical and mental health, all of which represent useful information for making a diagnosis of specific EDs [5].

In addition, this multiaxial approach provides clinical case formulations useful for the assessment and differential diagnosis of abnormal eating behaviours and EDs. Published data show encouraging results in using DC-LD criteria to achieve a more accurate description of psychiatric disorders and problem behaviours in adults with ID [14, 132], but further studies are needed to address the limitations of these criteria in reference to EDs, especially among adults with more severe ID and/or co-occurrence of ASD.

DM-ID 2 encompasses seven types of applications of the DSM-5 criteria to persons with ID, which includes addition of symptom equivalents, omission of symptoms (not existing or not identifiable in persons with ID), changes in symptom count, modification of symptom duration, modification of age requirements, addition of explanatory notes and exclusion of criteria sets (not applying to persons with ID). Unlike the DSM system, DM-ID 2 does not rely on self-report and its criteria subsets are principally concerned with observational or behavioural symptoms and signs [13].

The DM-ID 2 criteria for FEEDs are presented with two columns: one column lists the DSM-5 criteria and the other one lists the adapted criteria for the full range of severity of ID. Other diagnostic chapters have three columns, with the middle column referring to mild-to-moderate ID and the right-hand one to severe-to-profound ID [13].

Despite recent advances, also due to the use of these adapted criteria, many issues of FEEDs diagnosis in persons with ID are still far from being solved, starting from the definition of a boundary between abnormal eating behaviours and EDs.

25.6 Presentation and Clinical Issues of FEEDs in Intellectual Disability

25.6.1 Abnormal Eating Behaviours: Clinical Features and Hypothetical Meanings

Abnormal eating behaviours may have different impacts and meanings in subjects with ID and/or low functioning ASD. Apart from nutritional consequences, some studies underline that eating behaviour seems to assume a more communicative/relational function for people with ID in comparison with the general popula-

tion [133, 134]. This hypothesis is partially supported by some data which show that people with moderate-to-severe ID and physical illness that makes it difficult for them to both eat and communicate, are more likely to have eating dysfunctions [135]. The same study reported an interesting association between dysfunctional eating behaviour and aggressive behaviour, which would deserve further investigation.

Kuhn et al. [133] evaluated the possible relationship between rumination and social skills in persons with ID. They found that these persons show significant deficits in social skills (e.g. participating in leisure or occupational activities with others) when compared with non-ruminant persons with ID [133]. This result confirms previous data about the association between lower level of social adaptive skills and stereotypies [134]. Researchers have hypothesized that stereotypic behaviour such as rumination may develop in the absence of environmental/relational stimuli as a kind of compensatory sensory self-stimulation, and get to structure progressively until it becomes the only possible relational modality [108, 133, 136]. Rapid eating has also been associated with relational difficulties, especially with preventing one from contacting the reinforcers associated with eating with others and sitting down over a period of time [112].

Tip

The compensatory role of rumination for lack of environmental/relational stimuli in persons with ID/ASD represents an interesting field of research that could also lead to new therapeutic strategies aimed at improving dysfunctional eating behaviour by increasing social skills.

25.6.2 Key Aspects of Clinical Presentation and Differential Diagnosis of Specific FEEDs

25.6.2.1 Anorexia Nervosa

Cognitive symptoms such as fear of gaining weight and distorted body image can be difficult to assess, especially in persons with more severe ID and communication impairment. The presence of persistent behaviour that interferes with weight gain has to be considered the most important symptom and needs accurate assessment through additional observation from family members and other significant caregivers. The use of pictures may also be helpful to evaluate self-perception of body image.

Food refusal may present with atypical symptoms and problem behaviours such as temper tantrums, keeping lips sealed, turning the head away, spitting, throwing cutlery, self-injury (up to self-mutilation), vomiting in the face of caregivers, masturbation, compulsive singing, compulsive laughing and apathy [113].

In persons with ID/low functioning-ASD, anorexia nervosa has to be carefully differentiated from physical illnesses and other psychiatric disorders, especially depression, ADHD, obsessive compulsive disorder, psychotic disorders (delusions of poisoning) and anxiety disorders. In fact, as aforementioned, understanding the reasons of food refusal may be challenging as well as identifying cognitive symptoms of anorexia nervosa. Clinicians should also consider that anorexia nervosa may co-occur with other psychiatric disorders, especially anxiety disorders, mood disorders and ADHD [113, 137].

25.6.2.2 Bulimia Nervosa

As for anorexia nervosa, cognitive and self-reported symptoms can be difficult to observe and to elicit. Furthermore, careful assessment and comprehensive information is needed to ascertain secretive inappropriate behaviours.

25.6.2.3 Binge Eating Disorder

Feelings of self-disgust, sadness or guilt after binges may be difficult to identify. The presence and frequency of binge eating episodes should be defined at the light of availability of food, which can be excessive in many living contexts of person with ID/ASD.

25.6.2.4 Avoidant/Restrictive Food Intake Disorder

As general food refusal, also selective food refusal may present with atypical symptoms and problem behaviours, especially in persons

with major cognitive and communication difficulties, who cannot communicate which aspects of food are aversive. Inability to identify and express one's feelings may also contribute to restrictive eating as a means of internalizing distress [138].

The sensory peculiarities often present in persons with ID and ASD can lead to refusal of some or many foods, as the colour, texture or smell may result unpleasant. In this case a diagnosis of co-occurrent ARFID should be made with caution, only if the food restriction cannot be explained by the specific manifestations of the ASD.

Food refusal can also be a component of a co-occurrent anxiety disorder or ADHD. Loss of appetite is a common response to anxiety and stress, due to the activation of the sympathetic nervous system and the other changes the body makes as part of the fight-or-flight response. ADHD can affect eating behaviour because of distractibility, lack of interest in eating and high levels of arousal at mealtimes [137, 139]. Even in these cases and in those of other psychopathological co-occurrences, the issue of differential or adjunctive diagnosis of ARFID must be carefully considered.

25.6.2.5 Rumination Disorder

In individuals with ID and/or low functioning-ASD, regurgitation could be due to self-stimulatory or self-soothing behaviours. It could also be associated with physical problems that individuals are not able to describe, such as dysphagia, gastroesophageal reflux disease, pyloric stenosis or gastroparesis. Careful history taking and specific testing such as high-resolution oesophageal manometry with pH impedance are key to an effective differential diagnosis.

The occurrence of regurgitation in the context of an anxiety disorder, especially a generalized anxiety disorder, should also be excluded.

25.6.2.6 Pica

Persistent eating of non-nutritive, non-food substances should be distinguished from the inability of many persons with ID and/or low functioning-ASD to control hunger and to differentiate between food and non-food substances as well as prepared and unprepared or discarded food [113].

There is a strong correlation between pica and other mental illnesses, including ADHD, depressive disorders, anxiety disorders, post-traumatic stress disorder, obsessive compulsive disorder and other FEEDs. In all these cases, the issue of differential or adjunctive diagnosis of pica must be carefully considered.

25.6.3 Feeding Disorders: Quantitative and Qualitative Medical Implications

As described above, feeding disorder is characterized by inadequate quantity of food intake that does not satisfy nutritional needs and causes quantitative and/or qualitative alterations of nutritional status. Persons with ID/ASD often have overweight and obesity as consequences of abnormal eating behaviours, poor education about dietary habits, healthy life styles and nutritional potential of food, as well as low physical activity, due in turn to frequent pain, fatigue, athletic inability and limited mobility [4, 140–142]. Research has found that sex and age can also increase the risk of obesity, with females and adults showing higher rates than males and adolescents [143, 144]. Obesity appeared also significantly associated with some genetic syndromes, such as Down syndrome [145] and Prader-Willi syndrome [146]. Persons with Prader-Willi syndrome report compulsive eating and binge eating probably because of their hypothalamic and endocrine abnormalities which are responsible of an impairment of the food satiety response [147].

On the other hand, low weight (BMI < 18 kg/m^2) also represents a frequent quantitative alteration of nutritional status in individuals with ID, with rates ranging from 5% to 43% [4] and generally higher than those of the general population [148], although much less attention has been paid to this condition than to overweight and obesity.

There are a number of factors associated with ID that increase the risk of becoming underweight such as abnormal eating behav-

iours, avoidant/restricted intake of food, food refusal and food selectivity. Neuromotor alterations of the upper gastrointestinal tract (i.e. chewing problems, dysphagia, regurgitation, gastroesophageal reflux, cyclic vomiting) and consequent dehydration, aspiration and asphyxiation, and upper respiratory infection are frequent and may also contribute to nutritional deficiency [149].

The causal contribution to low weight of sex-related factors is not clear, as is the sex ratio of low weight prevalence. Hove found it to be higher in men with ID (9%) than in women (7%), while Emerson did not find any difference in reference to any age [148].

In summary, overweight/obesity and low weight in people with ID/ASD can be defined as a multifactorial manifestation of their complex mental and physical vulnerability. Both conditions determine a high risk of further serious health problems. Low weight is associated with increased susceptibility to infections, anaemia, osteoporosis, fertility issues, persistent fatigue and several dermatological problems, while obesity increases the risk of coronary heart disease, ischemic stroke, type II diabetes, osteoarthritis and cancers of the breast, colon, prostate, endometrium, kidney and gallbladder [7].

In persons with ID and/or ASD, overweight and obesity are the most common medical consequences of abnormal eating behaviours, although underweight is also rather frequent.

Rapid eating and low mastication also present threats to one's safety and health, in both the short- and long term. Immediate risks include the possibility of choking or aspiration, vomiting, acid reflux and poor digestion, whereas more long-term risks include overweight, obesity, insulin resistance, type 2 diabetes and metabolic syndrome [150–153]. Rapid eating has also been found to decrease the taste and smell sensory stimulation by food and determine lower satisfaction with food. Fast eaters tend to rate their meals as less pleasant and to eat more food to compensate, compared with slow eaters [154].

Table 25.3 Potential complication of pica

Toxicity	Hypokalemia or hyperkalemia, poisoning, deficiency of vitamins and iron
Obstruction	Intestinal obstruction with possible perforation, peritonitis, and death
Excessive caloric intake	As a consequence of the craving for food items such as carbohydrate and fats
Nutritional deficit	Failure to thrive, achlorhydria
Other	Dental injury, infections, infestations

Adapted from Sayetta [122] and Rose et al. [77, 155]

Rapid eating is a relatively understudied behaviour that also falls under the broad group of FEEDs, although it does not easily fit into either the skills deficit or behavioural excess categories since the alteration refers to the amount of time between one bite of food and another, which is too short and does not allow adequate chewing.

FEEDs can also have serious negative implications on qualitative nutritional status and consequently on general health. Among FEEDs, pica which represents the most common condition determining qualitative alteration of nutritional status in person with ID, with prevalence rates ranging from 4% to 26% [4]. Pica has numerous medical complications that have been gathered in five groups: inherent toxicity, obstruction, excessive calorie intake, nutritional deprivation and other complications [155] (see Table 25.3). Complications depend on the substance ingested. Iron deficiency anaemia is particularly common in persons with geophagia and pagophagia, while lead intoxication is more frequently associated with ingestion of flaking paint or plaster. Parasitic infestation (e.g. toxocariasis, toxoplasmosis, ascariasis, giardiasis, cysticercosis) often follows the ingestion of soil contaminated with the excreta of dog, cat and pork [35].

Definition and clinical characteristics of pica have been provided in the ▶ Sect. 25.2.1 of this chapter.

Rumination, another frequent feeding/eating disorder determining qualitative alteration of nutritional status, may also cause considerable health issues such as halitosis, lethargy, irritability, malnutrition, dehydration, electrolyte abnormalities, renal damage, parotid swelling, dental erosion and aspiration pneumonia, which can be lethal in 12–20% of chase, if not treated [4]. Also definition and clinical characteristics of rumination disorder have been provided in ▶ Sect. 25.2.1 of this chapter.

ARFID encompasses clinical conditions previous categorized by GOS (Great Ormond Street) classification system in: food avoidant emotional disorder (significant food intake restriction in absence of body and shape concerns), selective eating (selectivity based on food non nutritional characteristics such as taste, colour, temperature, texture, and smell), and functional dysphagia (fear of swallowing, vomiting or choking that cause anxiety) [156]. GOS developed this classification in order to distinguish different clinical patterns that encompass in the DSM-IV group of Eating Disorders not Otherwise Specified (EDNOS) [22].

Very little is known about ARFID prevalence and its medical consequences, with some exception for malnutrition and low body weigh that often necessitate hospitalization and tube feeding [157].

In summary, we can affirm that abnormal eating behaviours and feeding disorders in individuals with ID/ASD cause serious medical problems as a result of quantitative and qualitative alterations of nutritional status. FEEDs also complicate the management of all other health, social and educational needs of persons with ID/ASD, especially during infancy and childhood and in presence of more severe cognitive and relational impairment [41, 158]. Furthermore, these conditions have a strong negative impact on the quality of life of persons with ID and their caregivers.

These data indicate the need for clinicians and other professional operators to pay more attention to risk factors, early warning signs and symptoms of FEEDs in order to achieve timely diagnosis, prevention of complications and treatment. Standardized assessment tools can assist clinicians and the whole multidisciplinary team in the systematic identification of problems to be targeted for further clinical assessment. Beside generic screening tools for any psychiatric disorder (including FEEDs), few specific tools for FEEDs have been produced such as the STEP (Screening Tool of Feeding Problems; [42]).

25.7 Anorexia Nervosa and Autism Spectrum Disorder

A large body of literature considering the association between ASD and EDs, especially AN, has been published over the years since 1983, when Gillberg identified some similarities in the cognitive profiles of the two conditions [159, 160] and proposed an empathy defect to be a core feature of AN and its pathogenesis [53]. Some communalities between AN and ASD pointed out by the scientific literature refer to impairments of theory of mind (the ability to impute independent mental state in other individuals), central coherence (the ability to have a global prospective instead of focusing on details) and set shifting (the ability to shift from one action to another fluently, with no difficulty) [161]. Other similarities concern rigid attitudes and behaviours, especially towards food, and excessive focus on self. This tendency to an extreme self-attention can be interpreted as a consequence of the difficulty to empathize with other persons and associated psychopathological features such as anhedonia, deficit of emotional intelligence and alexithymia, which have been reported for both AN and ASD [162].

In ED populations, ASD prevalence is reported at rates of around 23–30%, especially among female individuals and in reference to anorexia nervosa [161, 163, 164]. Furthermore, it has been suggested that autistic traits in childhood increase the risk

of developing an ED during adolescence and adulthood, especially AN [165]. Conversely, eating problems and food selectivity are frequent in individuals with ASD, especially during childhood and adolescence [166].

ASD has been identified as a possible non-specific familiar risk factor for AN and a family history of AN has been associated with a risk increase of ASD [167].

Nevertheless, it is not clear whether ASD aspects in EDs represent psychopathological dimensions (quantitative variations along a continuum) that can be found also in other psychiatric disorders or they really describe the coexistence of the two nosographic categories (qualitative differences from other psychiatric disorders). In fact, beside the above-mentioned aspects in common, AN and ASD show main differences such as later age of onset of AN, higher prevalence in females for AN and in males for ASD, and more heterogeneous IQ levels for ASD than for AN [168].

Tchanturia et al. [169] found ASD aspects not to correlate with the core physical symptoms of AN, such as low BMI, malnutrition or starvation, suggesting that the link between ASD and AN could go through mental dysfunctions rather than their physical consequences. Moreover, the same authors showed that in persons with ASD, AN presents with more severe symptoms, higher association with depressive symptoms and poorer social functioning than in the general population.

Some researchers and clinicians indicated that persons with ASD may require longer and more intensive treatments for AN than other patients without ASD, suggesting that the presence of ASD can contribute to treatment resistance to conventional therapies [169]. AN patients with ASD-related social cognitive difficulties have been reported to have lower engagement in family-based treatment and other main psychotherapeutic interventions [169]. Nevertheless, no clear evidence have been provided to date on the impact of ASD traits on treatment outcomes of patients with AN.

Tip

The main similarities between AN and ASD are some cognitive and behavioural patterns: restricted interest and activities, difficulties in social interaction and communication, repetitive behaviours.

25.8 Treatment

FEEDs include a host of heterogeneous problems that vary in nature and it is highly unlikely that one or two therapeutic interventions can be effective in all instances. Multi/interdisciplinary and tailored therapy is key, based on the complex interactions among individual and disease characteristics, including type of behaviour, frequency, intensity, chronicity and cause.

Clinical management of FEEDs implies continuous risk monitoring for medical complications and the need for hospitalization, including monitoring of vital signs (e.g. heart rate < 40 bpm), weight status (e.g. extremely low body mass index), severe blood abnormalities (e.g. severe anaemia or electrolyte imbalances), treatment fails (e.g. decline in oral intake or family and caregivers support) and other psychiatric conditions (e.g. suicide risk) [170].

Normalization of nutritional status and dietary patterns are central features of the treatment of many FEEDs, especially anorexia nervosa, bulimia and avoidant/restrictive food intake disorder. As far as possible, oral-enteral nutrition should be favoured over parenteral nutrition, which should only be used as a last option. When appropriate, nutritional supplements should also be considered, including phosphate, thiamine, zinc, potassium and vitamins [170].

Psychotherapy is deemed as a central part of the treatment of all FEEDs, with behavioural therapy being the most evidence-based approach in persons with ID and/or ASD [112]. Several researchers have found antecedent interventions to be effective in altering the

aversive characteristics of the feeding context and/or non-preferred food as well as increasing the value of reinforcement for food consumption, and reducing the response effort for engaging in the target behaviour [112]. Examples of antecedent procedures are represented by simultaneous presentation, high-probability instructional sequence, food texture manipulation, presentation of bites of food and fading of bites demand. Simultaneous presentation involves the provision of non-preferred foods at the same time as preferred foods and the association of specific preferred tastes or stimuli (e.g. preferred condiments) to non-preferred foods [171, 172], while the high-probability instructional sequence consists in presenting 2–3 responses which the individual has a high probability of complying with, just before the non-preferred food [173–175].

Reinforcement and extinction procedures, especially differential reinforcement of alternative behaviour and escape extinction, have also received considerable attention in research and practice on treatment of FEEDs. Several kinds of reinforcers such as attention, favourite objects and/or preferred foods have been provided contingent upon food consumption and not at any other time [176–178]. Two of the most utilized extinction procedures include non-removal of the spoon and physical guidance [179, 180].

Some specific behavioural treatments have been practiced and studied for specific FEEDs. Rapid eating has been treated through reinforcement of pauses between bites of food, shaping of longer pauses and prevention of attempts to eat rapidly by prompting the person to wait [181], while some cases of rumination disorder have been improved by use of modelling and reinforcement in diaphragmatic breathing training and/or behavioural relaxation training [182]. Pica has been addressed mainly through behavioural treatments involving reinforcement and response reduction, with and without aversive components. Most studied aversive stimuli methods are lemon juice, food aversion and water mist [183, 184], while overcorrection has been the most considered of the work and effort methods, although it is now largely out of favour and replaced by other methods such as negative practice, habit reversal and response effort. In general, current clinical practice is drifting away from aversive techniques and relying more on positive procedures such as environmental enrichment, reinforcement schedules and eclectic methods [184].

Family-based therapy is increasingly being recommended for younger neurotypical patients [185] and could be of some utility also for many persons with ID and/or ASD but evidence on practicability and effectiveness is still lacking.

Research on pharmacological treatments of FEEDs in persons with ID and/or ASD is scarce. Antidepressants represent the most used class of drugs, specifically the SSRI (selective serotonin reuptake inhibitor) fluoxetine for the treatment of bulimia nervosa and co-occurrence of depressive disorders in anorexia nervosa, albeit with several restrictions and in combination with psychotherapy whenever possible. Selective serotonin reuptake inhibitors seem to be useful also for those cases of pica that seem to lie within the obsessive-compulsive spectrum [30].

Topiramate has been shown to have some efficacy on bulimia and binge eating disorder, although reports are limited and contradictory [186–188]. Orlistat and liraglutide could be of some utility for weight loss in these conditions and obesity [189–191].

Baclofen, a γ-aminobutyric acid agonist with antispasmodic activity, has been used for rumination disorder. It increases post prandial LES (lower oesophageal sphincter) pressure and reduces the number of its transient relaxation episodes. Another pharmacological therapy that has shown some benefit for rumination disorder is the antipsychotic levosulpiride, a selective dopamine D2-receptor antagonist with prokinetic activity.

Surgical treatment represents an extreme therapeutic ratio for rumination syndrome, in particular patients with low basal LES pressure on HREM (high-resolution oesophageal manometry) with evidence of GERD (gastroesophageal reflux disease) based on 24-h pH impedance testing may benefit from Nissen fundoplication refractory to behavioural and pharmacological therapy [192].

Key Points

In this chapter, we focused our attention on the clinical manifestations and the impact of eating and feeding disorders for individuals with ID. It appears that there is still a lot to study in order to clarify the role of these clinical syndromes in people with ID and other developmental disorders. This topic could be crucial for the development of a new management of eating symptoms in people with ID.

The main key points of this chapter are the following:

- With respect to people with ID it is challenging to investigate their disturbance of body image, one of the most important elements in the development of EDs. This issue limits the knowledge about prevalence data, psychopathological and clinical features of EDs in adults with ID.
- Subjects with ID are often victims of physical, sexual and psychological abuse. The role of abuse in the aetiopathogenesis of EDs, well known in adults without ID, could be a key element and should be evaluated also in ID population.
- People with ID often demonstrate serious medical consequences of feeding disorders and/or abnormal eating behaviour such as overweight and obesity, most frequently.
- Autism spectrum disorder and anorexia nervosa demonstrate to have some clinical features in common. The relationship between these two psychiatric conditions needs further studies.

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Sleep Disorders/ Sleep-Wake Disorders

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Learning Objectives

- To understand the biological, genetic, and psychosocial risk factors for the development of sleep abnormalities in individuals with intellectual disability and/or low-functioning autism spectrum disorder.
- To understand how sleep disorders in the ID/ASD population can be diagnosed especially when there are comorbid conditions such as seizure activity, medical conditions, problems behaviours, and neurodegenerative and psychiatric disorders.
- To understand the issues around diagnosis and management at different levels of intellectual functioning.
- To discuss treatment options and approaches to sleep disorders in this population.

26.1 Introduction

Sleep disturbance is a common problem in people with intellectual disability (ID) and/or autism spectrum disorder (ASD) due to their biological (including genetic) and psychosocial risk factors that predispose them to sleep abnormalities [1, 2].

Sleep is a dynamic state that is essential for human physical and mental health, including cognitive performance and emotional reactivity. Experimental sleep deprivation has been shown to negatively impact on all the human body communication systems (nervous, endocrine, and immune) and determines a wide range of negative effects on mood, cognitive, and psychomotor performance, glycogen sensitivity, lipid metabolism, immune responses to endotoxins, and many other physiological set points, also suggesting that persistent sleep restriction may accelerate some processes of ageing [3–9].

From a neurophysiological point of view, two main types of sleep can be distinguished: desynchronized sleep or sleep with rapid eye movements (REM), which is often associated with vivid dreams and intense brain activity, and synchronized sleep also known as sleep without rapid eye movements (non-rapid eye movements, NREM), which is associated with reduced brain activity. According to Rechtschaffen and Kales' model, which is meant as a reference method for sleep analysis and scoring, NREM sleep accounts for 75–80% of total sleep time in adults and can be split into four stages [10].

REM and the four NREM stages make up the sleep macrostructure, in which brain electrical activity occurs in coordinated and repeated cycles, normally four to six cycles per night. The initial cycle lasts 70 to 100 minutes while the following cycles are longer, 90–120 minutes each, with the amount of REM also increasing over the cycles up to 30% in the later cycle of the night [3, 11].

The average individual's sleep episode begins in NREM stage 1, which usually lasts 1–7 minutes in the initial cycle, constituting 2–8% of total sleep, and is easily interrupted by disruptive noise. In this stage brain electrical activity [as detectable through the electroencephalogram (EEG)] varies from rhythmic alpha waves to low-voltage, mixed-frequency waves. Alpha waves are associated with a wakeful relaxation state and are characterized by a frequency of 8–13 cycles per second. Stage 1 sleep may also play a transitional role in sleep-stage cycling [3].

Stage 2 sleep lasts approximately 10–25 minutes in the initial cycle and lengthens with successive cycles, constituting between 45% and 55% of the total sleep episode. To awaken from this stage of sleep, a person needs more intense stimulation than in stage 1. Brain electrical activity shows relatively low-voltage and mixed-frequency waves, with characteristic sleep spindles and K-complexes. It has been hypothesized that sleep spindles and K-complex are important for memory consolidation because of their high presence in individuals who learn a new task. K-complex has also been suggested to have other relevant functions such as the maintenance of interruption of sleep when encountered with a stimulus that the brain evaluates as harmless or potentially dangerous, respectively, and the maintenance of synapse homeostasis, which would be exerted by

determining a sort of "reboot" for the neural connections [3, 12].

Stage 3 lasts only a few minutes and constitutes about 3–8% of sleep while stage 4 lasts approximately 20–40 minutes in the first cycle and makes up about 10–15% of sleep. Stage 4 presents the highest arousal threshold of all NREM stages. Because of the increased amount of high-voltage and slow-wave activity on the EEG (i.e. delta waves), stages 3 and 4 are collectively referred to as slow-wave sleep, most of which occurs during the first third of the whole sleep episode [3].

In the last 20 years, Rechtschaffen and Kales' description of sleep processes and associated scoring rules have increasingly been criticized [13] and alternative guidelines have been developed such as those by the American Academy of Sleep Medicine, in which sleep stages S1–S4 are referred to as N1, N2, and N3, with N3 merging S3 and S4; stage REM is referred to as stage R [14]. Other approaches have also been employed for the automated categorization of sleep stages using modern multi-channel EEG [15–17].

Sleep structure and sleep propensity derive from the interactions of circadian and sleep-wake-dependent oscillatory processes developed by a hierarchy of tissue-specific structures located throughout the body and coordinated by the suprachiasmatic nucleus (SCN) of the hypothalamus [18]. These tissue-specific rhythms are based on light input from the outside world and cortisol release during daytime and by melatonin secretion during the dark hours. Damage to the SCN eliminates the circadian rhythms of many behaviours, including sleep [19]. The melatonin secretion pathway extends from the SCN to the paraventricular nucleus (PVN) of the hypothalamus, the upper thoracic spinal cord, superior cervical ganglion, and the pineal gland [20]. Cortisol level is reduced during slow-wave sleep while it rises rapidly in the middle of the night and peaks during the biological morning. Cortisol pathway projects from the SCN to the subparaventricular zone of the hypothalamus, the dorsomedial nucleus, and the medial parvocellular part of the PVN. The latter stimulates the release of corticotropin releasing factor, which has also been implicated in the control of sleep-wake rhythm as well as in the promotion of REM sleep [21].

The sleep phase is regulated by neural structures that switch off the brain's arousal systems, allowing it to fall asleep. The preoptic region of the hypothalamus contains a large number of these neurons but the sleep system is also heavily influenced by inputs from other parts of the brain such as the emotional and cognitive areas of the forebrain as well as the lower brainstem, which relay information about the state of the body (e.g. a full stomach makes it easier to fall asleep). The sleep-generating system also involves neurons in the pons, which regulate the transitions from NREM to REM phases during the night. These neurons transmit signals to the lower brainstem and spinal cord, resulting in muscle atonia, rapid eye movements, and erratic autonomic activity, all of which are hallmarks of REM sleep [3]. NREM sleep is controlled mostly by the thalamus and the cerebral cortex, although other structures like the basal forebrain, anterior hypothalamus, cerebellum, caudal brain stem, spinal cord, and peripheral nerves contribute to its regulation and modulation [22]. REM sleep is activated by the brainstem, particularly the pons and adjacent portions of the midbrain, and deactivated by the dorsal nuclei of the raphe, the locus coeruleus, and the deep mesencephalic reticular nucleus [23, 24].

The major pathways of the ascending arousal influence originate in the upper brainstem. One of these arises from cholinergic neurons in the upper pons, which stimulates portions of the thalamus that are responsible for preserving sensory input delivery to the cerebral cortex. Another pathway starts from monoamine neurotransmitter-containing cell groups in the upper brainstem and joins the hypothalamus rather than the thalamus, where it receives input from nerve cells containing peptides such as orexin and melanin-concentrating hormone. These signals then travel to the basal forebrain, where they receive additional information by acetylcholine and gamma-aminobutyric acid-containing cells, before entering the cerebral cortex to provide a diffuse activation and

preparation for the interpretation and analysis of sensory data [3].

Sleep is induced and maintained by the activity of acetylcholine, gamma aminobutyric acid, serotonin, norepinephrine, and a number of cytokines, hormones, nucleosides, and prostaglandins, while waking is more associated with glutamate, dopamine, hypocretin, and histaminergic systems, although many of these and other neurotransmitters and neuromodulators are involved in both states [25].

Insomnia and other sleep disturbances are common in mental disorders, especially depression and anxiety disorders, and many researchers have proposed a causal interplay between sleep and psychopathological conditions, which has been largely supported by contemporary neurobiology studies [26]. This association appears to be even stronger in persons with ID, ASD, attention deficit hyperactivity disorder, and other neurodevelopmental disorders than in the general population, with repeated findings of very high co-occurrence of sleep issues, problem behaviours, and psychiatric disorders [27–30].

The most common theory describes disturbed sleep as a causal mechanism for other psychiatric problems, which seems to be supported by the common observation that treating insomnia reduces the occurrence of other mental health issues. Common causes for sleep and mental disorders have also been proposed to explain this high coexistence and interrelation [31].

In persons with ID and/or ASD these difficulties of differential diagnosis and other diagnostic issues are even greater than in neurotypical persons since the peculiarities of clinical presentation and consequent overshadowing with common comorbid conditions such as epilepsy, respiratory illnesses, problem behaviours, neurodegenerative, and psychiatric disorders.

26.2 Prevalence

Sleep problems are commonly reported complaints of individuals with ID, with an estimated prevalence exceeding that reported for individuals in the general population [32–34]. According to the literature, the prevalence of sleep disorders in children with ID ranged from 25.5% to 36.2% with an average duration of between 6 and 9 years [35]. In adults with ID, the prevalence seems to vary between 8.5% and 34.1% and in the most severe forms, it reaches 9.2% [29]. However, when actigraphy has been used to evaluate insomnia, 72% of a large community sample of people with ID was found to suffer from sleep problems [36]. It is important to notice that the majority of the available studies on sleep disorders in people with ID concentrate on insomnia. The sleep-wake rhythm of adults with ID has been reported less stable, fragmented, and with a lower relative amplitude [37]. Furthermore, the evidence indicates that individuals with higher severity or comorbid disabilities are at an increased risk of developing sleep difficulties [38]. The factors that have been most frequently associated with the presence of sleep disorders are problem behaviours, respiratory disease, vision problems, psychiatric disorders, and intake of psychotropic drugs, especially anti-epileptics [28, 29]. Fragmented sleep has also been associated with lower physical activity, dementia, epilepsy, sensory impairments, and spasticity [37]. Sleep disorders have been reported more commonly in the case of difficult temperament, anxious or disorganized attachment behaviours, parental exhaustion, inexperience, psychosocial stressors and anxieties, conflicts over sleeping arrangements. Particularly, in the case of family psychopathology, the condition of maternal depression has been observed to lead to a higher chance of transferring to the child genetic risk factors for sleep and mental disorders [39–41]. Moreover, depending on the changes during the lifetime, sleep disorders may be treated as part of developmental disorders. Indeed, it is well established that the prevalence of sleep disorders varies depending on brain maturation and development and with the modification of environmental expectations and demands. At the molecular level, the timing of gene-environmental interactions changes across the lifespan and is regulated by the modifications of the environment previously stated [42, 43]. Finally, it has been observed that different rapid eye movement

(REM) characteristics are typical of different psychiatric situations and could concur to define endophenotypes specific for the considered disorder. For example, depression has been found to be characterized by a decreased REM sleep latency and an increased REM density. In schizophrenia, on the other hand, electrophysiological, pharmacological, and neurochemical activities have all been found to be encountered during REM sleep. Similarly, in ID and dementia-specific REM sleep disturbances have been reported [44].

Sleep problems are frequent in people with ID, with an estimated prevalence higher than that of the general population. These disturbances are common in both children and adults, especially in individuals with higher severity or comorbidities.

26.3 Development and Maturation of Physiological Sleep Timing

The sleep in infancy is marked by the circadian biological rhythms that correspond to zeitgebers, which is the night-day cycle, and in this case, can be identified as the night sleep and day attentiveness [45–48].

Most of the sleeping processes are learned and not innate, as it is commonly believed. Indeed, during early infancy, the schemas of sleep are characterized by "active sleep", which has short inactivity to REM-like activity, except for some characteristics of the REM phase such as atonia and muscle paralysis.

The period between 3 and 6 months of the child is characterized by non-REM sleep. In fact, the majority of the sleep cycle is characterized by the sleep spindles, K complexes, and slow wave activity.

On the other hand, the older infants' sleep cycle is dominated by the slow wave, as the non-REM activity is present for 50% of the time during sleep [45–48].

Finally, as the age progresses, there are changes in REM latency and density, sleep efficiency deteriorates as it becomes more desynchronized and less restorative. Indeed, there are some physiological changes such as the reduction of melatonin production and the decline in access and sensitivity to light and other zeitgebers.

26.4 Etiopathogenesis of Sleep Abnormalities in Neurodevelopmental Disorders

26.4.1 Biological Factors

The incorrect function of melatonin activity has been found to have effects on sleep phase disorders and also to affect psychopathology. Specifically, the production of melatonin is found to be dysfunctional in people with ASD and ID [49, 50]. Indeed, in these populations, sleep is fragmented, erratic, and the transition between cycles can be challenging to identify. Besides, people with ASD or people who are visually impaired are more likely to have an irregular sleep-wake cycle that is not regulated by light, as they have a desynchronized sleep rhythm with zeitgebers that lead them to depend on social cues to have a regular sleep [51, 52].

However, there is no evidence that poor levels of melatonin are the cause of disruptive behaviour in the ID population despite it is clear that subjects with severe/profound ID, ASD, and visual impairments do not produce enough melatonin, they may secrete it during the day instead of at night, or they metabolize it promptly. These characteristics can explain the reason why these people may not respond to melatonin.

Hypersomnia may be caused by another sleep disorder (e.g. narcolepsy or sleep apnoea), dysfunction of the autonomic nervous system, or drug or alcohol abuse. In some cases, it results from a psychopathological condition such as depression, a neurological problem such as multiple sclerosis, epilepsy, encephalitis, a tumour, head trauma, or injury to the central nervous system, or other physical health issues such as obesity. Certain medications, or medicine withdrawal, may also cause hypersomnia. Some people appear to

have a genetic predisposition to hypersomnia; in others, there is no known cause.

Narcolepsy-cataplexy is most commonly caused by a loss of hypocretin-producing cells in the lateral hypothalamus. Hypocretin is a neuropeptide vital for the regulation of the sleep-wake cycle by its influence on the histaminergic, nonadrenergic, serotonergic, and cholinergic systems. Low cerebrospinal fluid hypocretin levels are found in the wide majority (90–95%) of patients with narcolepsy and typical cataplexy and can be used to diagnose the conditions, while they are present in only 10–30% of cases of narcolepsy without cataplexy. Narcolepsy-cataplexy is tightly associated with a specific human leukocyte antigen (HLA)-allele, namely HLA-DQB1*0602, suggesting an autoimmune process to underlie the destruction of hypocretin-producing neurons [53].

For parasomnias, a link between a delayed maturational process of sleep associated with a greater amount of slow wave sleep (SWS) in adulthood has been postulated, which would be confirmed by the high prevalence of these group of sleep disorders in persons with ASD, ID, and other neurodevelopmental conditions [54].

26.4.2 Genetic Factors

People with ASD and Smith-Magenis syndrome can exhibit a switch in melatonin synthesis and release [49]. Besides, people with Angelman's syndrome and complex epilepsy can have an interrupted sleep caused by anticonvulsants, dysfunctional GABA receptors, arousal problems, and nocturnal seizures [2, 55]. Indeed, nocturnal frontal lobe seizures are related to nicotinic cholinergic receptors, and for this reason, they have to be distinguished from parasomnias and REM-sleep related behaviours [56, 57].

Moreover, some physical changes can be related to sleep problems. Indeed, the changes of the facial morphology that are present in Down syndrome and Fragile X syndrome and the obesity that characterizes Prader-Willi syndrome are associated with breathing disorders which can lead to obstructive sleep apnoea syndrome (OSAS) [58, 59]. Owing to midface and maxillary hypoplasia as well as relative macroglossia and glossoptosis, persons with Down syndrome have a narrower bony dimension of the upper airway with superficially positioned tonsils, relative tonsillar and adenoidal encroachment, and increased secretions. Furthermore, they show generalized hypotonia and increased risk of hypothyroidism and obesity, which are also risk factors for OSAS [1, 58–60]. Main predisposing factors of persons with Fragile X syndrome are represented by high arched palate and generalized hypotonia [2]. Many persons with Prader-Willi syndrome show altered hypocretin levels in the cerebrospinal fluid that are associated with hypersomnia, and cataplexy, but only one-third of these cases possess the above-mentioned HLA-DQB1*0602 haplotype [2].

Diverse behavioural phenotypes have been recognized in people with ID, such as the presence of polysomnographic phenotypes [61], while people with ASD, Down, and Fragile X syndromes have problems in sleep architecture [2, 62]. Persons with Williams syndrome show an increase of non-rapid eye movement percentage, slow-wave sleep, number of leg movements, irregular sleep cycles, respiratory-related arousals, and slow-wave sleep, while rapid eye movement sleep percentage is reported to be significantly decreased [63, 64]. Rett syndrome is characterized by frequent night-time awakenings, bruxism, and difficulty falling asleep, although these issues show an age-related decreased across the life-span. Night screaming and night laughing in particular are strongly related to large deletions in MECP2 [2].

Moreover, a link has been found between the level of ID and total sleep time, frequency of awakenings, eye movements during REM-sleep, and stability in sleep architecture [65]. In addition, the Smith-Magenis syndrome presents a sleep disorder as the main symptom, which in part is correlated with the inverted process of melatonin production [66].

26.4.3 Psychosocial Factors

Nowadays, there are some lifestyle and habit changes within the society that can lead to sleep disorders or insufficient sleep. These are high-stress levels that can be due to work or academic demands, time spent in natural and artificial light, use of computers in late evenings, TV and other electronic devices, obesity, fixed bedtimes, use of pharmaceuticals, and other psychosocial or cultural factors [67].

Mainly, the same psychosocial and environmental issues affect normal population as well as neurotypical and people with ID. However, their impact on people with ID can be different depending on the level of ID and the presence of other risk factors for disrupted sleep. Besides, the adverse effects of relative insufficient sleep can negatively affect the quality of life of people with ID and increase the rates of disruptive and destructive behaviours [50]. Indeed, children and several adults with ID, when they experience insufficient sleep, instead of fatigue and sleepiness, may show hyperactivity, disruptive or stereotypic behaviours, irritability, and self-injurious or aggressive behaviour [68].

26.4.4 Developmental Factors Through the Lifespan

There is still little information on the developmental trajectory of brain "ageing" in several subpopulations with ID. Indeed, despite the neuroimaging and post-mortem studies made to gain data on the brain ageing and dementia in people with Down syndrome [69], there is a general lack of information on the remaining and more heterogeneous ID population. Currently, some investigators are addressing the link between emerging tauopathies, synucleopathies, cerebrovascular disease, and REM parasomnias in people with ID.

In persons with ID and/or ASD non-organic sleep disorders can be caused by various biological, psychological, and environmental factors, including genetic anomalies, alterations in the melatonergic system, psychiatric problems, distress, medications, and ageing.

26.5 Criteria and Clinical Features

There are many distinct sleep disorders, but the majority of them are characterized by one of the following symptoms: "excessive daytime sleepiness, difficulties initiating or maintaining sleep, or abnormal movements, behaviours, and sensations occurring during sleep".

DSM-5 [70] offers the classification of ten main sleep-wake disorders (eight main groups), conditions that are manifested by disturbed sleep that cause both distress and impaired functioning during the daytime:

1. Insomnia disorder. It is the most frequent sleep disorder. The diagnostic criteria include problems falling asleep, difficulties maintaining major sleep, and difficulties with daytime attention, concentration, memory, and executive function.
2. Hypersomnolence disorder. It is characterized by daytime sleepiness.
3. Narcolepsy. It is characterized by periods of sleep attacks, rapid eye movements, sleep paralysis, and in some cases episodes of cataplexy (partial or total loss of muscle control, often triggered by a strong emotion such as laughter).
4. Breathing-related sleep disorders. They are associated with daytime sleepiness and fatigue and include the following:
   - Obstructive sleep apnoea-hypopnoea. It is characterized by repetitive episodes of airflow reduction (hypopnoea) or cessation (apnoea) due to upper airway collapse during sleep.
   - Central sleep apnoea. It is characterized by a lack of drive to breathe during sleep, resulting in repetitive periods of insufficient ventilation and compromised gas exchange.
   - Sleep-related hypoventilation. It is characterized by an abnormal increase in partial pressure of carbon dioxide ($PaCO_2$) and a decrease in PaO_2 during sleep.
5. Circadian rhythm sleep disorders, including:
   - Advanced sleep phase syndrome. It is a disorder in which the major sleep episode is advanced in relation to the

desired clock time that results in symptoms of compelling evening sleepiness an early sleep onset, and an awakening that is earlier than desired.
   - Irregular sleep-wake type. It is characterized by numerous naps throughout the 24-hour period, no main night-time sleep episode, and irregularity from day to day.
   - Non-24-hour sleep-wake type. It is defined as comprising daily delays in sleep onset and wake times.
6. Parasomnias, characterized by abnormal behavioural, experiential, or physiological events that occur during sleep, specific sleep stages, or sleep-wake transitions, including the following:
   - Non-REM (NREM) sleep arousal disorders. They are recurrent episodes of incomplete awakening from sleep. During these episodes, patients may experience sleepwalking or sleep terrors. Sleepwalking can be associated with sleep-related eating or sleep-related sexual behaviour (sexsomnia).
   - Nightmare disorder. It involves repeated severe anxiety dreams during REM sleep.
   - REM sleep behaviour disorder. It involves abnormal behaviour during the sleep phase with REM sleep.
7. Restless legs syndrome. It is characterized by an unpleasant tickling or twitching sensation in the leg muscles when sitting or lying down, relieved only by moving the legs.
8. Substance- or medication-induced sleep disorder. It is the diagnostic name for insomnia and other sleep problems which are caused by the use of alcohol, drugs, or certain medications.

The ICD-10's [71] categorization of sleep disorders was based on the now-outdated distinction between organic and non-organic illnesses, resulting in the inclusion of "non-organic" sleep disorders in the ICD-10's [71] mental and behavioural disorders chapter, and "organic" sleep disorders in other chapters (i.e. diseases of the nervous system, diseases of the respiratory system, and endocrine, nutritional, and metabolic disorders). In ICD-11, a distinct chapter for sleep-wake disorders has been formed, which includes all pertinent sleep-related illnesses, regardless of whether they are more or less biological or psychological in nature, as following: [72]

- Insomnia disorders
- Hypersomnolence disorders
- Sleep-related breathing disorders
- Circadian rhythm sleep-wake disorders
- Sleep-related movement disorders
- Parasomnia disorders
- Other specified sleep-wake disorders
- Sleep-wake disorders, unspecified

Sleep-related movement disorders are characterized by basic, often stereotyped movements that disrupt sleep or its onset, and include restless legs syndrome, periodic limb movement disorder, sleep-related leg cramps, sleep-related bruxism, sleep-related rhythmic movement disorder, benign sleep myoclonus of infancy, propriospinal myoclonus at sleep onset, sleep-related movement disorder due to a medical condition, and sleep-related movement disorder due to a medication or substance. Unlike other sleep-related movement disorders, restless legs syndrome is mainly characterized by a waking, sensorimotor experience, but it has been included in this group of disorders because it almost always also includes periodic limb movements during sleep.

Parasomnias include disorders of arousal from non-REM sleep, parasomnias related to REM sleep, and other parasomnias. In the ICD-11 [72] the articulation of disorders of arousal from non-REM sleep is somewhat different from that of the DSM-5 [70] and includes confusional arousals, sleepwalking disorder, sleep terrors, and sleep-related eating disorder. Confusional arousals are marked by mental confusion or disturbed behaviour (e.g. disorientation, becoming unresponsive, slurred or slow speech, poor memory) during partial arousal from deep sleep. Like in all other disorders of arousal from non-REM sleep, partial or complete amnesia for the event can be present as well as inappropriate

or absent responsiveness to efforts by others to intervene or redirect the person during the episode.

The category "other parasomnias" includes hypnagogic exploding head syndrome, sleep-related hallucinations, and parasomnia disorder due to a medical condition. The impression of a sudden, loud noise, or a sensation of a violent explosion in the head is the main symptom of the hypnagogic exploding head syndrome, which usually happens while the person is falling asleep but episodes may also occur with awakening during the night. Sleep-related hallucinations are hallucinatory events that occur during the falling asleep phase (hypnagogic hallucinations) or while awakening from sleep (hypnopompic hallucinations). Visual hallucinations are the most common, but they may also involve auditory, tactile, or kinaesthetic experiences.

Sleep disorders in the population with ID can be the consequence of decreased adaptability, an attachment disorder, and poor sleep-related hygiene, combined with immature or dysfunctional sleep networks, primary psychiatric disorders, visual impairments, and psychosocial factors [68].

Most people with severe ID generate wrongly timed or insufficient melatonin production and sleep-induced peptides to initiate sleep. In contrast, others are unable to suppress the production of melatonin during daylight hours, which disturbs the circadian cycle and sometimes leads to behavioural disturbance [73].

Most people with ID show aberrant sleep rhythms, such as slow wave/REM, sleep fragmentation, restless legs syndrome, sensitivity to medication side effects, nocturnal epilepsy, or other physiological problems associated with sleep onset. Indeed, it has been stated by Kaplan and colleagues [74] that people with Rett, Down, Prader-Willi, Smith-Magenis, and Cornelia de Lange syndromes, along with those with orofacial and upper airway anomalies, are more likely to have obstructive sleep apnoea and disrupted sleep [65, 75]. In this case, these people are more likely to develop irritability, hyperactivity, and increased aggression or self-injurious behaviour (SIB) instead of excessive daytime somnolence. A correlation has been established between regressive subtype of ASD and REM-related parasomnias [76]. REM may bring to the integration of the cortical, mesencephalic, and pontine management of motor generators and maybe to the rise of atypical cortical neurones and head circumference associated with these syndromes [77, 78].

Moreover, it has been established that sleep disruption and fragmentation are extremely common in people with ID. Shibagaki, Kiyono, and Mastruno [79] carried out a study on 79 children with ID that suggested an increased rate of abnormal sleep physiology in people with ID and cerebral palsy. Besides, the majority of sleep dysfunctions are common pathways for several excitatory or inhibitory brain networks.

Also, Okawa and colleagues [80] studied four congenitally blind children with ID and stated that the relationship between social deficits and severe ID inhibited the use of alternative zeitgebers.

Several other studies identified sleep problems in children with autism [81–83], sleep-related breathing disorders in adults with Down syndrome [84], and a more complex correlation between insomnia and ID [29], suggesting that different sleep disorders can be associated with specific genetic disorders. Besides, more complex genetic disorders along with neuropsychiatric disorders imply a multidirectional correlation between primary and secondary sleep disorders.

26.5.1 Children with Intellectual Disability

Brain maturation and the interaction between neurones related to sleep mechanism drive sleep physiology. It has been stated that many sleep disorders are developmental, such as primary non-REM parasomnias, which are more likely to occur along with stressors or medical or psychiatric disorders in adulthood [42, 85, 86]. There are other disorders that are

highly heritable such as restless legs syndrome and periodic limb movements [87, 88].

However, the majority of sleep disorders come from the interaction between genes and the environment and the boundaries between syndromes are semi-permeable. In children, the supremacy of slow wave sleep overlaps with the emergence of non-REM parasomnias. In addition, the rate of nightmares is influenced by sleep phase instability and traumatic experiences in childhood, while, during adolescence, they grow less but are replaced by sleep phase delay, narcolepsy, persistent enuresis, chronic sleep deprivation, insomnia, substance use, and increase in psychiatric disorders [86, 89].

Finally, insomnia, irregular sleep-wake cycles, fragmentation of sleep, and REM-related parasomnias are common in children with ASD and ID [50, 90, 91].

26.5.2 Adults with Intellectual Disabilities

Sleep difficulties are a lifelong condition in ID population, continuing to persist into adolescence [92] and adulthood [93, 94].

Insomnia is frequent in adults with ID, ASD, and other neurodevelopmental disorders, comorbid psychiatric or neurological disorders, post-traumatic stress disorder [95], and in certain behavioural phenotypes [65]. Nevertheless, people with severe ID, ASD, and insomnia can also have other sleep dysfunctions that remain unidentified or misunderstood with seizure activity or severe psychiatric disorder [51, 96].

Also, the prevalence of sleep-related breathing, central and hypoventilation disorders, REM, periodic limb movements, declines in vision and hearing, and less sensitivity to the light (zeitgebers) usually increase with age. Besides, blindness and severe visual problems are more frequently related to irregular sleep-wake cycle disorder. On the other hand, non-24-hour sleep wake cycle disorder has been observed in adult patients with ID and/or ASD, sensory impairments and mood disorders. Fragmented sleep has also been found to be common in people with ASD, traumatic brain injury, and dementias. Furthermore, REM-related movement disorder has a link with the neurodegenerative disorders and progressive supranuclear palsy (synucleopathies), dementias, and vascular disorders.

Insomnia, irregular sleep-wake cycles, fragmentation of sleep, and other sleep difficulties represent frequent life-long conditions in persons with ID and/or ASD, with onset in infancy and continuous course across adolescence, adulthood, and old age.

26.6 Specific Assessment

26.6.1 Issues Related to Diagnosis in People with Neurodevelopmental Disorders

In order to diagnose sleep disorder in people with ID it is necessary to analyse the available sleep-related evidence, identify the link between sleep changes and daytime behaviours, and complete the assessment and differential diagnosis. However, several aspects need more in-depth investigations. Indeed, people with ID insufficiently report or cannot report their own sleep disorders. Besides, clinicians do not always have the most up-to-date in-depth knowledge of the links among sleep physiology, atypical brain development, and the different phenotypes of sleep disorders. Furthermore, the steps for allowing reasonable adjustments to make sense of the information gathered from people with varied levels of severity of ID can be difficult [32]. Finally, there can be some training deficits as clinicians belong to different schools that give importance to different aspects. For example, some of them do not collect the sleep history of the patient, while others do not use direct observations.

Boyle and colleagues [28], in an epidemiological review of sleep-related disorders, argued that the prevalence rate can be

overestimated in some published literature. However, the effect of some health pertinent changes that can be correlated with the level of severity of ID and the co-occurrence with some health, psychological and behavioural disorders, can represent exceptions.

Generally, available studies often have design flaws such as a small sample size and selection bias. However, there is an urgent need for further research on the correlation between sleep disorder and the level of severity of ID [97].

26.6.2 Differential Diagnosis

In order to diagnose a sleep disorder in a patient, it is fundamental to know the individual's history and carry out an appropriate medical evaluation. Indeed, the changes in the physical and chemical processes involved in sleep in people with ID can lead to difficulties in the interpretation of polysomnography data because of the instability of the phasic shifts during sleep. Besides, difficulty in diagnosing sleep disorders can vary depending on the level of the ID, because of some people's limited ability to self-express and controlling for the confounding factors such as the higher prevalence of comorbid medical, neurological, and psychiatric disorders. These aspects should raise a high index of doubt and an efficient investigation for sleep disorders.

The two rating instruments to measure sleep disorders in the population with ID are the Stanford Sleepiness Scale [98] and the Sleep Disturbance Questionnaire [99, 100].

High rates of co-occurrence of primary sleep disorders have been reported to date, as well as a significant permeability in the boundaries between different subtypes of sleep disorders. Moreover, elevated levels of comorbidity with other neurodevelopmental, metabolic, genetic, medical, and neurological factors have been found in people with ID with sleep disorders. These data imply that the physiology and the reasons that lower the threshold for sleep disorders in people with ID need further investigation.

Subjects with ASD have several sleep disorders. These disorders can be exclusive to the neurophysiology of the ASD or also present problems for people with ASD, such as high rates of insomnia seem to be related to sleep hygiene and psychophysiological conditioning.

Finally, the high prevalence of psychiatric disorders, trauma, neurodegenerative disorders, and psychosocial stressors, makes it difficult to tease apart some primary sleep disorders from sleep disorders secondary to an underlying cause [101].

To diagnose sleep disorder in people with ID and/or ASD, it is necessary to analyse the available sleep related evidence, identify the link between sleep changes and daytime behaviours, review the medical history, and carry out appropriate medical evaluation.

Difficulties in diagnosing sleep disorders can vary depending on the level of the ID, because of some people's limited ability to self-express, and controlling for the confounding factors such as the higher prevalence of comorbid medical, neurological, and psychiatric disorders.

26.7 Functional Consequences

Sleep-phase shifts, restless legs syndrome, sleep-related breathing disorders, and parasomnias are related to insomnias. The restless legs syndrome is similar to the neuroleptic effect, akathisia; both of them can be difficult to detect in people with severe ID and problems in sleep initiation [68].

Besides, adults with ID can react to sleep deprivation with an increase in irritability, hyperactivity, aggression, and self-injurious behaviour (SIB), while the normal adult population feels fatigue or excessive daytime sleepiness [90, 102, 103]. Finally, it has to be considered that sleep in people with ID and ASD can also be susceptible to cyclical and seasonal changes, with a particular sensitivity to rapidly changing photoperiods and to sunlight on longer rhythms.

26.8 Treatment

Most studies on treatment strategies for sleep disturbances and sleep disorders have been carried out in children and younger adolescents. There is little research assessing sleep in very young children, older adolescents, or adults with developmental disabilities [32]. Indeed, there is limited evidence on intervention strategies for adults with ID experiencing sleep problems, and many studies have serious risk of bias [97]. Most research concentrates on the pharmacological treatment of sleep problems. However, current evidence primarily supports the use of non-pharmacological interventions [29]. Psychosocial intervention is the first-line treatment of sleep problems in the general population [104], with evidence of effectiveness. In studies of diverse ID, there is also strong evidence that behavioural and sleep hygiene measures should be the first line approach [105]. Sleep hygiene focuses on lifestyle and environmental factors such as reducing the consumption of caffeine, exercising regularly, avoiding daytime naps, and managing stress to promote sleep [106].

Most common cognitive-behavioural interventions include bedtime fading, extinction, distancing/desensitization, sleep-wake scheduling, multicomponent behaviour plan based on the function of sleep disrupting behaviour [29, 107, 108], and stimulus control, which aims to generate in a person's mind and body a strong association between going to bed and sleeping [109]. Other interventions that may be effective in the adult population include optimal scheduling of sleep, light therapy, and relaxation [97].

Furthermore, a combination of individualized behavioural therapy and medication has been proven effective [91, 110]. Educational training for staff supporting adults with ID has demonstrated improvements in sleep efficiency, declines in time in bed, and daytime napping [111]. A study by Gunning and Espie [109] assessed the effects of behavioural sleep management on adults with ID, finding that such management may improve behavioural sleep patterns or sleep-related functioning in the majority of adults with ID who are experiencing sleep problems.

Pharmacological treatments of sleep disorders include benzodiazepines, anticonvulsants, antidepressants, atypical antipsychotics, and agomelatine [33]. In general, psychopharmacological treatments are reported to be associated with longer sleep duration and better sleep efficiency [36].

Benzodiazepines act on γ-aminobutyric acid (GABA) neuroreceptors and have far less overdose danger and abuse potential than previous medications used for sleep disorders such as barbiturates [112]. Nevertheless, they can also cause some side effects, resulting in impaired sleep quality, residual sedation, drowsiness, dizziness, memory and cognitive impairment increased risk of falls, reduction of reflexes, or rebound insomnia. Paradoxical reactions to benzodiazepines are also possible, although relatively uncommon (less than 1% of patients), and may include increased talkativeness, emotional release, excitement, excessive movement, and aggression [113, 114]. Long-term benzodiazepine use may determine dose escalation, tolerance, dependence, and withdrawal issues. Physiological tolerance and dependency have both been linked to an increase in the negative effects of benzodiazepines [115].

The newer non-benzodiazepine agents zopiclone, zolpidem, and zaleplon have a hypnotic action comparable with that of benzodiazepines, but they display specific pharmacokinetic and pharmacodynamic properties. These three "Z" agents all share a short plasma half-life and limited duration of action as well as a selective interaction with omega (1) receptors (responsible for sedative effect), whereas benzodiazepines also interact with omega (2) receptors (responsible for some adverse effects such as memory and cognitive reduction) [116, 117].

Antipsychotics are usually prescribed for psychotic and mood comorbidities but also for the management of problem behaviours [118–120] and secondarily have a beneficial effect on sleep, although there are reports of specific prescription for sleep disturbances,

both in children and adults [121–123]. When used for the treatment of schizophrenia or bipolar disorder, the positive effects of atypical antipsychotic medications on sleep architecture are well documented [124–128]. Typical antipsychotics, including haloperidol, chlorpromazine, fluphenazine, and thioridazine, are associated with higher incidence of extrapyramidal side effects and daytime somnolence. Newer, second-generation antipsychotics, including clozapine, risperidone, olanzapine, quetiapine, and aripiprazole, have a lower propensity for extrapyramidal side effects and are generally less sedating (with the exception of clozapine, which can cause significant sedation, particularly during the initiation of treatment). Few studies examining the effect on sleep architecture have shown that slow wave sleep is increased by olanzapine, ziprasidone, and risperidone, whereas REM suppression is greatest for ziprasidone and risperidone [124, 126, 127, 129]. These agents are prescribed off label for the treatment of insomnia or the management of sedation, and are not recommended to be prescribed routinely for this indication, especially as a first-line pharmacotherapeutic agent [125, 130–132].

Limited data exist regarding use and efficacy of sedating antidepressants, selective serotonin reuptake inhibitors (SSRI), and tricyclic antidepressants (TCA) for treatment of sleep disturbances in people with ID/ASD. Sedating antidepressant such as mirtazapine and trazodone may be beneficial in case of co-occurrent depression [133–135]. In the general population the off-label use of trazodone for insomnia has surpassed its usage as an antidepressant [135]. The doses used for the treatment of insomnia are generally lower compared to those used for the treatment of mood disorders. Amongst TCAs, amitriptyline, imipramine, and doxepin are the most sedating and the most used for the treatment of insomnia in adults [136].

Agomelatine, an antidepressant analogue of melatonin and agonist of melatonergic MT1 and MT2 receptors, is reported to be effective and well tolerated for treating insomnia and sleep-wake alterations in adults with neurodevelopmental disorders [137]. Agomelatine produces both subjective and objective improvements in sleep quality and continuity, with increased slow-wave sleep and no effect on REM sleep [138].

Even anticonvulsants have little research on their use in the treatment of sleep disturbances in people with ID/ASD. The agents that have commonly been used in this category include valproate, lamotrigine, gabapentin, and carbamazepine [97].

In neurotypical persons with epilepsy, valproic acid has been shown to decrease REM activity and increase delta activity, although clear definitive evidence is lacking [139–142]. For lamotrigine, a minority of studies in epileptic patients indicate slight improvements in sleep duration and/or quality while many others show no effect on total sleep time, sleep efficiency, sleep latency or REM latency, and NREM duration [139, 143]. Lamotrigine has been reported to even cause insomnia, in a dose-dependent manner [144]. Gabapentin seems to shorten sleep latency, reduces awakenings and fast-wave sleep, enhances slow-wave sleep, and improves overall sleep quality and duration. Nevertheless, these effects have been reported in persons with sleep disorders secondary to various neurological or psychiatric conditions and effectiveness for primary and sleep disorders remains to be verified [141, 145]. Carbamazepine has been associated with decreased sleep fragmentation, increased delta NREM sleep, and increased total sleep time when used to treat bipolar disorders or epilepsy. Nevertheless, these effects have not been reported in other conditions and appear to be time-limited [141, 142].

Melatonin as both a dietary supplement and a drug (at a higher dose and/or prolonged release) has been shown to reduce sleep onset latency in primary insomnia and to regulate sleep-wake patterns in the neurotypical population. However, evidence is insufficient and controversial about long-term utility and safety as well as effects on sleep quality and duration, although slightly higher agreement exists on effectiveness in older people, who may be deficient in the hormone and present mitochondrial dysfunction [146, 147].

Specific studies, including open-label data, controlled trials, and meta-analysis, have been

conducted in people with ID/ASD, indicating melatonin to be effective on sleep onset latency, sleep duration, and sleep-wake alteration. Night awakenings have also been reported to improve, although some controversial findings, especially in persons with ASD [73, 97, 148, 149], who also showed higher rates of rather rapid loss (within a few weeks of treatment starting) of melatonin general efficacy, due to low genetically determined (single nucleotide polymorphism) metabolic activity of cytochrome P450 CYP1A2 [150]. Other differences in treatment response can be attributed to individual variations in receptor sensitivity [151]. Melatonin has also been used to treat severe sleep problems associated with various genetic syndromes such as Angelman syndrome, tuberous sclerosis complex, Rett syndrome, Smith-Magenis syndrome, and Sanfilippo syndrome [2, 152]. Except for tuberous sclerosis [153, 154], all other genetic syndromes reported significant decrease in sleep onset latency and other sleep improvements [155].

Most studies of melatonin involving people with ID and/or ASD have reported no adverse effects, although agitation, aggression, and restlessness have been described in a small number of cases, mostly when melatonin has been used in combination with other drugs [73, 148, 156–159].

The most used dosage in studies and clinical reports is between 1 and 2 mg in children/adolescents and between 1 and 5 mg in adults/elderlies, administered 30 minutes before bedtime [160–162]. Controlled-release formulations are likely to be more appropriate for those individuals who have issues with sleep maintenance and night awakes while immediate-release may be more appropriate for those with sleep onset delay [32, 162].

The implementation of evidence-based sleep intervention strategies can also help to reduce some of the short-term and long-term health and behavioural consequences of poor sleep [163–165].

Excessive daytime sleepiness can benefit from behavioural treatments aimed at extending total time asleep and supporting sleep onset. Medication to support sleep can also be considered [33]. Excessive daytime sleepiness associated with narcolepsy, idiopathic hypersomnia, and other forms of hypersomnias can be treated with amphetamine-like central nervous system stimulants or modafinil and its R-enantiomer, armodafinil, and wake-promoting compounds unrelated to amphetamines. Methylphenidate is the most commonly prescribed amphetamine-like stimulant and is reported to be quite efficacious and well tolerated by most narcoleptic patients. For excessive daytime sleepiness associated with narcolepsy, modafinil is considered a first-line treatment, also due to its safety and low side-effect profile. These compounds, however, lack efficacy for cataplexy and dissociated manifestations of REM sleep, so antidepressants, especially selective norepinephrine, and serotonin reuptake inhibitors are additionally prescribed to treat these issues. Sodium oxybate, a salt of gamma-hydroxybutyrate (GHB), is indicated as the most effective drug for the treatment of cataplexy, but its prescription status is limited in many countries across the world since the high risk of serious side effects, craving, and physical dependence [166, 167].

The treatment of obstructive sleep apnoea-hypopnea syndrome includes weight loss and exercise, positive airway pressure, continuous electrical stimulation, and surgical enlargement of the upper airway. Continuous positive airway pressure (CPAP) is the standard treatment for moderate to severe OSAS and can be selectively used also for mild OSAS. For persons with intolerance to CPAP, bilevel positive airway pressure (BPAP) can represent a valid alternative since the lower pressure is exerted during exhalation. Other alternative treatments are represented by positional therapy, mandibular advancement devices, continuous transcutaneous electrical stimulation, and hypoglossal nerve stimulation. Surgical treatments include nasal surgery, oral/palate surgery, hypopharyngeal surgery, and other operations, such as orthognathic surgery, tracheostomy, and multi-lateral surgery. There are currently no effective pharmacological therapies, although corticosteroids and leukotriene receptor antagonists have been reported to have some efficacy on mild OSAS in children [168, 169].

There is little high-level evidence for the treatment of parasomnias, especially in adults [170]. Priorities include avoiding potential triggers such as scary movies, caffeine, alcohol, or late-night meals, as well as maintaining a consistent and healthy sleep-wake pattern. It is also critical to adopt behavioural techniques and environmental measures to protect the patient, bedmate, nearby youngsters, and other fragile persons, such as closing windows, bolting doors, removing any sharp objects or furniture near the bed area, and sleeping on the bottom floor [108, 170–172]. Cognitive-behavioural therapy seems to be effective for most parasomnias and further progress is expected in the near future [172]. In particular, schedule awakening, a behavioural intervention based on the involvement of briefly awakening the patient approximately 15–30 minutes prior to the expected episode, has been reported to provide good results in sleepwalking, sleep terrors, and confusional arousal, while nightmare disorder seems to benefit more from imagery rehearsal therapy, exposure relaxation and rescription therapy, imagery rescripting and exposure therapy, self-exposure, lucid dreaming treatment, and eye movement desensitization reprocessing therapy [172].

Benzodiazepines, especially clonazepam, are the mainstay of pharmacological management for most persisting parasomnia, although imipramine, pramipexole, levodopa, and melatonin have also been used in the past with some efficacy [108, 170, 171, 173, 174]. Clonazepam has to be used with caution in patients with dementia, disorders of gait or balance, or concomitant OSAS [108].

Restless leg syndrome can benefit from dopaminergic therapies such as pramipexole, ropinirole, and rotigotine, both for sleep disturbance and daytime symptoms. Alternative treatments include α2δ ligands, opioids, and iron preparations, although no iron preparations have received regulatory approval. Oxycodone and naloxone are indicated for severe restless legs but, due to the commonly occurring adverse effects, they should be used exclusively as second-line therapy [175].

Nightmare disorder and other parasomnias have been reported to be triggered or worsened by many psychoactive compounds, including SSRIs (especially paroxetine), cholinesterase inhibitors, beta-blockers, levodopa, lamotrigine, and following withdrawal from antidepressants [108].

Current evidence primary supports the use of a combination of individualized behavioural (including sleep hygiene) and pharmacological therapies to treat sleep disorders in persons with ID and/or ASD. For insomnia, pharmacological treatments include benzodiazepines, anticonvulsants, antidepressants, atypical antipsychotics, and agomelatine. Melatonin has also been shown to have some efficacy.

Tip

Sleep disorders are a quite common comorbidity in people with ID, and their presence further compromises physical, cognitive, and behavioural functioning. New models of testing, using new technology, will allow researchers and clinicians to collect precise data on sleep while patients go about their daily lives in their normal environment.

Key Points

- Aetiology and pathogenesis of sleep disorders in ID is multifactorial. There are biological, genetic, and psychosocial risk factors for developmental of sleep abnormalities.
- There are several comorbid conditions which are associated with sleep disorders in ID. One must be able to evaluate comorbid disorders to make a differential diagnosis.
- Sleep disorders are important because they may affect the person's quality of life, lead to subsequent problem behaviours, and cause both distress and impaired functioning during the daytime.
- The diagnosis of sleep abnormalities in persons with ID is complex due to the

limited ability of people with ID to report their own sleep disorders. Besides, the changes in the physical and chemical processes of sleep in people with ID can lead to difficulties in the interpretation of polysomnography because of the instability of the phasic shifts during sleep.

- The diagnosis could be facilitated by collecting sleep history and identifying the link between sleep changes and daytime behaviours.
- Most effective treatment is based on a combination of individualized behavioural and pharmacological therapies. Benzodiazepines, anticonvulsants, antidepressants, atypical antipsychotics, and agomelatine represent the most studied drugs.
- Effects of exogenous melatonin have been widely investigated, with indications of effectiveness on sleep onset latency, sleep duration, and sleep-wake alteration.

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Sexual Dysfunctions

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Contents

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Learning Objectives

- To learn about the definition of sexual health and sexuality.
- To learn about the definition and ascertainment of sexual dysfunctions in people with ID and/or ASD.
- To know how much the issue around sexual health in the ID and or ASD population can be underestimated.
- To learn the peculiar presentation of sexual dysfunction in persons with ID/ASD and how to diagnose them.
- To know the course and treatment of the sexual health issues in people with ID and/or ASD.

27.1 Introduction

Several definitions of sexual health have been provided across time, the most recent of which have built upon the essential elements provided in the original definition of the World Health Organization (WHO) [1] but have added concepts of mental health, responsibility, and sexual rights [2].

According to the current working definition of the WHO, sexual health is "…a state of physical, emotional, mental and social well-being in relation to sexuality; it is not merely the absence of disease, dysfunction or infirmity. Sexual health requires a positive and respectful approach to sexuality and sexual relationships, as well as the possibility of having pleasurable and safe sexual experiences, free of coercion, discrimination and violence. For sexual health to be attained and maintained, the sexual rights of all persons must be respected, protected and fulfilled" [3, 4].

The WHO specifies that sexual health cannot be understood and made operational without a broad consideration of sexuality, which is defined as "…a central aspect of being human throughout life" and "encompasses sex, gender identities and roles, sexual orientation, eroticism, pleasure, intimacy and reproduction". "Sexuality is experienced and expressed in thoughts, fantasies, desires, beliefs, attitudes, values, behaviours, practices, roles and relationships", and "is influenced by the interaction of biological, psychological, social, economic, political, cultural, legal, historical, religious and spiritual factors" [3, 4].

The fulfilment of sexual health is tied to the extent to which sexual rights are respected, protected, and complied. In their position statement on the sexuality of persons with intellectual disabilities and developmental disabilities, the American Association on Intellectual and Developmental Disabilities (AAIDD) and The Arc [5] denounce that for a long-time persons with intellectual disability (ID) "have been thought to be asexual, having no need for loving and fulfilling relationships with others. Individual rights to sexuality, which is essential to human health and well-being, have been denied. This loss has negatively affected people with ID in gender identity, friendships, self-esteem, body image and awareness, emotional growth, and social behaviour. People with ID frequently lack access to appropriate sex education in schools and other settings. At the same time, some individuals may engage in sexual activity as a result of poor options, manipulation, loneliness or physical force rather than as an expression of their sexuality". The same AAIDD and The Arc, in alignment with the United Nations Convention on the Rights of Persons with Disabilities [6] and the WHO framework for an operational approach to sexual health [7], state that the presence of ID, regardless of severity, does not justify the loss of rights related to sexual health and sexuality, which include safe and pleasurable experiences, equality, privacy, marriage, choice around reproduction, information or education, opinion, and expression [5].

The aforementioned framework for an operational approach to sexual health that has been recently produced by the WHO in partnership with the Gender and Rights Advisory Panel (GAP) of the UNDP/UNFPA/UNICEF/WHO/World Bank Special Programme of Research and the Development and Research Training in Human Reproduction (HRP) identifies six crucial, cross-cutting principles to be incorporated into the design of all sexual health (and reproductive health) interventions and which can also serve as evaluation criteria against

which these interventions should be assessed. These principles are the following: (1) holistic approach to sexual health; (2) linked nature of sexual health and reproductive health; (3) respect, protection, and fulfilment of human rights; (4) multilevel influences on sexual health; (5) diversity of needs across life course and populations; and (6) evidence-based, respectful and positive approach. The fifth principle has particular relevance for persons with intellectual and developmental disabilities since it states that "sexual health exists on a dynamic continuum, with needs that …vary depending on a complex mix of individual characteristics, as well as the cultural, socioeconomic, geopolitical and legal environment. Particular combinations of these factors can create vulnerabilities … that may increase susceptibility to ill health and/or hinder access to health care. For example, certain sexual health interventions may not be available because they are not culturally acceptable or legally permissible; available sexual health interventions may be difficult to access for a range of reasons; and the sexual health needs of some individuals, populations or age groups may not be recognized or acknowledged. Sexual health programming and research must therefore be inclusive of the diversity of needs among individuals at various points across the life course and in various settings or circumstances" [7].

In the last decades, the interest in the sexual health and sexuality of persons with ID has increased. Nevertheless, there has been a tendency to focus on sexual rights and societal attitudes, while less attention has been paid to sexual health [8, 9].

Very little information is available on sexual problems in people with ID, but it is known that they have more vulnerability factors than the general population to mental and physical ill-health, including sexual dysfunctions. Furthermore, some genetic syndromes including ID and/or autism spectrum disorder (ASD) also include fertility issues [10, 11].

Sexual dysfunctions are a set of clinical conditions characterised, in both genders, by the significant difficulty to respond sexually or to experience sexual pleasure, that regard different behavioural aspects, including the psycho-physiological functioning profile. With the exception of substance/medication-induced sexual dysfunctions, they must be present for at least 6 months and be associated with individual distress [12]. The lack of sexual education, low-grade support systems, negative social attitudes, mental and physical health problems, life events, and medications seem to represent significant aetiological factors for the development of sexual dysfunctions in people with ID and/or ASD [13].

27.2 Prevalence

The epidemiological studies on sexual dysfunctions in ID are very limited [13]. There are different reasons that can account for this finding such as the lack of interest in this comorbidity, the difficulty in assessing the sexual functioning in this population, and the methodological difficulties in conducting research in this area.

Expert opinion reports and interesting summaries are available to date, but no systematic reviews of the literature [14–19].

The only prevalence data available indicate that 35% of men with severe to borderline ID and epilepsy had never experienced spontaneous erections while awake, 44% had never had an orgasm, and 21% had never had intercourse [15]. Furthermore, it is possible to forecast that some sexual dysfunctions could accentuate with ageing.

People with ID and/or ASD have more vulnerability factors than the general population to develop specific comorbidities, including sexual dysfunctions. Nevertheless, scientific evidence on these health issues is limited.

27.3 Aetiopathogenesis

The aetiology of sexual dysfunctions in persons with ID/ASD is probably similar to that of the general population, and it comprises biological, genetic, psychosocial, developmental, and cultural factors and various life events.

27.3.1 Biological Factors

One or more psychotropic medications are prescribed to the wide majority of persons with ID/ASD across their life span, especially in order to prevent and treat problem behaviours. These treatments include antipsychotics, antidepressants, mood stabilisers, and anxiolytics [20, 21], most of which can cause sexual dysfunction even though some of the most recent antipsychotics are found to have less impact on sexual functioning. Moreover, some people with ID/ASD are administered androgen-depleting drugs as a consequence of inappropriate sexual behaviour [22]. For the high prevalence of many physical health issues, persons with ID/ASD also receive many other non-psychopharmacological treatments and are often exposed to polypharmacy [23, 24]. Because of this high rate of use of psychotropic and not psychotropic drugs, persons with ID/ASD are more likely to develop a substance/medication-induced sexual and endocrine dysfunction, including erectile dysfunction, ejaculation disorder, and priapism in males and vaginismus, inadequate lubrication, anorgasmia, dyspareunia, and menstrual disorder in women, along with hyperprolactinaemia, weight gain, and movement disorders in both genders.

In addition, a higher rate of psychiatric disorders is found in people with epilepsy, and epilepsy is common in people with ID/ASD [25]. Anti-epileptic drugs (AEDs) are also used as mood stabilisers in people with ID/ASD. Seizures and AEDs can affect hypothalamic-pituitary-gonadal axis, causing changes in hormones production and, thus, sexuality [26, 27].

Furthermore, these people might encounter decreased sexual activities, listlessness, and depression as a consequence of the sedative effects of the AEDs. Besides, AEDs are also linked to hypo-sexuality, abnormal morphology, low sperm count and androgen, and elevated oestrogen, testosterone, and sex hormone-binding globulin [26].

Other neurological conditions can be linked to sexual dysfunctions in people with ID/ASD. Parkinson's disease may be responsible for affect arousal, orgasm, and sexual desire in women and erectile dysfunction, sexual dissatisfaction, and premature ejaculation in men [19].

In multiple sclerosis, the sexual dysfunctions may be due to pain or physical fatigue or movement disorders or the body self-image. Similar problems are caused by spinal cord injuries [19].

27.3.2 Genetic Factors

People with ID tend to have genetic syndromes that can be associated with sexual dysfunction. Downs syndrome, for example, is related to hypogonadism. The Prader-Willi syndrome is linked to small penis-size, hypogonadism in males, and delayed puberty in females [28]. The Klinefelter syndrome, which is characterised by having 47 XXY or more than one X chromosome in males, is related to hypergonadotropic hypogonadism, reduced body hair, small testes, decreased libido, and increased breast tissue [29]. Turner syndrome (45 X) is linked to hypogonadism, delayed puberty, amenorrhoea, lack of breast tissue, and infertility [30]. Finally, Fragile X syndrome is related to macroorchidism and premature menopause [31].

27.3.3 Psychosocial Factors

Services can influence sexuality in various ways. Most of them are restrictive, aversive, and do not help people with ID and/or ASD in their free expression or privacy. Indeed, there is a general fear that talking about sexuality could lead them to abusive conducts or to sexually offensive behaviours [32]. Besides, there is less support for lesbian, gay, bisexual, and transgender (LGBT)-related issues [33] or masturbation. Because people with ID have limited privacy, education, economic independence, social skills, and environmental mastery, they are at higher risk for abuse [34, 35], more exposed to an unplanned pregnancy [36] and sexually transmitted disease, and for some because of lack of communication skills, they are unable to express their experiences, needs, and desires. Given appropriate opportunities and support, many people with

ID and/or ASD are likely to develop an intimate relationship and sexual interactions, but the misconceptions about sexuality in the ID/ASD population still represent social barriers for them to develop such relationships.

27.3.4 Developmental Factors Through the Lifespan

The social, emotional, and psychosexual development of people with ID and/or ASD often do not synchronise with that of the general population. People with ID and/or low-functioning ASD may not have age-appropriate sexual knowledge and often be unable to determine whether they have sexual dysfunctions [25, 37, 38]. Indeed, people with ID would need support from their family or carers with their understanding of the sexual knowledge as well as finding potential partners and spending time alone with them. On the contrary, in the past, people with ID often were desexualised and prevented from having sexual intercourse by punishing them and suppressing that behaviour. Even today, many adults with ID/ASD are still likely to be viewed as childlike and asexual by their families and service workers [39].

27.4 Criteria and Clinical Features

The assessment of sexual dysfunctions in people with ID and/or ASD presents many problems. First of all, the inability of these people to recognise and communicate their own health and sexual needs, thus failing to ask for help leading to appropriate diagnosis and necessary treatment [40]. The social stigma and the attitude of some familiars and caregivers may hinder the sexual experience, the sexual identity development, and the identification of dysfunctions [14].

Furthermore, in this population, although the physical growth may follow the normal stages, the development of cognitive, emotional, and social aspects is generally atypical and asynchronous.

The clinicians need to be aware to include important issues in assessing sexual dysfunctions in ID and/or ASD. These are, for example, societal attitudes and sexual education of people with ID/ASD; the influence of family and care-staff in the expression of individual sexual needs and behaviours; the frequent use of medications; the presence of many mental health problems, genetic syndromes, and neurological disorders; and the lack of opportunity for the person to experience their own sexuality and to share with others.

In relation to clinical issues, it is very difficult to determine the criterion of distress, which is very important for making a diagnosis. In fact, in this population it is difficult to establish how the distress is experienced and shown. Indeed, often the distress could be hidden or expressed with unspecific irritability.

Therefore, a longer and deeper assessment is required in people with ID to explore their self-knowledge of sexuality and sexual needs, their gender identity and sexual orientation, and their medication history [33, 41, 42].

The diagnosis of sexual dysfunction in persons with ID/ASD is very complex due to the developmental, educational, and functional peculiarities and the lack of a specific diagnostic methodology.

▪ Delayed Ejaculation

This disorder is characterised by the inability or considerable delay in reaching ejaculation during sexual intercourse with the partner for at least 6 months. The exact prevalence of this disorder in the ID population is unknown. Indeed, it increases with age and the relation between delayed ejaculation and medical causes still needs to be investigated in-depth, such as the role of medications and substances. Moreover, the psychological factors related to delayed ejaculation also need further investigation to understand if there are specific circumstances that can promote healthy ejaculation.

▪ Erectile Disorder

This disorder is characterised by experiencing at least one of three following symptoms for no less than 6 months: (a) having difficulty with achieving or (b) maintaining erections

during sexual activity and (c) a considerable decrease in erectile rigidity.

As for many other sexual dysfunctions, the prevalence of this disorder is unknown, although there are reports of an increase with age [4]. However, there is evidence that erectile disorder is related to having sexual intercourse with an unknown partner, substance abuse, and peer pressure. The erectile disorder can also be associated with medical conditions and comorbid mental illnesses [43–45].

Female Orgasmic Disorder

The female orgasmic disorder is characterised by the absence, considerable delay, infrequency, or reduced intensity of the orgasmic sensations. The symptoms must be present for at least 6 months in order to meet the diagnostic criteria. The prevalence of the female orgasmic disorder in the general population is 10–42% [46]. However, these data do not take into account distress as a causative factor leading to fewer women reaching the diagnostic criteria. Besides, it has been calculated that 10% of women in the general population never reach an orgasm in their lifetime [47]. The female orgasmic disorder may be related to psychological factors such as feelings of inadequacy or guilt as well as relationship problems and cultural factors. Moreover, there is a link between female orgasmic disorder and vulvovaginal atrophy and mental health problems, especially anxiety and depressive disorders [48, 49].

Female Sexual Interest/Arousal Disorder

The disorder is characterised by having at least three of the following criteria for at least 6 months: (a) a decline or absence of interest in sexual activities; (b) a reduction or lack of sexual thoughts and fantasy; (c) a reduction or loss of desire to take the initiative for the sexual intercourse or a negative response to the partner's initiative; (d) a decline or loss of sexual pleasure or excitement during sexual activities; (e) a decline or absence in sexual desire or arousal to stimuli that are related to sexual activities; and (f) a reduction or lack of sensations during the sexual intercourse. The prevalence of this disorder in the ID population is still unknown. Moreover, this disorder can be associated with depression, anxiety, urinary incontinence, thyroid problems, arthritis, and irritable bowel syndrome [49–51].

Genito-pelvic Pain/Penetration Disorder

The disorder consists of having difficulties with vaginal penetration, pain in the vulvovaginal or pelvic area when trying to penetrate or during the sexual act, and fear and anxiety that is related to the pain and tense pelvic floor muscles upon attempted penetration, for no less than 6 months. Besides, while the diagnosis can be made based on one symptom dimension, all four aspects should be assessed. The peculiarity of this disorder is that it can take place with a partner (during sexual intercourse) or even during medical examinations or while inserting a tampon. The prevalence of this disorder is unknown, but in North America, nearly 15% of women in the general population noted recurrent pain during the sexual act. The genito-pelvic pain/penetration disorder can occur along with other sexual dysfunctions leading to the exclusion of sexual activities or intimate situations. However, both physical and sexual abuse and cultural factors have been considered predictors of this disorder, as evidence leads to believe that there is possibly a higher prevalence in non-Western countries [52, 53]. The disorder can be also anticipated by vaginal infection, which can persist even when the symptoms are treated. Although the DSM-5 [12] diagnoses it only in women, there are shreds of evidence that men can feel genito-pelvic pain as well [54]. A common cause in men is represented by pelvic floor overactivity, which is encountered in up to 50% of chronic prostatitis and/or chronic pelvic pain syndrome and characterised by persistent muscle pain that can also affect the bowel and bladder [55].

Male Hypoactive Sexual Desire Disorder

The prevalence of this disorder is still unknown, and it is characterised by a decline or absence of sexual fantasy or desire for sexual intercourse for a minimum of 6 months. However, these difficulties can be preferential or associated with other sexual dysfunctions such as difficulties in erection or ejaculation.

Besides, as for previous sexual disorders, there is a relation between age and fantasy or desire. Moreover, it is related to mental health problems, such as depression, anxiety, and alcohol abuse. Endocrinologic factors, such as hyperprolactinaemia and hypogonadism, should also be considered as aetiologies [45, 56].

Premature (Early) Ejaculation

Premature ejaculation is characterised by ejaculation within 1 minute of penetration. This fact is consistent and undesired and has to be present for at least 6 months. It seems to be only applicable for vaginal penetration, as there are not specific duration criteria indicated for the manual stimulation, oral and anal sex, and other activities. Nevertheless, the DSM-5 [12] states that this diagnosis can be applied to individuals who engage in non-vaginal sexual activities. Based on two separate observational, cross-sectional surveys the overall prevalence of the of premature ejaculation is founded to be 19.8% and 25.80% with a lifelong rates of 2.3% and 3.2% [57]. Besides, there could be a correlation among anxiety disorder, self-esteem, and premature ejaculation, while cultural and religious factors still need to be assessed [46, 58].

Substance-/Medication-Induced Sexual Dysfunction

Because of the lack of reports on treatment-emergent sexual side effects, the prevalence of these dysfunctions in persons with ID and/or ASD is unclear. A relation between psychotropic medications, such as antidepressants, opioids, and antipsychotics, and sexual dysfunction has been reported [5, 45, 56].

The disorder is characterised by problems in sexual activity that are due to substance or medication. The problem can appear before or after the substance misuse, after stopping the substance or following the introduction of medications.

27.5 Treatment

Treatment of sexual dysfunctions must take into account underlying problems or problems that are causing the dysfunction. If the cause is physical, medication and mechanical aids should be considered, while counselling, psychotherapy, behaviour modification, and education may work in presence of prevalently psychological causes [59].

When a medication is the cause of the dysfunction, a change in the medication may help. Hormone therapy, both local and systemic, can be considered for men and women with hormone deficiencies. In men, sexual function may be improved with phosphodiesterase-5 inhibitors such as sildenafil, tadalafil, vardenafil, and avanafil by increasing blood flow to the penis [60, 61]. For women, hormonal options such as oestrogen and testosterone can be used, although specific approval is lacking [62]. In premenopausal women, flibanserin and bremelanotide have been used (and approved by some agencies) to treat low desire [62].

A good quality health and sex education presented in an accessible format describing body parts and relationship using pictures, role-play, guided practice, corrective feedback, modelling, or reinforcement is a good start in addressing the problem of sexual disorders in adults with ID [63, 64].

People with ID should see a sex therapist in order to present the concerns that are related to their particular disability. However, most of the readings and written homework that is an essential/intrinsic part of sex therapy are complicated/problematic/challenging to these people because of their intellectual impairments. Moreover, the therapist should be cautious in the use of body-awareness and touching exercises that can be seen as humiliating by these people that have in general a negative self-image. It is also essential, for those individuals who are not able to have a sexual intercourse, to pay attention to masturbation, as it may offer an outlet and a way to experience sexuality [65]. Once a person with ID or a couple finds their way into the sex therapist's office, he/she or they can present treatment concerns unique to their particular disabilities. Many couples who were referred to this kind of therapy were experiencing some level of relational or family issues, exacerbated by poor communication. In this case, the interplay between the relational and sex-

ual difficulties requires that the focus of treatment should include working with issues of intimacy and communication as well as with the specific sexual dysfunctions [18].

Since professional caregivers may play a key role in the development and management of sexual dysfunctions in persons with ID and/or ASD, it can be useful for those of them who have difficulties in approaching sexuality and sexual issues of their clients with ID/ASD, to receive specific training on these topics [33, 66]. Chou and collaborators found that workers of a service for people with ID who participated in an intervention programme aimed at fostering positive attitudes towards the sexual identities, health, and rights of people with ID changed their perception of sexual behaviours of their clients with ID from problematic to normal. Parents, who participated in the same study, were also found to shift their approach to the sexual needs of their children with ID from prohibition to communication and support [67].

27.6 Course and Prognosis

The lack of sexual knowledge and the absence of appropriate sexual models with which to compare people with ID and the attitudes of carers can delay or hinder the recognition of problems. It is also probable that people with cognitive, communicative, and relational impairment have limited skills to understand and express needs, feelings, and difficulties as well as behavioural issues related to sexual life [65]. Furthermore, people with ID may have fewer interpersonal experiences and present various problems regarding finding, entering, and maintaining sexual friendships and relationships [68].

In the meantime, people may develop a childlike self-image or compensate for their sexual problems and dissatisfaction developing psychological problems and challenging or stereotypical or compensative behaviours. Typically, it's possible to note aggression, criminal acts, compulsive masturbation, exhibitionism, sexual victimisation, etc. [13].

Accordingly, the prevalent sexual activity in this group of ID is masturbation, but in most cases the person needs support to ensure proper execution of this act. In fact, the frequent problem in people with severe ID is excessive masturbation or unsuitable behaviours towards others, often in an inappropriate context.

Finally, persons with ID who are also LGBT sexual may have much more difficulties establishing intimate and sexual relationships with others [33, 42].

Educational intervention should be very useful in promoting knowledge of the parts of one's body and their functions, pubertal changes, and interpersonal, relational, and life context adequacy as well as acquiring personal hygiene skills and self-protective behaviours.

This chapter does not consider issues related to the sexuality of persons with ID/ASD that have not been included in the group of sexual dysfunctions and disorders as defined by the DSM-5 or the ICD-11. Nevertheless, readers have to be aware that some of these untreated issues such as masturbation in public, touching others inappropriately, or sexual offending are frequently encountered in persons with ID/ASD and would deserve specific coverage. An emerging area of interest is related to the psychological implications of having a homosexual relationship because of the lack of opportunity for a heterosexual relationship.

> The diagnosis of sexual dysfunctions becomes possible later in the life of persons with ID compared with the general population. During the developmental period, people may develop a distorted self-image or compensate for the sexual dissatisfaction developing psychological and behavioural problems.
>
> Psychotherapy and psychoeducation can help these people to improve their sexual health and sexuality.

Tip

Sexual health is an important element in the functioning of people with ID. The scientific community has long neglected the study of sexuality in these persons, probably due to the cultural attitude and the clinical difficulty posed by the condition of ID itself. Early diagnosis and treatment of sexual dysfunctions can promote quality of life and discourage other physical, psychological, and relationship problems.

Key Points

- Data on the prevalence of sexual dysfunctions in persons with ID and/or low-functioning ASD are lacking.
- Aetiopathogenesis of sexual dysfunctions in ID/ASD seems to be multifactorial, although knowledge is limited.
- Assessment of sexual dysfunctions in persons with ID/ASD is complex, for different reasons, ranging from individual communication deficits to social stigma.
- Diagnostic criteria of sexual dysfunction in persons with ID/ASD are the same as those used for the general population. There is not available specific adaptation.
- Treatment should be approached at a multidimensional level, with early affective-sexual education having a significant role. Programmes to develop specific skills in relation to specific sexual dysfunction may be particularly effective.

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Dementia in People with Intellectual Disabilities

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Contents

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Learning Objectives

- Rate of dementia in people with intellectual disabilities.
- Rate of dementia in people with Down syndrome.
- Neuropathology of dementia in intellectual disabilities.
- Clinical features of dementia in people with intellectual disabilities.
- Assessment of dementia in intellectual disabilities.
- Differential diagnosis of dementia in intellectual disabilities.
- Investigations for dementia in intellectual disabilities.
- Rating scales for dementia in intellectual disabilities.
- Pharmacological treatment of dementia in intellectual disabilities.
- Non-pharmacological intervention for dementia in intellectual disabilities.
- Service needs for people with intellectual disabilities and dementia.

- The rate of dementia is higher among people with intellectual disabilities than the general population who do not have intellectual disabilities.
- The rate (particularly of Alzheimer's disease) is even higher among people with Down syndrome with an earlier age of onset compared with the general population and people with intellectual disabilities who do not have Down syndrome.
- Early diagnosis of dementia in people with intellectual disabilities may be difficult, and a combination of informant-rated instrument and direct examination of the person with intellectual disabilities is desirable.
- Loss of memory of recent events in the context of intact distant memory which is common in early-stage dementia among people who do not have intellectual disabilities seems to be the early manifestation of dementia in people with mild to moderate intellectual disabilities.
- Skill loss, on the other hand, seems to be an early sign of dementia in people with severe intellectual disabilities.
- Features of frontal lobe dysfunction including apathy and other behavioural changes seem to be common in early stages of dementia diagnosis in people with intellectual disabilities.
- Regular yearly assessment from an early age (30–35 years) is recommended to compare disease onset with a pre-determined baseline.
- Screening instruments used for the general population are often not useful in people with intellectual disabilities.
- In the absence of any convincing randomised controlled trial (RCT)-based evidence in intellectual disabilities, the treatment recommendations for dementia in people with intellectual disabilities should be based on evidence based on that for the general population who do not have intellectual disabilities.

Tip

- Dementia is more prevalent in people with intellectual disabilities and more so in people with Down syndrome who are affected early in their lives.
- Dementia at its early stage may be difficult to diagnose in people with intellectual disabilities.

28.1 Introduction

Life expectancy of people with intellectual disabilities (ID), perhaps with exception of those with profound ID, has increased considerably in the last five decades [1]. Although this is very good news this also means that people with ID are exposed more to the risks of developing medical conditions that affect people in older age such as neurodegenerative disorders including Alzheimer's disease (AD) and other dementias. In this chapter,

we discuss dementia in people with ID in general and Down syndrome (DS) in particular.

28.1.1 Dementia in Persons with Intellectual Disabilities Who Do Not Have Down Syndrome

Studies of dementia in people with non-DS ID have reported an increased prevalence of dementia compared with the non-ID general population. Cooper reported dementia in 21.6% of 134 adults with ID aged 65 years and above [2]. In a cross-sectional study involving adults with non-DS ID (aged >60 years), AD was found to be the most common subtype (prevalence of 8.6%), followed by Lewy body, frontotemporal and vascular dementia [3]. Strydom and colleagues found an increased prevalence (18.3%) of dementia in 284 adults (≥65 years) with non-DS ID. This prevalence was not affected by the severity of ID [4]. The study confirmed a downward shift in age-associated risk, with an earlier age of dementia onset in this population compared with the general population. In contrast, Zigman and colleagues reported the prevalence of AD in 2.7% of adults with the non-DS ID of age 65 and over, and 4.1% in adults aged 75 years and older [5]. These rates are comparable with or lower than the expected rates in the general population. Patel and colleagues found a prevalence of dementia of 11.4% among 105 adults with ID aged 50 years and older [6]. The incidence rates were also higher compared with the general population [7]. Between 31% and 78% of non-DS adults with ID show AD neuropathology, whereas almost all people with DS over age 50 are known to develop AD neuropathology [8].

Autopsy examinations on 60 brains of individuals with ID of whom 15 had DS and 29 were aged 40 years and over showed a variety of acquired neuropathological changes, the most common being AD pathology with or without cerebrovascular disease (CVBD) (n = 8) followed by CVBD alone (n = 6) and Parkinson disease (n = 3) [8]. Of note, four brains showed neurofibrillary tangles (NFTs) confined to entorhinal cortex, compatible with the concept of primary age-related tauopathy (see ◘ Fig. 28.1).

A number of non-DS ID genetic syndromes are shown to be associated with increased prevalence of dementia [1]. These include Fragile X-associated tremor/ataxia syndrome (FXTAS), Cockayne syndrome (CS), Williams syndrome (WS), Prader-Willi syndrome (PWS), mucopolysaccharidosis type III (MPS III), Angelman syndrome (AS), and untreated phenylketonuria (PKU). It is not always clear whether this clinical presentation is due to premature neurodegeneration associated with specific syndromes as dementia neuropathology could not always be confirmed.

The CGG repeat size has been shown to be an independent risk factor for the FXTAS dementia [9]. Neuropathological characteristics are heterogeneous ranging from AD pathology to Lewy bodies in cortical and brainstem dopaminergic neurons, white matter disease, and astrocytic pathology [10].

In mildly affected people with CS, the course is slower, resulting in survival into adulthood with the development of dementia. Neuropathological examination discloses profound microencephaly, basal ganglia calcification, white-matter atrophy due to severe patchy myelin loss (tigroid leukodystrophy) with string vessels (i.e. thin connective tissue strands, remnants of capillaries, with no endothelial cells and no blood flow), and severe CVBD [11].

A few people with WS and premature 'senile dementia' have been reported, but neuropathological post-mortem examinations are lacking. However, people with WS may be considered to be at risk for developing both AD and vascular dementia, due to the high prevalence of hypertension and a high incidence rate of glucose intolerance or type 2 diabetes mellitus [12].

Also, in people with PWS, some cases of early-onset dementia have been reported but, again, without autopsy confirmation [13]. Maternal uniparental disomy (mUPD) with female sex and a history of psychosis appear to increase the risk for dementia [14].

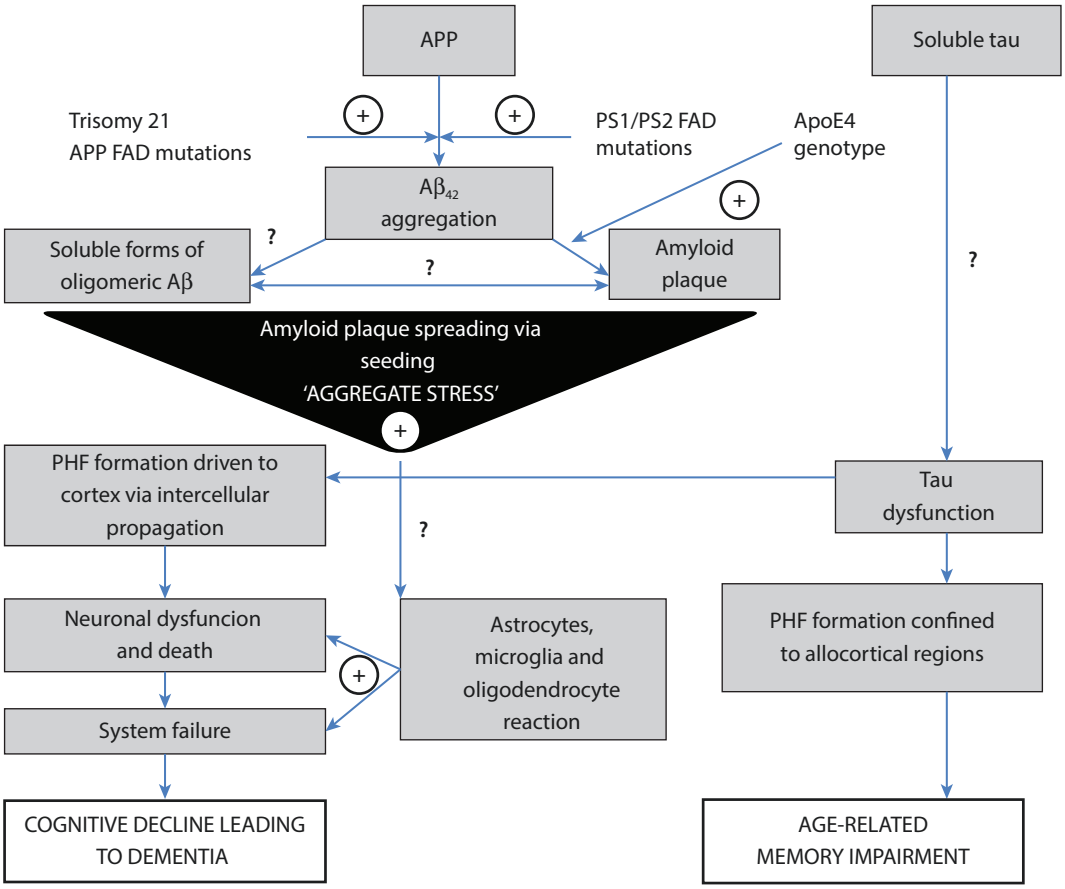


Fig. 28.1 Amyloid Cascade Hypothesis: current state of knowledge. APP amyloid precursor protein, FAD familial Alzheimer 's disease, PS presenilin, ApoE4 apolipoprotein epsilon4, Aβ amyloid beta, Tau tubulin-associated unit. (Modified from Karran & De Strooper (2016) [84])

MPS IIIB, also known as Sanfilippo syndrome, although associated with premature death, a number of less severe cases have been described to be living into late adulthood (age 30s to 60s) [15]. To the best of our knowledge, there is only one case report of an early-onset dementia in a female patient with MPS IIIB with a history of autistic-like features [16].

AS, albeit being a severe form of ID, may be compatible with long life (72–75 years) [1, 17]. There are, however, no published case reports of AS with dementia apart from a report on a 75-year-old male with a reduced cerebral circulation compatible with 'organic dementia' [17].

Untreated PKU although is typically associated with severe neurological dysfunction beginning in early childhood; there is one case report of a 57-year-old woman with mild ID who manifested four years of progressive spastic paraparesis and dementia with only partial (motor) improvement after protein-restricted diet [18].

Although there is an improved life expectancy in individuals with Rubinstein-Taybi syndrome and Cornelia de Lange syndrome [1], no case report of dementia could be found in the literature involving these syndromes.

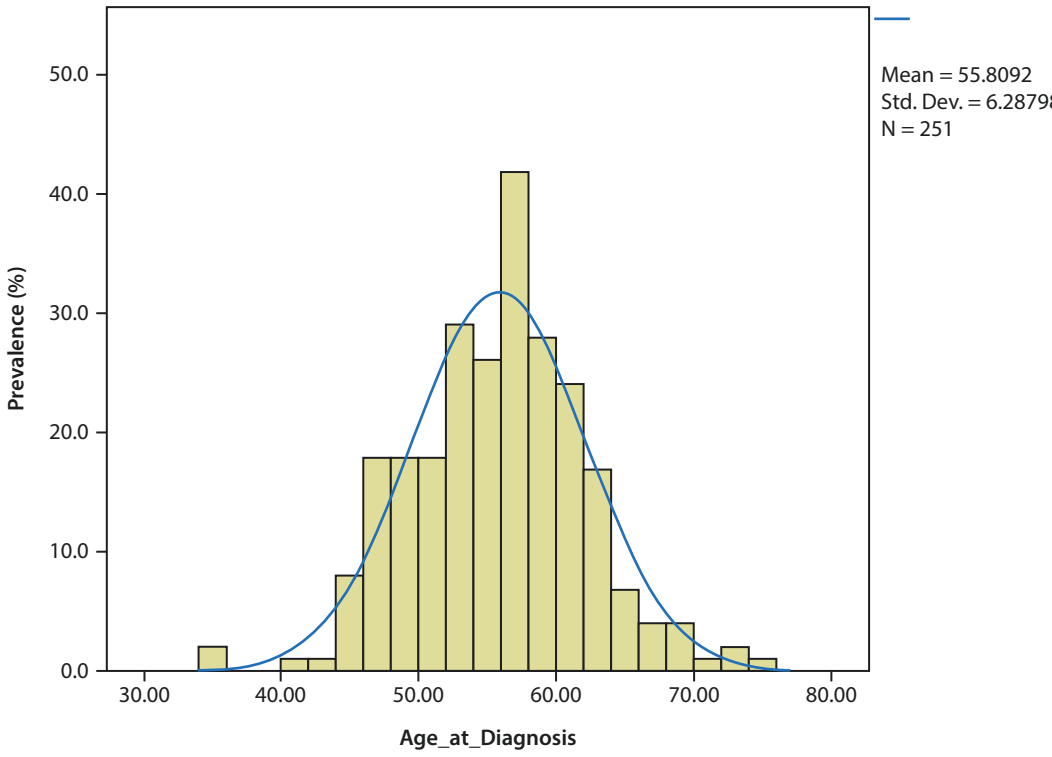
28.1.2 Dementia in Persons with Down Syndrome

Among people with DS not only is AD much more common, but the age of onset of dementia is much earlier than in the non-ID general population [19].

Reported average ages of diagnosis vary between 50 and 56 years, with median survival times of 45 to 84 months [20–22]. Despite near universal AD neuropathology in DS, there is great inter-individual variability in dementia presentation. Studies using information from medical records, informant report, and direct assessment of cognitive abilities show ages of diagnosis range from 35 to 74 years (◘ Figs. 28.2 and 28.3), with some individuals showing barely any signs of decline over the age of 65 [22, 23].

Before the age of 40, less than 5% of the DS population show dementia. However, the prevalence over age 40 increases steeply with age. The prevalence of dementia in DS was reported to be between 10% and 25% in the 40 to 49 years age group, 20% and 50% in the 50 to 59 years age group, and 30% and 75% in those older than 60 years [24–29]. This diversity in the prevalence rates may be caused by difficulty in diagnosing dementia in people with DS since their baseline cognitive abilities can vary considerably. The incidence similarly increases with age. ◘ Table 28.1 shows dementia incidence from a study of 506 adults with DS in the Netherlands [29]. A 20-year longitudinal study of 77 women with DS found all but two had developed dementia [21]. From these data, the calculated cumulative risk for developing dementia increased from 23.38% at 50 years of age to 88% at age 65.

The average prevalence of dementia in DS is about 15% at age 45 compared with 13% among individuals with non-DS ID at age 60 and over, and less than 12% for AD at age 65 in the non-ID general population [30]. Thereafter, the rate of dementia doubles with each 5-year interval in both groups with DS and non-DS ID (see ◘ Fig. 28.3) [31]. A recent publication of a cross-sectional study of 388 individuals with DS from Barcelona, Spain, and Cambridge, UK, have reported prodromal AD at a median age of 50.2 years (IQR 47·5–54·1), and AD at 53.7 years (49·5–57·2). The prevalence of clinically confirmed AD increased with age, reaching 90–100% in the seventh decade of life [32].



◘ **Fig. 28.2** Age of dementia diagnosis in a Down syndrome population [22, 23]

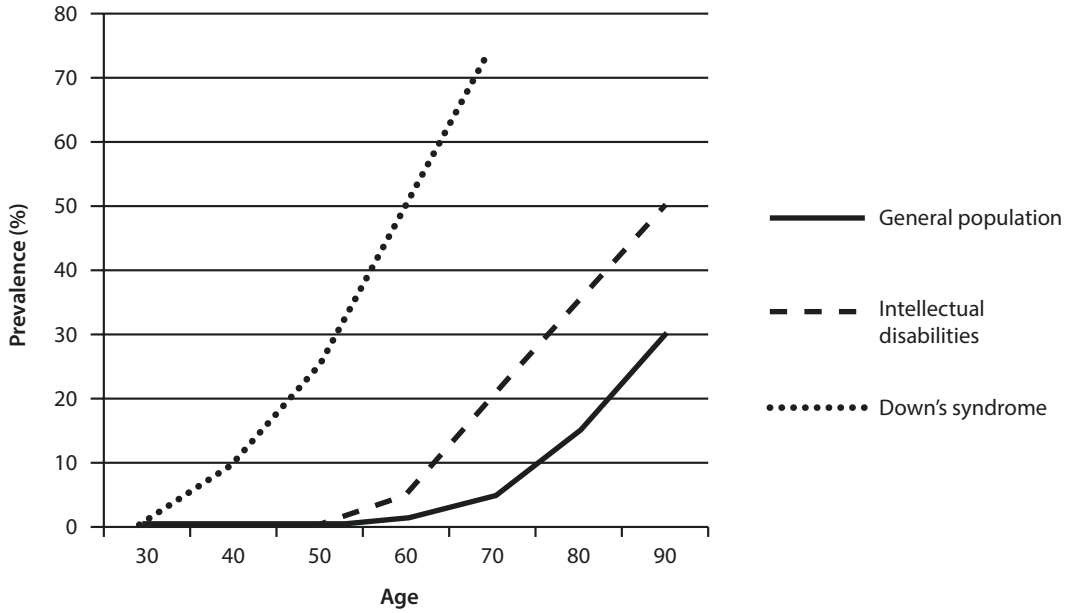


Fig. 28.3 Comparison of dementia prevalence rates by age in different populations [145]

Table 28.1 Incidence per 100 person years from Coppus et al. (2006) [29]

Age (years)	Incidence per 100 person years
<50	2.53
50–54	2.82
55–59	4.88
≥60	13.31

28.1.3 Diagnostic Criteria

The fifth edition of the *Diagnostic and Statistical Manual of Mental Disorders* (DSM-V) replaced criteria for 'dementia' with 'mild or major neurocognitive disorder', with specifiers indicating the likely underlying cause (e.g. AD, vascular disease) [33, 34]. While previous diagnostic criteria framed dementia primarily as a disorder of impaired memory, these new criteria view decline in any one of six cognitive domains (learning and memory, language, executive function, complex attention, perceptual-motor skills, and social cognition) as a key feature. This must not occur exclusively in the context of a delirium or be better explained by another mental disorder. In mild neurocognitive disorder, the decline would be modest and ability to live independently would not be affected. In major neurocognitive disorder, the decline would be significant, and the individual would require assistance in, at the very least, more complex instrumental activities of daily living, such as managing their bills or medication. A mild neurocognitive disorder has to be backed up by an objective neuropsychological test, which remains difficult to achieve in a number of individuals with ID, particularly those with severe and profound ID. A similar problem exists in determining the effect of cognitive impairment on everyday activities as often in the case for more severe ID these activities are dependent on external support. The ICD-10 criteria for dementia view dementia as a chronic or progressive disturbance of memory as well as other higher cortical functions, together with deterioration in emotional control, social behaviour, and motivation [35].

Both ICD-10 and DSM-IV dementia criteria were found to be reliable in individuals with ID, but when used in individuals with more severe ID and sensory deficits, predictive validity was less good than in the general

population [36]. It was noted that the ICD-10 criteria were more specific but less sensitive than the DSM-IV criteria. The ICD-10 criteria are more demanding to apply because they are more dependent on reliable information from informants. The ICD-10 and the Diagnostic Criteria for Learning Disabilities (DC-LD) criteria [37] require behavioural and emotional changes to make the diagnosis, but the DSM-IV does not. ICD-10 dementia criteria have also been shown to be reliable in DS, but a clinical diagnosis of dementia following comprehensive assessment in DS may identify cases before ICD or DSM-IV criteria for dementia is met [38].

28.1.4 Diagnosis of Dementia in Persons with Intellectual Disabilities and Down Syndrome

Making a diagnosis of dementia in individuals with ID could be difficult for various reasons [19]. For example, (a) the main criterion for making a diagnosis in the general population is the gradual deterioration in memory and other cognitive functions. However, many people with ID have a pre-existing cognitive impairment, therefore, creating a floor effect for cognitive assessment. Therefore, in many cases, the only way to assess a cognitive impairment will be to compare current state with a pre-morbid baseline of cognitive skills. (b) The level of cognitive abilities varies widely among people with ID. On one end, people with mild ID may have fairly intact cognitive skills to start with and may live independently in the community with minimum support. On the other end, people with severe/profound ID have a very poor cognitive skill and are almost totally dependent on others for their day-to-day needs. Therefore, it is difficult, if not impossible, to use the same diagnostic method for people with different levels of severity of ID. (c) There is also difficulty in obtaining reliable baseline levels of cognitive functioning from which to measure change. Therefore, it remains difficult to accurately identify when the cognitive decline begins. (d) Many people with ID will not have enough communication skills to express the symptoms associated with dementia. (e) For a diagnosis of dementia in the general population, it is important to establish that cognitive impairment has affected the person's functional abilities, but in case of people with severe ID, functional abilities are poor to start with, therefore, making it difficult to assess the impact of any cognitive decline on their day-to-day functioning [19].

Studies that have mapped out cognitive decline among adults with ID (both with and without DS) using prospective designs show a trend of relatively greater decline in verbal abilities compared with non-verbal abilities that has been the case in the non-ID general population [39, 40]. Krinsky-McHale and colleagues [41] observed changes in both the storage and retrieval of information using a modified Selective Reminding Task [42] several years prior to the diagnosis of AD. Urv and colleagues, on the other hand, showed evidence of behaviour change prior to the onset of dementia [43]. However, none of these symptoms are definite markers of future dementia. The validity of mild neurocognitive disorder as a definite diagnostic category among people with ID remains questionable at this stage. Also, the exact features associated with normal ageing and their overlap with dementia symptoms have never been properly studied among individuals with ID.

28.1.5 Clinical Features of Dementia in Intellectual Disabilities and Down Syndrome

There is an ongoing debate as to whether the presentation of AD in DS/ID population is similar to that in the non-ID general population. Many factors such as age, level of ID, pre-morbid personality, state of physical and mental health, and the individual's circumstances are likely to influence the pattern of clinical presentation of dementia in this population. The level of ID may also modify and in some cases mask the early manifestation of dementia. Both typical and atypical pre-

sentations of dementia have been reported in people with ID/DS [44]. The main clinical features of dementia in individuals with ID are presented in ► Box 28.1.

Dementia in people with DS and non-DS ID, as in the general population, is characterised by progressive decline in cognitive abilities and everyday skills (see ► Case study 28.1). Behavioural and psychological symptoms of dementia (BPSD), such as apathy, aggression, and sleep disturbances are equally noted, and in some adults with DS may occur a few years before dementia is clinically diagnosed [45, 46]. Many memory measures are insufficiently sensitive to assess the decline in lower functioning participants; [46] however, with sensitive tests, it has been shown that memory changes may occur earlier than the decline in executive function, similar to AD in the general population [47]. Caregivers are also more likely to respond to excessive behaviour (problem behaviour such as aggres-

Box 28.1: The Main Clinical Features of Dementia Among Individuals with ID

- Forgetfulness
- Confusion
- Apraxic symptoms: e.g. putting on clothes the wrong way round, doing up buttons the wrong way
- Speech and language problems: e.g. difficulty instigating a conversation, lack of expression and flattening of the tone of voice, repetitive questioning, and problems with understanding
- Slowness
- Loss of skills
- Problems with socialising
- Lack of confidence
- Obsessional behaviour
- Paranoia/suspiciousness
- Sleep problems: early morning waking, wandering at night
- Change in appetite, often weight loss
- Balance problems: unsteady gait, fearful of kerbs, leaning to one side, shuffling
- Taking a big step over a crack or uneven surface or pattern on the floor
- Emotional problems: crying episodes, easily upset and frustrated
- Shouting and screaming
- Agitation/restlessness
- Aggression (verbal and physical)
- Hallucinations, delusions, and illusions
- Loss of interest/apathy
- Covering up/confabulating
- Seizures

Case Study 28.1: Molly

Every Saturday morning, Molly takes the 439 bus across town to visit her 'little' brother, Harry. On the way, she usually stops in the local shop to buy a magazine for her nephew, Tommy.

But today, Molly was late, and Harry was annoyed. Harry reminded Molly last night about leaving before 11 am to take Tommy to his friend's birthday party, but at 12 pm, Molly was still not there. When Molly finally arrived at 6 pm, it was courtesy of the local police. She had been spotted by a member of the public on a country lane 10 miles from home, clearly lost and distressed. Molly had taken the wrong bus. Normally, she would call Harry if anything went wrong, but she had forgotten to bring her mobile phone. Harry was concerned – Molly had been taking that same bus route for years.

As Harry reflected over the past few months, several other moments sprang to mind. This year in particular, Harry is realising how much older Molly seems.

She remembered the time Molly shouted at Tommy, because he was sat in 'her' place. At other times, she seemed withdrawn. Harry remembered the tins of beans in the freezer and wondering at the time why Molly would have put them there, and she realised that she hadn't seen the diary that Molly religiously carried with her for several months. These were so out-of-character – Molly was usually calm, meticulously well organised, and outgoing. Harry had heard about people with Down syndrome being at risk of developing dementia, but Molly had only just turned 43, surely she was too young?

sion) than behavioural deficits (cognitive and functional impairment) [48], which may mean subtle early changes that cause no disruption are missed. Adults with DS who live with their families receive dementia diagnoses 4 years earlier on average than those in other settings, further highlighting how differences in sensitivity to change may influence diagnosis [22]. Gender does not appear to influence the age of onset [22, 29]; however, earlier onset follows early menopause in women with DS [49].

Using the Severe Impairment Battery, Dick and colleagues found similar neuropsychological profiles among 59 individuals with AD when compared with 51 non-ID individuals with moderate to severe AD [31]. However, overall it appears that in most people with mild to moderate ID, the early presentation is similar to that in the non-ID general population. In that, it seems that an impairment of recent episodic memory in the context of relatively intact distant memory is an early manifestation, whereas, in severe ID loss of skills, neurological symptoms such as seizures and possible problem behaviours seem to be the early manifestation. The first appearance of epilepsy in later life could be a sign of dementia in DS. There is a bimodal distribution of epilepsy in the DS population, with early-onset epilepsy (primarily infantile spasms which may or may not lead to Lennox-Gastaut syndrome) occurring in about 10% of children and a late-onset epilepsy associated with dementia [50]. The presence of seizures is a poor prognostic feature being associated with death within 3 to 5 years [51]. Occasionally, late-onset progressive myoclonic epilepsy may develop [52]. This is of interest as the gene for Unverricht-Lundborg myoclonic epilepsy has been identified adjacent to the critical site for DS on chromosome 21 [53].

Deb and colleagues gathered unconstrained information by interviewing caregivers of 24 individuals with DS and dementia regarding the symptoms, particularly the early symptoms of dementia using a qualitative methodology [54]. Findings show that many 'frontal lobe'-related symptoms such as lack of motivation, apathy, inability to socialise, general slowness, and lack of interest in life in general that are usually manifested later in the process of AD in the general population were common at an early stage of dementia among individuals with DS. Loss of interest in activities and problems with speech and language are also common in the early stage. Caregivers reported general confusion, physical and mental slowness, deterioration in pre-existing skills, and emotional and behavioural problems in the early stage of dementia in individuals with DS, whereas these symptoms tend to appear later in the course of AD in the general population. Some caregivers reported, although not frequently, early symptoms such as illusions (e.g. thinking a cat is rubbing against them, while in fact, the cat is sitting still in the room at a distance), visual agnosia (e.g. picking things out of the air), and tactile hallucinations (e.g. feeling of touch sensation that is not there). Change in personality in the form of aggression was rarely manifested in early stages, but personality change in the form of apathy and social withdrawal were not uncommon in early stages of dementia in people with DS.

Some had problems in following more than one instruction at one given time. One example of such a problem given by the caregivers is that a person would pick up all the knives or all the forks when she/he was asked to pick up a knife and a fork from a drawer in the next room. An early symptom of dementia in people with DS was evidence of slowness that affects almost all aspects of their functioning such as walking slowly, eating slowly, speaking slowly, particularly failing to initiate conversations, and slowness in the overall body movement. Some caregivers reported that the individual with DS they cared for would stop in the middle of a task such as eating or drinking.

Caregivers reported that many individuals with DS at an early stage of dementia showed speech and language problems in the form of tardiness in instigating a conversation, lack of expression and flattening of the tone of voice, and repetitive questioning, which was not present prior to the development of dementia. Other examples included speech becoming slow and difficult to understand (which was not the case before). Sleep problems in the form of early morning wakening and wandering at night were reported as early prominent features of dementia in individuals with DS in a number of cases. Some reported evidence of

catnapping during the day at the expense of less and less sleep at night.

Loss of self-help skills could be an early symptom of dementia among individuals with DS. In the beginning, the caregivers often reported that the person with DS needed more and more prompts to carry out simple self-help chores such as wiping face with a flannel or brushing teeth or drinking from a cup. Eventually, individuals with DS lost these skills altogether and could not carry them out even with a lot of prompts. Many caregivers reported that the individuals with DS took part in fewer socialisation activities and withdrew from other people around them and also did not show interest in activities that they used to enjoy before the onset of dementia. Another prominent feature of dementia in people with DS was unsteady gait. Some people tended to lean to one side while walking while others took a big step over a crack on the surface or an uneven surface, including kerbs and uneven patterns in the carpet. One unusual symptom such as 'pica' (eating/mouthing inedible substances) was reported by one caregiver.

There are two possible explanations for prominent features of 'frontal lobe' dysfunction early in dementia among adults with DS. First, even before developing dementia individuals with DS show structural brain abnormalities in the frontal and the temporal lobes, and are, therefore, vulnerable to loss of frontal and temporal lobe functions early if there is any brain insult. Second, it is often not possible to detect dementia in its early stage among individuals with DS. Therefore, dementia tends only to be diagnosed in these individuals once the disease has already progressed to its middle or late stage. This is also evident from the shorter period of time that is clinically observed between making the diagnosis of dementia and death among individuals with DS compared with the general population who do not have ID. The features of 'frontal lobe' dysfunction are not uncommon in the later stage dementia in the general population; therefore, it is not surprising to detect these features fairly early in the diagnosis among individuals with DS, given that these individuals are probably already in an advanced stage of dementia by the time a diagnosis is made [55]. Spatial disorientation was a frequently reported symptom and was present in both individuals with moderate and severe ID.

Prasher's study of common early symptoms of dementia among people with DS included mental deterioration, slowing, confusion, reduced output of speech, and deterioration in gait and personality change [56]. Evenhuis, on the other hand, described apathy and withdrawal symptoms as the most common early features of dementia among people with both moderate and severe ID [57]. Prasher described late features of dementia in individuals with ID as severe intellectual deterioration, marked change in personality and mood, loss of sphincter control, onset of epileptic seizures, loss of mobility with increased muscle tone, and eventual complete loss of all self-help skills [58]. Holland and colleagues found in an 18-month follow-up study that in the initial assessment, caregivers reported changes predominantly in behaviour and personality in individuals with DS who later developed dementia [27]. According to Holland and colleagues, the frontal-like dementia was more prevalent, particularly among the younger groups [27]. On the basis of their findings, the authors hypothesised that the frontal lobe functions are the first to be compromised with the progressive development of Alzheimer-like neuropathology in individuals with DS. Crayton and colleagues [59] in another 4-year follow-up study found that those individuals with DS who developed dementia at the follow-up showed evidence of impaired orientation and visuospatial memory at the beginning of the study, but language function and praxis deteriorated later. The progression of dementia in people with DS has been reported to be more rapid than in the general population, one of the reasons being a late diagnosis in this population [60].

28.2 Etiopathogenesis

DS initially described by J. Langdon Down in 1866 [61] is the most common highly variable developmental genetic condition that primarily (≅95%) arises from an additional copy of an entire chromosome 21 (Hsa21) [62]. Other genetic errors associated with DS

are Robertsonian translocations (i.e. some genetic material from Hsa21, most often from the long arm, is translocated to Hsa14 or 22, or from the long to the short arm of Hsa21) and mosaicism (i.e. the whole Hsa21 is triplicated in only a proportion of cells with other cells being euploid), accounting for 1–5% and 1–2% of DS cases respectively [63, 64]. Exceptionally, DS can also occur with partial trisomy 21 (i.e. duplication of only a delimited segment of Hsa21) [65]. These multiple individual differences in DS genetics need to be taken into account when discussing the aetiopathogenesis of DS-related AD [66].

28.2.1 Down Syndrome-Related Alzheimer's Dementia Neuropathology

The neuropathology observed almost universally among aged individuals with DS is almost identical to those found in people with AD in the general population. The two major hallmarks of this neuropathology are extracellular amyloid plaques (see Fig. 28.4) and intra-cellular neurofibrillary tangles (see Fig. 28.5) composed of amyloid β (Aβ) and phosphorylated tubulin-associated unit (tau) protein, respectively [67]. The mechanisms behind this are thought to be overexpression of amyloid precursor protein (APP), locus for which is found on chromosome 21 that leads to overproduction of Aβ [68] as well as overexpression of Dyrk1A (dual specificity tyrosine phosphorylation-regulated kinase 1A) and RCAN1 (regulator of calcineurin 1) also located on chromosome 21 which are both involved in hyperphosphorylation of tau [69–72]. Neuropathological changes begin many years before clinical symptoms [73]. Positron emission tomography (PET) studies showed that both tau and amyloid binding in vivo are related to age and cognitive status [74, 75]. There is evidence of elevated levels of total-tau (t-tau) [72], and markers of oxidative stress and amyloid deposition in blood and cerebrospinal fluid may be of particular value; however, further research is needed and as yet, there are no biomarkers that allow clear predictions of future clinical course on an individual basis [44].

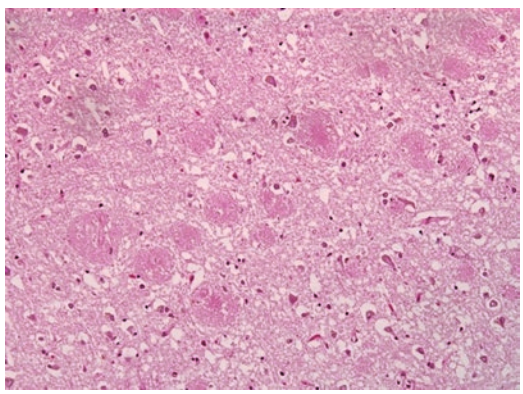


Fig. 28.4 Amyloid plaques, Alzheimer's disease, HE stain

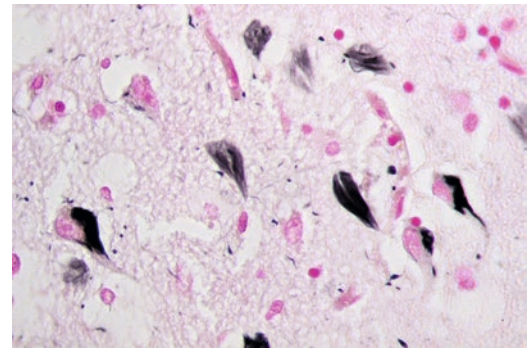


Fig. 28.5 Neurofibrillary tangles, Alzheimer's disease, Gallyas silver stain

There have been at least three seminal events in AD research: (a) the identification of the gene located on Hsa21 coding for the APP [76]; (b) the discovery of a missense mutation in the APP gene causing autosomal dominant familiar AD (FAD) in typically developing people [77]; and (c) the subsequent articulation of the amyloid cascade hypothesis (ACH) by John Hardy and Gerry Higgins [78] which posited that the extracellular deposition of the aggregated amyloid-β (Aβ) peptide – the major constituent of amyloid and neuritic (when associated with dystrophic and degenerating neurons) plaques in the brain parenchyma, initiates a sequence of events that ultimately lead to AD dementia (Fig. 28.1). Apart from the ACH and tau mechanisms, neuroinflammation is another important hypothesis in relation to AD which is relevant to DS as amyloid plaques lead to the production of neurotoxic pro-inflammatory cytokines [79]. Additionally, relevant to DS neuropathology, some other genes on Hsa21 include SOD-1 (superoxide

dismutase 1) and SLC5A3 (Solute Carrier Family 5 Member 3) [80, 81]. Mitochondrial dysfunction in neurons and astrocytes could also be involved, either independently or via altered APP metabolism [81–83].

28.2.2 The Amyloid Cascade Hypothesis and Down Syndrome-Related Alzheimer's Dementia

There is still a significant number of gaps in the ACH (see Fig. 28.1), among which at least three are fundamental. For example, we do not know (a) whether neurotoxicity and/or synaptotoxicity are caused by abnormal Aβ species as intracellular accumulations, or as extracellular free-soluble Aβ monomers, insoluble amyloid fibrils, and plaques or rather by the increase in the ratio of the longer (Aβ42) to the shorter forms (Aβ40) of the Aβ peptide; (b) how deposited Aβ triggers or accelerates the formation of neurofibrillary tangles (NFTs); and (c) what initiates ultimately the neuronal cell death that accompanies AD? The earlier outlined deficiencies in the ACH fostered in recent years the postulation of alternative hypotheses [84, 85], supported by the failure of multiple clinical trials targeting amyloid [86]. Yet, the ACH accommodates a wide range of data that we have on the aetiopathogenesis of DS-AD into a coherent hypothesis.

28.2.3 The Pathophysiology of Down Syndrome-Alzheimer's Dementia

Although our understanding of genetic and pathogenetic features in DS-AD has dramatically evolved in the last decade, some issues still remain unanswered [87, 88]. For example, besides the *APP* gene, approximately 13 protein-coding genes on HSa21 and four non-Hsa21 genes are thought to play a direct or indirect role in amyloidogenic *APP* processing and Aβ clearance, and in tau phosphorylation [88]. There are at least another 12 Hsa21 genes that are strongly implicated in neuro-inflammation, and oxidative stress deemed to be relevant in neuronal dysfunction and death according to the ACH [89]. Another 11 Hsa21-encoded genes and a five-cluster microRNAs flanking the *APP* gene are assumed to be involved in far-reaching epigenetic mechanisms [45, 64].

Also, the mechanisms underlying the wide variability in the onset of dementia in DS ranging from very early (<40 years) to very late (>70 years) onset remain largely unknown. Indeed, DS with trisomy 21 presents with many individual differences at the genome, single gene, epigenetic, cellular, neuro-developmental, cognitive, behavioural, and environmental levels, attesting the hypothesis that DS-AD may be considered a 'developmental disease' across the lifespan [66] so that, if individuals with DS live long enough, the development of clinical features associated with AD pathology appears almost inescapable as recently illustrated by McCarron and colleagues [21] who showed a prevalence of virtually 100% of dementia in 77 females at the end of a 20-year follow-up period.

28.3 Risk Factors and Biomarkers

An appropriate management would be helped by an understanding of the important risk factors for dementia in individuals with ID. Increasing age, a positive family history of AD, genetic factors, low educational level, sedentary lifestyle, history of head trauma, dyslipidaemia, diabetes mellitus, hypertension/hypotension, obesity, sleep apnoea, insomnia, depression, sensory impairments, periodontal disease, osteoporosis, chronic anticholinergic medication use, heavy smoking and drinking of alcohol, and cardiovascular disease have been identified as risk factors for AD and cerebrovascular dementia in the non-ID general population [90]. The use of non-steroidal anti-inflammatory drugs, antioxidants, and a high educational level may act as protective factors. There are likely to be similarities in the risk factors associated with AD in individuals with DS and the general population [19].

The Healthy Ageing in ID study involving 980 people with ID with a mean age of

61.5 years (range 50–93), of whom 861 had a diagnosis of DS, found that hypertension (53%), diabetes mellitus (14%), and metabolic syndrome (45%) were present similarly as in the general Dutch population matched for age [91]. Moreover, 50% of the people with hypertension had not been previously diagnosed with this condition. There is robust evidence that effective management of vascular risk factors delay the onset of AD in the general population [92], which is consistent with increasing research data that AD and cerebrovascular dementia have shared pathophysiology and neuropathological substrates [93].

Whereas the presence of trisomy 21 is a major risk factor for majority of people with DS, there is evidence that adults with DS who have atypical karyotypes including translocations, partial trisomies, and mosaicism have an improved survival and decreased rates of AD [94]. The presence of Apolipoprotein E allele 4 (ApoE e4) on chromosome 19 is shown to be a risk factor for AD in the general population, and there is some evidence that it may act as a risk factor for individuals with DS [95]. Some studies [96], though not all [95], have suggested that ApoE status, particularly the presence of the ApoE ε4 allele, affects the age of onset of dementia in people with DS. It is also suggested that ApoE status may influence the age of death [97]. Apart from the APP, there are other genes located on Hsa21 that may mitigate the occurrence of AD in individuals with DS [98, 99].

Mothers under 35 years of age of children with DS show a greater susceptibility to aneuploidy and trisomy 21 non-disjunction and have a five-fold increased risk for AD later in life compared with older mothers of a child with DS, to all fathers of a child with DS, or to the general population [100]. Low levels of oestrogen and an early-onset menopause may bring forward the age of onset of dementia in women with DS [101].

The cognitive reserve hypothesis has been proposed to explain how adults with similar brain insults may present with different clinical pictures [102]. The hypothesis proposes that there is a critical threshold of reserve capacity that needs to be breached by pathological processes before symptoms and signs develop. Individuals with DS will have reduced cognitive reserve due to their underlying decreased cognitive function and the likelihood that they will have had less education and may have resided in environmentally impoverished institutions. The cognitive reserve hypothesis predicts that adults with ID should be at risk for dementia and that they will be at risk of developing dementia at a younger age. There is a need for further research into this hypothesis to clearly elucidate its possible effects on individuals with DS.

There are controversial findings in the literature regarding whether or not the severity of ID acts as a risk factor for dementia [103]. Age, however, is the strongest risk factor for AD and in the case of individuals with DS, the effect of age is independent of severity of ID and gender [4]. Multiple vascular risk factors influence risk for AD and may act by increasing cerebrovascular pathology or also through a direct effect on AD pathology [104]. Coppus and colleagues showed that in individuals with DS, higher levels of plasma Aβ40 and Aβ42 were associated with increased risk of dementia [105]. Adjusted risk of dementia was increased in people with higher level of anti-fibril antibodies. Others have failed to show this association with dementia but showed Aβ40 level to be associated with cognitive decline in people with DS [106]. On the other hand, Matsuoka and colleagues showed that the ratio of Aβ42 to Aβ40 rather than absolute levels of the peptides is important to the pathophysiology of AD in genetically vulnerable population such as people with DS [107]. For example, a recent biomarker study of 388 adults with DS found that the Aβ1-42/1-40 ratio in the cerebrospinal fluid (CSF) and neurofilament light chain (NFL) values in the plasma changed among the participants as early as the third decade of life, and amyloid PET uptake changed in the fourth decade [18].F-fluorodeoxyglucose PET and CSF p-tau changes occurred later in the fourth decade of life, followed by hippocampal atrophy and changes in cognition in the fifth decade of life [32].

Other factors such as the raised mean corpuscular volume (MCV) of red blood cells (RBC) [108], impaired balance between

excitatory and inhibitory neurotransmitter system, particularly involving GABAA system [109] and higher levels of macrophage inflammatory protein-1α (MIP-1α) [110] have been implicated as risk factors, but further proof is required to confirm these findings. Triggering receptor expressed in myeloid cells 2 (TREM2) genetic variants are risk factors for AD and other neurodegenerative diseases. Plasma-soluble TREM2 (sTREM2) and other inflammatory markers are raised in adults with DS and dementia [111, 112].

The use of certain medications particularly with anticholinergic properties may enhance the expression of the neurocognitive disorder. A number of medication classes have been reported to be associated with possible worsening of cognitive function in people with dementia. These include antihistamines, especially first generation (diphenhydramine, hydroxyzine, promethazine), bladder agents (oxybutynin, tolterodine), certain pain medications (meperidine, propoxyphene), tricyclic anti-depressants, certain antipsychotics (chlorpromazine, clozapine, pimozide), and benzodiazepines [113].

There are also a number of psychological risk factors for dementia in individuals with ID. Esbensen and colleagues found that higher initial maternal and paternal levels of depressive symptoms and lower initial levels of paternal relationship quality were significant predictors of a higher likelihood of a dementia diagnosis in individuals with DS 10 years later [100]. Furthermore, lower initial levels of maternal positive psychological well-being predicted a higher likelihood of developing dementia in individuals with DS 10 years later, but the effect was only marginally significant.

Poorer overall physical health and depression are shown to be associated with an increased rate of dementia in individuals with ID [114]. However, it is not clear to what extent other known risk factors for dementia in the non-ID general population such as lack of engagement in mentally stimulating leisure and social activities, poor educational achievement, emotional trauma and associated post-traumatic stress disorder, poor diet and lack of exercise, also have a role to play in individuals with DS.

28.4 Diagnostic or Screening Instruments

Two main approaches have been taken for screening or making a diagnosis of dementia in individuals with ID with and without DS. These include instruments completed by caregivers of individuals with ID (informant-rated questionnaire) (see ◘ Table 28.2) or face-to-face interviews and examination, primarily including neuropsychological tests (see ◘ Table 28.3). There are pros and cons for each of these approaches. For example, direct cognitive assessment is impossible in people with severe ID and may be unreliable in people with even moderate to mild ID. On the other hand, different informants may provide different information on the information-rated scales. However, to counter this problem, most scales have assessed inter-informant reliability scores and found them to be good. The second problem is that many informants may not have known the person with ID since before the time of developing dementia, therefore, being unable to compare pre- with post-dementia symptoms. It is, therefore, recommended that both informant-rated and face-to-face cognitive tests should be administered periodically from age 30/35 years onwards in order to establish a pre-dementia baseline score. Zellinger and colleagues recently published a systematic review of assessment instruments for dementia in people with ID [115]. They found 35 instruments that are used for face-to-face cognitive testing in individuals with ID and dementia. They have also found 15 informant-rated scales that have been used either as a diagnostic tool or a screening tool for dementia in individuals with ID and/or DS. There are other recent reviews on rating scales for the detection of dementia in ID/DS [116, 117].

The face-to-face cognitive tests available involve assessment of various aspects of memory, language, executive function, orientation in time, place and person, attention, abstract thinking, visuospatial abilities, apraxia/dyspraxia, fine motor skills, dexterity, agnosia, etc. The informant-rated scales include assessment of behaviour and memory including adap-

Table 28.2 Informant-rated dementia rating scales

Domain assessed	Names of the instrument
Behaviour and memory	Assessment for Adults with Developmental Disabilities (AADS) [206]
Adaptive behaviour	Adaptive Behaviour Dementia Questionnaire (ABDQ) [131]
Cognitive functions	InterRAI-ID-cognitive performance test (InterRAI-ID-CPS) [207]
Cognitive function	Cognitive Scale for Down Syndrome (CS-DS) [208]
Dementia symptoms related	Dementia Screening Questionnaire for Individuals with Intellectual Disabilities (DSQIID) [125]
Dementia symptoms related	National Task Group-Early Detection Screen for Dementia (NTG-EDSD; adapted from DSQIID) [130]
Dementia symptoms related	Dementia Scale for DS (DSDS) [138]
Dementia symptoms related	Plymouth Dementia Screening Checklist (Plymouth-DSC) [209]
Behaviour and function test based	Dementia Questionnaire for People with Learning Disabilities (DLD) [123]
Behaviour and function test based	Multidimensional Observation Scale for Elderly Subjects-adapted for persons with DS (MOSES-DS) [129]
Behaviour and function test based	Behavior and Function Test (BFT) [210]
Behaviour and function test based	CAMDEX informant interview-DS [120]
Behaviour and function test based	Johanson interview from Johanson Test Battery [211]
Global outcome scale	Clinical Global Impression (CGI) [212]

tive behaviour, cognitive functions, but some are related to dementia-related symptoms. A global outcome scale has also been adapted for dementia screening in individuals with ID (see Table 28.2). A number of test batteries have also been developed (see Table 28.4).

The original CAMDEX (Cambridge Examination for Mental Disorders of the Elderly) was developed by Roth and colleagues [118] and revised in 1998 [119]. The full schedule includes an informant interview, an interview with the participant, an objective examination of cognitive function (CAMCOG) (Cambridge Cognitive Examination), and a physical examination and information on laboratory investigations. The original CAMDEX has been adapted for use in individuals with DS (CAMDEX-DS) (now updated as CAMDEX-DS-II) [120]. This is an informant-rated scale which has been combined with an adaptation of the CAMCOG as CAMCOG-DS (now updated as CAMCOG-DS-II) for use in individuals with DS, which is a direct cognitive assessment of individuals with DS [27]. Therefore, this battery combines an informant-rated interview with cognitive testing. Hon and colleagues [121] and Beacher and colleagues [122] found that the difficulty level of the CAMCOG led to exclusion of participants and limits the degree to which the results can be generalised to the DS population as a whole.

28.4.1 Informant-Rated Scales

Some of the early symptoms of dementia may be subtle or may present as an exacerbation of the existing behavioural traits or manifest differently in people with DS than they do in people who do not have ID. Only caregivers will notice these early and sometimes, unusual changes in the individual's behaviour and only by asking caregivers we can ensure that such symptoms are included in any case detection instrument. Therefore, an informant-rated instrument is ideal for screening or diagnostic purposes among individuals with ID and DS.

Table 28.3 Face-to-face neuropsychological assessment scales

Domains assessed	Names of the instrument
Cognitive function	Modified MMSE [140, 213]
Visuospatial function	Spatial Recognition Test (SRT) [214]
Delayed and immediate memory	Buschke Verbal Selective Reminding Task (4–6-year version) (SRT) [42]
Digit and sentence recall	McCarthy Scales of Children's Abilities (MSCA) [135]; Modified Boston Naming Test [215]
Memory for sentences, object and picture; delayed object memory	Rivermead Behavioral Memory Test (RBMT-C) [216]; Peabody Picture Vocabulary Test-revised (PPVT-III) [217]; The Fuld Object Memory Evaluation [218]; Modified Fuld [219]
Shoebox memory task, and delayed match-to-sample task	(Devenny et al., 2002) [220]
Verbal and semantic fluency	Controlled Oral Word Association Test (COWAT) [221]; Semantic verbal fluency [222]
Nonverbal reasoning/mental status	Leiter International Performance Scale (LIPS) [223]; Bayley Scales of Infant Development [224]; Test of Severe Impairment-Modified (TSI-M) [225, 226]
Visual perception, visual-motor integration and precision, spatial recognition span	Developmental NEuroPSYchological Assessment-II (NEPSY-II) [227]
Fine/perceptual motor skills	Modified Purdue Pegboard [143]
Dyspraxia, praxis	Dyspraxia Scale for Adults with Down Syndrome called the Brief Praxis Test (BPT) [228]
Executive function	Modified version of the Tower of London Test [3]
Language	British Picture Vocabulary Scale (BPVS) [229]
Not all tests mentioned in the above Table are specific for ID and AD [230]	

Table 28.4 Cognitive batteries used for detection of dementia in people with DS

Domains assessed	Names of the test battery
Cognitive function	CAMCOG-DS (part of the CAMDEX-DS) [120, 121]
Cognitive function	Cambridge Neuropsychological Test Automated Battery (CANTAB®), Cambridge Cognition Ltd., Cambridge, UK. 2016.
Cognitive function	Cognitive Assessment System (CAS) [231]
Memory, language, picture and object naming and identification	Neuropsychological assessment of dementia in individuals with intellectual disability (NAIID) [59, 232]
Memory, orientation, language, praxis, calculation	Prudhoe Cognitive Function Test (PCFT) (one long and two short Forms) [233]
Memory, language, executive functioning, motor performance, new learning	Rapid Assessment for Developmental Disabilities (RADD) [234]
Memory, orientation, language, attention, perception, construction, praxis, social interaction	SIB (Severe Impairment Battery) [235]

The Dementia Questionnaire for people with Learning Disabilities (DLD) [123] (formerly the Dementia Questionnaire for people with Mental Retardation; DMR [124]) and the Dementia Screening Questionnaire for Individuals with Intellectual Disabilities (DSQIID) [125] are perhaps the two most widely used informant-rated scales.

The DLD/DMR has 50 questions that have been divided up into the categories of 'short-term memory', 'long-term memory', 'spatial and temporal orientation' (making up the sum of cognitive scores; SCS), 'speech', 'practical skills', 'mood', 'activity and interest', and behavioural disturbance (making up the sum of social skills score; SOS). Each of the behaviours is rated as present either 'normally yes' (0), 'sometimes' (1), or 'normally no' (2). The overall score is calculated under two headings, namely, the 'sum of cognitive scores' (SCS) and the 'sum of social scores' (SOS). The cut-off scores for screening positive for dementia are different for individuals who have mild, moderate, or severe ID, respectively. The main problem with the scale is that it has shown low reliability when completed by caregivers [114]. Another practical problem is to determine accurately the severity of an individual's ID, which is required to determine cut-off scores for dementia.

Visser and colleagues followed up adults who had DS in a Dutch Institution with the Early Signs of Dementia Checklist (ESDC) [126], and Dalton and co-workers have used the Multi-dimensional Observation Scale for Elderly Subjects (MOSES) [127]. Neither has been properly validated for use in individuals with ID. Some studies have used the Adaptive Behaviour Scale [128] to estimate the rate of decline in adaptive behaviours in individuals with DS over a period of time [129]. The Adaptive Behaviour Dementia Questionnaire (ABDQ) is a 15-item questionnaire used to detect changes in adaptive behaviour and can be used as a screening tool [129]. However, it is not clear whether an assessment of the decline in adaptive behaviour alone is sufficient to make a diagnosis or screen for dementia, although adaptive behaviours are likely to be affected by dementia.

The DSQIID is unique in many ways [125]. Whereas most scales used professionals' views in developing the scales, a 'bottom-up' approach incorporating the views of caregivers regarding the symptoms of dementia was adopted for the DSQIID. Whereas almost all the scales included an inadequate number of participants with dementia to validate their scales, the DSQIID is the only scale that recruited an adequate number of participants with dementia for its validation. Unlike the other scales, the DSQIID avoids floor effect by comparing the current symptoms with a pre-morbid baseline. Also, unlike many other scales, the DSQIID is based purely on dementia symptoms rather than cognition-related items. The DSQIID has shown very good psychometric properties and factor structure. The DSQIID is a screening instrument and not a diagnostic tool and should not be used as such. In a clinical setting, the DSQIID is best used to explore each item with the caregiver in more detail in order to build up a clinical picture. If the DSQIID is used in a prospective manner, certain items may show floor effect as dementia progresses thus giving a lower total score as time progresses. Also, it is worth keeping in mind that there are certain physical and psychological conditions that may affect the scoring of certain items. For example, a cerebral stroke may affect scoring on items on 'walking and gait' and 'speech'.

The DSQIID has been translated into more than 16 languages for use worldwide, and in the USA, the National Task Group-Early Detection Screen for Dementia (NTG-EDSD) [130] has adapted the DSQIID and incorporated it into the NTG-EDSD [130], an early detection and screening tool. Validation of a Chinese (DSQIID-CV) [131], an Italian (DSQIID-I) [132], a Taiwanese [133], a German [134], and a Japanese version [72] have been published in peer-reviewed journals. Both the DSQIID-CV [131] and the DSQIID-I [132] have shown very good psychometric properties and good factor structure. Although in the original UK version Deb and colleagues found 20 as the best fit for specificity and sensitivity [125], in a subsequent recent study of the Japanese version of DSQIID (DSQIID-J), the

authors found a lower screening score of 10/11 [135]. Also, a recent independent validation of DSQIID items in the US NTG-EDSD an even lower score of 5 was seen to best fit the specificity and sensitivity for screening [136]. However, both the latter studies have shown good psychometric properties of DSQIID [135, 136]. A very recent study of the French version of DSQIID (DSQIID-F) has shown good psychometric properties and best fit between specificity and sensitivity at a total score of 19 [137]. However, as a screening instrument it may be better to use a lower cut-off score at the expense of detecting some false negatives, which could be excluded when a full clinical assessment is carried out at a later stage to confirm the diagnosis of dementia.

28.4.2 Neuropsychological Tests

Deb and Braganza highlighted the difficulty of using direct neuropsychological tests on people with ID [138]. The authors compared clinicians' diagnosis of dementia using Dementia Scale for Down Syndrome (DSDS) [139], DMR [124], and Mini-Mental State Examination (MMSE) [140] scores among 36 adults with DS without dementia and 26 adults with DS and dementia. DSDS and DMR scores correlated with each other and also with the clinicians' diagnosis (possibly because the same clinicians rated the DSDS and the DMR, and also have made the clinical diagnosis). It was only possible to administer the MMSE to 34 of the 62 (55%) participants. Thirty of these 34 adults (95%) had a score below 24 (the usual cut-off score for possible dementia diagnosis), but 23 (67.64%) of these had no diagnosis of dementia according to either DSDS or DMR or the clinicians' diagnosis.

Adult versions of neuropsychological tests are usually amended, or children's versions are used when testing adults who have ID. However, both these approaches carry the risk of losing the psychometric properties of the original scales. The 'McCarthy Verbal Fluency' Test [141], designed originally for children with normal cognitive function, has also been used for adults with ID. This test requires the individual to name as many words as possible within four specific categories, namely, 'food', 'animals', 'things to wear', and 'things to ride'. The initial simpler items of the 'Boston Naming Test' [142] can also be used to test language. The 'Purdue Pegboard modified' [143] tests constructional and psychomotor skills and requires the individual to place as many round pegs in round holes as possible in 30 seconds. The 'Brief Praxis Test' was developed by Dalton and co-workers [144] specifically to screen for early features of dementia in adults with ID. In this test, the individual is asked to carry out simple motor tasks such as raising an arm, shaking hands, and unscrewing the top of a jar.

Despite the use of all the above tests, their validity for use among adults who have ID still remains questionable. Perhaps an important strategy is to use these tests alongside an informant-rated behaviour questionnaire on a prospective basis.

28.5 Differential Diagnosis/ Co-morbidities

While making a diagnosis of dementia in individuals with DS, it is important to consider all the factors that may lead to cognitive impairment or give a picture similar to that of dementia. Sensory impairments, such as difficulty with hearing and vision, are not uncommon among people with DS, and they may lead to symptoms similar to that of dementia. There are a wide variety of potential causes for the decline in function in the ageing population with ID, many of whom have DS that are listed in ▶ Box 28.2 [113, 145].

Alongside their elevated AD risk, adults with DS experience higher rates of other common age-related issues, including sensory impairments and musculoskeletal disorders, than the general population and people with non-DS ID [146]. The development of dementia is further associated with higher levels of specific co-morbidities, including depression, hypothyroidism, epilepsy, and sleep disturbances [29, 147]. Capone and O'Neill [148] discussed other physical problems commonly associated with people with DS that may have

Box 28.2: Differential Diagnosis of Dementia/Decline in Function

- Depression
- Decline secondary to sensory impairment (visual and hearing)
- Endocrine disorders such as hypothyroidism
- Delirium
- Vascular disease: multi-infarct dementia
- Physical brain damage: tumour, head injury, epilepsy
- High-dose antipsychotics, medications with anti-cholinergic adverse effects, anti-epileptic drugs, and multiple medications
- Degenerative disorders: Parkinson's disease
- Infections: encephalitis, meningitis, Creutzfeldt-Jakob disease
- Anaemia, chronic infection, nutritional deficiency: vitamin B12, folate
- Impact of life events (bereavement, change in environment, move from home, loss of day activities, etc.)
- Physical and emotional abuse

an impact on dementia. Some of these include extrapyramidal symptoms, cervical spondylosis, atlanto-axial instability, feeding/swallowing problem, sleep apnoea, cardiac valve disease, peripheral vascular disease and atherosclerosis, autonomic dysfunction, the problem with bone metabolism, and early menopause. Individuals with the non-DS ID may also present with a range of physical health co-morbidities, including sensory deficits [149].

Assessment for, and treatment of, such co-morbidities should form an essential part of diagnostic procedures for individuals with suspected dementia. Thyroid disorders, depression, and sleep apnoea, in particular, could lead to misdiagnosis of dementia, as each can cause impairments in cognitive and/or adaptive functioning that could be reversed with appropriate treatment [150–152].

Thyroid Disorders

Thyroid disorders are more common in DS and non-DS ID than the general population and more common in older people with DS than younger [153, 154]. Prevalence of thyroid disorders is not directly related to dementia status in DS [147, 155]; however, thyroid disorders may be more common in those with end-stage than early-stage dementia [147]. Both hypo- and hyperthyroidism can impair cognitive abilities [150], thus given the increased risk, thyroid disorders must be first ruled out when assessing suspected dementia. Subclinical hypothyroidism (high basal level of thyroid-stimulating hormone (TSH) and low total or free T4) is present in approximately one third, and anti-thyroid antibodies are present in 18% of adults with DS compared with 6% of matched healthy control subjects [156]. The presence of anti-thyroid antibodies makes individuals with DS vulnerable to develop clinical hypothyroidism, and this must be differentiated from other causes of dementia as clinical hypothyroidism could be treated with supplementary thyroxine.

Depression

Depression can be both a co-morbid condition and a confounder of dementia (see ▶ Case study 28.2) [21, 155]. Some authors have reported a higher prevalence of depression among adults with DS, compared with adults who have non-DS ID [157], whilst others have not confirmed this [158]. There may be an overlap between the clinical expression of depression and dementia, which is important to distinguish, as most cases of depression are treatable. Untreated depression may exacerbate the cognitive decline in DS or ID, and cognitive abilities following extended periods of decline in these cases can recover upon appropriate treatment [159]. In adults with DS, treatment with anti-depressants regardless of the existence of depression has been reportedly associated with delayed onset of dementia and increased longevity, though this was not a randomised trial [152]. However, the previous history of depression and treatment with anti-depressants were both found to increase the likelihood of dementia in a longitudinal study conducted in the Netherlands [29].

Epilepsy

Epilepsy is strongly associated with dementia in the DS population [160, 161]. Between

Case Study 28.2: Vignette 2: Jerome

It had been 6 months since Jerome moved in to the care home. Jerome had lived in his family home for the previous 60 years, however when his mother passed away, with no other family to support him, Jerome had to move.

When he first arrived, Jerome was withdrawn and often tearful. He spoke very little with the other residents and required help from staff to get ready in the morning and make his own food and drinks. When staff took Jerome out, he would often try to go 'home' to his family home. The staff thought perhaps he kept forgetting that he had moved.

One member of staff had known Jerome as a younger man, when he attended a day centre that they were working in. They were surprised to see Jerome needing so much help. While Jerome had always required some support with shopping and travelling, he would always dress and clean himself and enjoyed helping to prepare lunch for everyone at the day centre. Being aware of the risk of dementia in this population, and knowing Jerome was now in his 60s, the staff arranged a review with a psychiatrist.

While Jerome's age and apparent deterioration could well be indicative of the beginnings of dementia, the psychiatrist was also mindful of the major bereavement and change in living circumstances Jerome was experiencing, so suggested a course of anti-depressants. Three months later, the staff reported a marked improvement in Jerome. He was managing his own self-care and chatting readily with the people he lived with, although he was sometimes getting his words mixed up. He was more settled in the home, although found it difficult to remember new staff member's names and would occasionally forget where things belonged.

40% and 77% of those who develop dementia will also have seizures [29, 147, 162]. For the majority, these seizures will be late onset, starting after the age of 35. Retrospective patient data showed a median time of seizure onset of 2 years following the dementia diagnosis, with generalised tonic-clonic seizures being the most frequently reported type [162]. However, in a recent clinical-video-EEG study of 22 adults with DS, Vignoli and colleagues [163] found focal seizures among nine (41%), late-onset myoclonic epilepsy among nine (41%), and unclassified type among four (18%) remaining participants. Indeed, some suggested that many generalised seizures observed in people with ID are focal seizures secondarily generalised, which could be wrongly classified in the absence of an appropriate investigation [50, 51]. The presence of seizures is associated with more severe cognitive decline, highlighting the importance of seizure management in this group [164].

Neurological complications, including extra-pyramidal symptoms, incontinence, and mobility issues are also common features of dementia in individuals with ID, particularly in later stages [165].

28.6 Assessment

In the USA the National Task Group (NTG) on ID and Dementia Practices [113] and in the UK the British Psychological Society and the Royal College of Psychiatrists jointly [145] produced consensus recommendations for the evaluation and management of dementia in adults with ID. The US guideline recommends the following steps for assessment: (a) Gather a pertinent medical and psychiatric history. (b) Obtain a historical description of baseline functioning. (c) Obtain a description of current functioning and compare with baseline. (d) Perform a focused review of systems. (e) Review the medication list thoroughly (see the earlier Section on 'risk factors' in this chapter). (f) Obtain a pertinent family history. (g) Assess for other psychological issues or changes. (h) Review social history, living environment, and level of support. (i) Synthesise the information. (j) Carry out a physical examination. (k) Carry out a mental state examination including cognitive assessment. In addition, the UK guidelines recommend certain relevant investigations (see ► Box 28.3).

Box 28.3: Schema for the Diagnosis of Dementia in a Person Who Has Intellectual Disability

- History from the person
- History from an observer/carer
- Examination of the person
  - Test for sensory impairments such as hearing and vision
  - Check for physical disorders, particularly hypothyroidism
  - Check for other psychiatric disorders such as depression
  - Check for epilepsy
  - Check for associated drug treatment
- Laboratory investigations (for differential diagnosis and cause of dementia; FBC, Vitamin B12 etc.)
- Neuroimaging (structural to exclude space-occupying lesion, etc.; functional to help with the differential diagnosis)
- Longitudinal use of neuropsychological tests and observer-rated questionnaire including an assessment of adaptive function

The assessment of an individual with DS suspected of dementia should start with a history from the individual and their caregivers. A mental state and physical examination of the individual with DS is imperative in order to make a diagnosis. Mental state examination should be used to exclude specifically the presence of depression, delirium, sleep problem, and anxiety (https://spectrom.wixsite.com/project) disorder. The issue of so-called diagnostic overshadowing is worth keeping in mind in which symptoms of dementia may be mistakenly attributed to the effect of ID in individuals with ID/DS. Physical examination includes assessment of all bodily systems including neurological examination for focal neurological signs and cardiovascular system including high blood pressure, and importantly assessment of sensory modalities such as hearing and vision. Neurological examination should also emphasise on language assessment and gait and motor coordination. Special emphasis should be placed in excluding hypothyroidism and anaemia. Standard laboratory tests should include vitamin B12 and folate levels, thyroid function test, full blood count (FBC), serum glucose/HbA1c, lipid profile, urea and electrolytes, renal function test, and liver function test.

An electroencephalograph (EEG) will be useful for those who have seizures and for whom anti-cholinesterase treatment is being considered an electrocardiogram (ECG) is necessary. There are differing views on the usefulness of neuroimaging in the work-up of the individual. Structural neuroimaging is widely considered to be useful to exclude space-occupying lesions, and it may also have a role in early diagnosis of dementia. An MRI study in individuals with DS found reduced medial temporal and striatal volume as a marker of AD [121]. These grey matter changes can be seen before the onset of clinical dementia [166, 167]. Functional neuroimaging such as Single Proton Emission Computed Tomography (SPECT) [168] and Positron Emission Tomography (PET) [169] have been used in individuals with DS and dementia [167, 170].

Recent advances in PET using new ligands have enabled researchers to show deposition of cerebral amyloid and tau in the brains of individuals with DS and dementia. Both [^{18}F] FDDNP and carbon 11-labeled Pittsburgh Compound B ([^{11}C] PiB) have been used to measure cerebral beta-amyloid [171, 172]. 11-PiB is used for amyloid and FDDNP for both amyloid plaques and neurofibrillary tangles [170]. Amyloid deposition, particularly in the posterior cingulate gyrus, has been detected using 11PiB before the clinical onset of dementia. Some showed a different distribution of amyloid deposition in the brains of people with DS compared with people with AD in the general population, in that the first deposition is shown in the striatum at around age 40 years, which some suggested could be the precursor of dementia. The FDG-PET showed hypo-metabolism of glucose in the posterior cingulate and other relevant brain areas even before the onset of dementia in

people with DS [166, 173]. Although paradoxically in some adults with DS who did not develop clinical dementia, hyper-metabolism was also observed. This may be a possible compensatory mechanism before manifestation of hypometabolism [167].

Diffusion Tensor Imaging (DTI) studies showed impaired integrity in frontal tracts in people with DS and dementia compared with people with DS who did not have dementia [174, 175]. Increased white matter damage in-PiB positive subjects with DS, predominantly in the posterior tracts has also been reported [167, 176].

However, neuroimaging requires informed consent which will require accessible information to be given to the individual and also possibly an assessment of capacity to consent to the procedure. The procedures are often long and complex requiring the co-operation of the individual, and this is often difficult in this population. The British Psychological Society guideline [145] concluded that neuroimaging is not an essential investigation but that it may be of value where other brain lesions or vascular dementia are suspected.

Therefore, for regular prospective screening, an informant-rated dementia symptom-based scale such as the DSQIID/NTG-EDSD [125, 130] along with neuropsychological screening if appropriate could be used. A more detailed diagnostic battery such as the CAMDEX-DS [120] could be used if the decline is noted or if the diagnosis is unclear. An assessment of environmental factors is also mandatory. Ageing individuals with DS are likely to face many changes in their lives. For example, there could be a change in their living environment as their parents may not be available any longer to support them. Similarly, there may be a change in care staff or residents in the community home where they live. Many of these individuals are also likely to see the death of parents or other close family members. There may be a change in occupational status, leisure activities, or in friend circle. Given the limited coping skills, individuals with DS are likely to be affected at times quite profoundly by these life events which would have an effect on their cognition and functional skills. Therefore, these issues have to be considered carefully before a definitive diagnosis of dementia is made.

28.7 Management Options

28.7.1 Pharmacological and Medical Interventions

Two classes of medication are currently licenced for the treatment of dementia, particularly AD. Acetylcholinesterase inhibitors such as donepezil, galantamine, and rivastigmine are used for mild to moderate dementia. Adverse effects may include dose-related cholinergic effects such as nausea, vomiting, hallucination, sleep disturbance, dizziness, and agitation. Available evidence suggests that acetylcholinesterase inhibitors are well tolerated by individuals with DS/ID, although lower doses may be required due to bradycardia which is common in DS [177]. Therefore, it is advisable to carry out an ECG prior to initiation of treatment and monitor pulse rate regularly thereafter. Memantine is a glutamate receptor antagonist often used in severe dementia. Adverse effects include constipation, headache, dizziness, and hypertension.

A recent Cochrane review on the efficacy of drugs in improving cognition in people with DS included nine randomised controlled trials (RCTs) [178]. Of these, four studies were on donepezil, two on memantine, and one each on simvastatin, anti-oxidants, and acetyl-L-carnitine (a dietary supplement). Four of these studies included participants below age 30, before the age of expected onset of clinical dementia in this population. Only simvastatin had shown some positive difference on cognition when compared with the placebo group but given the very small number of participants whose data were analysed ($n = 13$) [179], this finding is of very limited value. Donepezil group showed a statistically significant higher rate of adverse effects when compared with placebo group (Odds ratio: 0.32). However, no significant inter-group differences were observed with other active interventions. Authors concluded that due to the low qual-

ity of the body of evidence synthesised, it was not possible to draw any definitive conclusion about the efficacy of any medication in the management of cognitive decline in people with DS. However, a recent practice-based cohort study of 310 adults with DS and AD in the South of England in the UK showed a statistically significant longer median survival time among those who were prescribed anti-dementia drugs (primarily acetylcholinesterase inhibitors) (5.9 years; 95% CI: 4.67–6.67; n = 145) compared with those who were not (3.45 years; 95% CI: 2.91–4.13; n = 165) during the period of 2000 and 2013 [180].

Current guidance suggests that medication for dementia in individuals with ID should be initiated by specialists [145]. Once started, treatment should be carefully monitored to assess benefit and adverse effects and may need to be stopped if not tolerated as dementia progresses. Medical management is generally supportive. Common issues include sleep problems with reversal of sleep-wake cycle, which can be associated with problem behaviour, but sleep apnoea, which is a common co-morbidity in DS, has to be excluded first. Sleep-wake problems can be managed using simple interventions such as reinforcing the cycle, and melatonin may be useful [177]. In the early stages of dementia, additional functional gains can be made by managing sensory impairments such as hearing and vision problems. Respiratory and other infections that are common in individuals with dementia should be treated early. In the case of dementia-related seizures, a partial or complete response to treatment with anti-epileptic medication, particularly levetiracetam, has been described [162].

Eventually, dementia will progress with increasing complications, including problems with eating and drinking and incontinence, myoclonus or seizures, increased risk of infections, and mobility issues. The person may become bed-ridden and need 24-hour support. Palliative and end-of-life care should be instigated in a timely manner with the aim to support quality of life, addressing both psychological needs and symptom management. Areas that may require particular attention towards the end of life include swallowing assessments and eating and drinking guidelines to avoid aspiration, pain relief, and posture.

28.7.2 Non-pharmacological Management

There is a range of non-pharmacological management that may be used to promote quality of life, potentially avert or respond to symptomatic issues, and/or support family and other caregivers [181]. Guidelines for the care and treatment of persons with and without ID affected by dementia recommend the use of non-pharmacological therapeutics over pharmacological options as a first approach in addressing issues that arise in dementia care [182]. Table 28.5 identifies an assortment of non-pharmacological therapeutic interventions commonly found in practice. Research on specific therapeutic interventions used with persons with ID experiencing dementia or aiding their caregivers is sparse [183], although the literature offers some evidence of the utility of select interventions. Dodd and colleagues [184] noted that these include developing an understanding of dementia [185], anxiety and stress reduction [186], life story work [187], reminiscence [188], validation techniques [189], and helping peers to understand dementia [190]. The use of support groups for persons with ID and dementia which employ various strategies (e.g. information and education using visual aids, role-playing) has its benefits [191, 192]. A range of other generic therapeutic approaches may also be effective with people with ID and dementia, including music therapy [193, 194], aromatherapy, sensory stimulation (including Snoezelen), touch, and the use of electronic devices, domotica (smart homes), and entertainment. Generally, the most commonly used approaches for people with ID are the same as those used in the general population; however, some applications may need to be adapted contingent on the innate level of ID [181]. Their utility will be stage- and function-based; the efficacy of each approach needs should be individually monitored and evaluated [195]. Family caregivers and paid-staff

Table 28.5 Possible applications of non-pharmacological management strategies

Therapeutics	Examples	Rationale
Counselling and group support	Individual, family, and significant others	Address psychosocial/emotional needs of individual affected and caregivers
Culture and arts based and other therapies	Dance/movement, music/music therapy, art, pet, massage	Enhance quality of life, maintain interests/abilities, address behaviours
Reminiscence	Life story work, review of photos and memories	Keep long-term memory active and permitting favourable memories to come forth
Education and skills training for caregivers	Dementia information, communication strategies, other specific interventions	Modify caregivers' interactions, reduce behavioural reactions/stress
Health discussions	Physical exercise, dining strategies, personal reviews of physical factors	Maintain health and improve diet, nutrition, and hydration of person
Multisensory approaches	Snoezelen, Montessori	Stimulate yet calm the individual
Advanced dementia physical care	Bathing, voice modulation and facial expressions, massages, sensorimotor activities (such as 'twiddle muffs'), aromatherapy, scented lotions, use of durable medical equipment, help with swallowing and breathing	Prevent skin breakdown, provide physical and sensory stimulation Maintain relationships

Source: Jokinen, 2014 [181]

should be given information, instruction, and receive follow-up to effectively incorporate non-pharmacological therapeutics into a person's daily or weekly routine.

The most recent systematic review on non-pharmacological interventions for people with dementia in the general population who do not have ID has shown that person-centred training, communication skills training, and adapted dementia care mapping decreased symptomatic and severe agitation in care homes immediately (Standardised Effect Size; SES range 0.3–1.8) and for up to 6 months afterwards (SES range 0.2–2.2). Activities and music therapy by protocol (SES range 0.5–0.6) decreased overall agitation, and sensory intervention decreased clinically significant agitation immediately. However, aromatherapy and light therapy did not demonstrate efficacy [196]. A recent Cochrane review has shown a decreased need for antipsychotic use among patients with dementia in the general population when psychosocial interventions including staff training were introduced [197, 198]. However no such evidence exists involving people with ID.

28.8 Service Needs

An ideal service for people with ID and dementia should ensure that (a) periodic, but regular and planned, reviews are undertaken of the person's programme/care plan to identify significant changes in health, function, and quality of life, and adjustments are made in activities and care practices to ensure that the person continues to receive quality person-centred care; (b) early identification of behavioural and psychological symptoms of dementia occurs and reviews of care practices and supports are undertaken when such symptoms are present; (c) support is offered to caregivers throughout the course of the condition, from both specialist and mainstream services, and there is a continuing provision of information; and (d) quality of life is evaluated at regular intervals from both the perspective of the person with ID and caregivers, across the course of the person's journey.

Generally, dementia-related care is stage-based and in Table 28.6 [182] we offer a structure for stage-based activities and

Table 28.6 Pre-and post-diagnostic supports for community care

Pre-diagnostic stage	*Recommended actions:* Make provisions for the ongoing information needs of individuals, friends, and families as well as staff training to better understand the diagnostic process and progressive nature of dementia Use a detection/screening tool on a regular basis to capture early warning signs that may or may not indicate dementia Assess for medication-induced adverse drug reactions or other conditions mimicking, exacerbating, or masking dementia Dialogue with a healthcare professional or clinician about the screening tool results and, if the suspicions appear supported, seek a referral for a formal health assessment Advocate that trained professionals familiar with assessment and diagnosis of adults with ID and cognitive/functional decline become involved Have a person familiar with the adult with an ID and his or her history and communication method always accompany the adult to the assessment appointment(s) Hold meetings with the individual, family members, and others important to the person, if a diagnosis of dementia is obtained, to explain the diagnosis, prognosis, and begin to map out priorities for future support
Post-diagnostic stages	
Early stage: Mild dementia	*Recommended actions:* Engage the individual and their family, and/or other caregivers or guardians in advance care planning (and prepare advance directives) consistent with state or other requirements Identify and plan to remediate the environmental challenges to help maintain community living Establish a daily regime that provides for purposeful engagement based on individual needs and preferences yet is organised so as not to cause anxiety and confusion Provide ongoing clinical support to address behavioural and psychological symptoms associated with dementia Redesign day activities and programmes so that participation in valued activities and opportunities for interaction with others continues and respite for families and other caregivers is possible
Mid-stage: Moderate dementia	*Recommended actions* Provide increased assistance with personal care and hygiene when needed Secure appropriate residential supports and consider housing options to accommodate increasing losses in independent functioning Continue surveillance and periodic assessments to determine extent of change and progressive dysfunction as well as the possible development of co-morbid conditions Monitor any medications being taken to prevent adverse drug reactions Enhance training of staff and family as well as consultation to caregivers around coping with behaviours and adapting routines Provide caregivers information about potential complications, and how to prevent these, including falls (footwear), swallowing issues (choking/coughing during eating, which needs rapid assessment), sleep issues, risk for infections, etc. Institute planning for long-term services and supports Ensure protections are in place to preclude abuse or harm in both formal and informal settings
Late stage: Advanced dementia	*Recommended actions* Reorganise care management towards non-ambulatory care Identify eating/drinking/swallowing issues, also infections, aspiration pneumonia, etc. Reassign staff to activities more structured around nursing and personal care including the support of family caregivers who wish to maintain the person at home Obtain support from palliative care or hospice specialists Institute procedures to maintain dignity, comfort, and address pain and symptom management Organise end-of-life supports and post-death arrangements

Source: Jokinen et al., 2013 [182]

adaptation in care practices as dementia-related decline runs its course. As noted in the pre-diagnostic stage, preliminary activities are designed to identify or validate the presence of mild cognitive impairment or early-stage dementia. In the post-diagnostic stages, activities and care practices are progressively more focused on coping with and management of gradually deteriorating function. Early-stage practices tend towards personal supports and enabling the person to remain-in-place. With the deterioration of function, the focus of mid-stage practices shifts to greater levels of supports as well as introducing various environmental adaptations. In structured settings, this stage often calls for more enhanced staffing and supervision, while providing personal supports to maintain the integrity of the person's function and skills. For more detailed information and recommendations related to stage-based care, we recommend sourcing the following International Summit on Intellectual Disability and Dementia statements: for general post-diagnostic supports (PDS) issues [184], for advanced dementia [199], for family supports [200], and for end-of-life care and dementia [145, 201].

A multi-disciplinary/multi-professional approach to assessment and service provision in which General Practitioners/Family Physicians, Psychiatrists, Neurologists, Nurses, Psychologists, Physiotherapists, Social workers, and Occupational Therapists are involved, is vital in order to lessen the burden of care. Watchman [202] suggests a need for increased awareness and training for professionals in primary care. She also suggests that one professional should take responsibility for sharing the diagnosis with the individual and their caregivers.

In terms of accommodation, the best practice is to facilitate ageing and caring in the same home where they have lived in if possible, to do so. Moving home at the time of increased confusion may be detrimental and hasten their decline and increases the chances of emerging problem behaviour. However, if a move away from family home really becomes inevitable, then relocation to a model service specifically designed to care for people with DS and dementia as suggested in De Vreese and colleagues' paper [203] would be ideal. In the late stage of the disease, a co-ordinated service from Health, Social, Voluntary, and Private sector may be required because in the majority of cases these individuals may have to be transferred away from their home settings and looked after within one of the services mentioned earlier in the text. It is important to ensure that caregivers are fully involved in the formulation and provision of care. Other important areas to assess are the organisational settings, systems, and processes in place to support the person with ID and dementia and their caregivers, and the need to address the possible absence of adequate support.

28.8.1 Cross-Cutting Management Issues

Abuse and neglect. With diminished capacity, adults affected by dementia become vulnerable to different kinds of abuse or mistreatment, which can take varied forms, such as emotional, physical/sexual, and financial. Neglect, a form of abuse, is the refusal or failure to fulfil caregivers' obligations or duties to a person under their care. Self-neglect, also a form of abuse, occurs when an adult with dementia who lives alone loses the ability to care for him or herself, lacks support resources, and poses a threat to his or her health, safety, and well-being. Whatever the source, abuse, mistreatment, and neglect should not be tolerated, and any instances immediately reported to the appropriate authorities [204].

Nutrition. Weight loss and under-nutrition are common complications that occur with dementia. Under-nutrition adversely affects an individual's functional abilities, resistance to infections, and general overall health status and independence. As dementia progresses, the person increasingly becomes more dependent on caregivers to adapt meals and meet nutritional needs. Malnutrition is preventable and can be addressed; early intervention strategies should be set up and management of emerging nutritional problems dealt with rapidly.

Relationships. In person-centred care for dementia, Kitwood [205] has related person-

hood to the relationship and social being. In the context of relationships, life stories can be used to nurture a sense of history and selfhood with people with dementia. Assisting caregivers to understand behaviours and offering them tools to connect with and 'be in the present' with the person 'as is' can enhance well-being for both the person with dementia and the caregivers. PDS also involves aiding families and paid care staff, to deal with emotions, such as grief (for multiple losses), guilt, and self-doubt which can colour interactions, and to receive education and counselling.

The shift in philosophy. In many cultures, the overall focus for people with ID is based upon fostering skill building, along with increasing independence and autonomy. Goals and strategies that value and promote skill maintenance and overall well-being ('being not doing') in dementia care should be employed that recognise and promote greater systems change.

Education and training. Best practice would dictate the exposure of staff or other workers to information about dementia and its effects. Education should include an understanding of best practices for providing day-to-day specialised dementia supports; a familiarity with adaptations and modifications to the physical and social environments, as well as to activities to promote active engagement; and exposure to a range of models for providing community-based dementia care and supports. Training should also lead to a familiarity with the range of resources available to help support people with dementia and their families [204].

28.9 Conclusion

A case could be made to make caregivers aware of the main themes of expression of dementia in individuals with ID so that the caregivers could raise suspicion at an early stage about the condition and an appropriate referral could be made to specialist services for confirmation of diagnosis. Whereas an early and accurate diagnosis of dementia in individuals with ID is necessary for timely management, it is equally important to bear in mind that a false positive diagnosis of dementia may cause unnecessary anxiety to the individuals and their families and lead to wrong management of the problem. A multi-disciplinary, person-centred approach to diagnosis is, therefore, recommended in which a baseline is established at a stage before dementia is likely to occur. Use of screening instruments followed by a full work-up by an appropriately qualified professional or a group of professionals when dementia is suspected is likely to provide the best yield in terms of diagnosis. A longitudinal approach to diagnosis would be desirable, where the intervals between assessments decrease in proportion to increase in age and subsequent amplification of risk. The effect of medical, psychological, social, and environmental factors on cognition of individuals with ID has to be carefully assessed. It would be desirable for local services to set up a register of adults with ID/DS in order to carry out a baseline assessment of cognitive and adaptive functioning which should be further assessed at regular intervals in a prospective manner. It is also desirable that each service should develop specialist skills in this area and offer training to other professionals, front line staff and caregivers. Caregivers and health professionals need more information on behavioural and cognitive changes in early stage of dementia so that an earlier and more effective referral to specialist services can be made. In terms of accommodation, the best practice is to facilitate ageing and caring in the same home where the individuals with ID have lived in if possible to do so. Moving home at the time of increased confusion may be detrimental and hasten their decline, although this may become unavoidable as the disease progresses to an advanced stage.

Key Points

- Dementia is more prevalent in people with intellectual disabilities and more so in people with Down syndrome.
- Onset of dementia in people with Down syndrome is earlier in their lives com-

pared with the general population who do not have intellectual disabilities.
- Dementia at its early stage may be difficult to diagnose in people with intellectual disabilities.
- A combination of caregiver-rated questionnaire and direct examination of the person with intellectual disabilities is necessary to reach a clinical diagnosis of dementia.
- Usually loss of recent memory in the context of intact distant memory as is the case in the general population is an early symptom of dementia in people with mild to moderate intellectual disabilities.
- Usually loss of skills is the early symptom of dementia in people with severe and profound intellectual disabilities.
- There is no cure for dementia, but the medication used for treating symptoms of dementia in the general population who do not have intellectual disabilities should also be used to treat people with intellectual disabilities.

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Disorders Due to Brain Damage and Dysfunction and to Physical Diseases (Excluding Neurocognitive Disorders)

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Contents

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Learning Objectives

- Concept and definition of organic mental disorders (other than dementia and mild neurocognitive disorder)
- Symptomatic similarity between organic and nonorganic mental disorders in persons with intellectual disability (ID) and/or autism spectrum disorder (ASD)
- Prevalence of organic mental disorders in persons with ID and ASD
- Relationship of organic mental disorders and some etiologically defined forms of ID and/or ASD
- Relevance of thorough and comprehensive physical assessments
- Bias that may limit the epidemiological estimate of epilepsy in people with ID and ASD
- Factors correlated with epilepsy that may determine adjunctive cognitive dysfunction
- Factors to be considered in choosing antiepileptic drugs
- Importance of multi-/interdisciplinary approach in diagnosis and treatment of organic mental disorders and epilepsy in persons with ID and/or ASD

29.1 Introduction

Besides dementia or mild neurocognitive disorders, there are other mental disorders caused by brain dysfunctions or brain damage. Brain dysfunctions may be a consequence of a primary cerebral disease (e.g., encephalitis) or a systemic somatic disease including endocrine disorders (e.g., hypothyroidism) secondarily affecting the brain, and of exogenous toxic substances disturbing the brain function. Because such disorders are based on identifiable organic changes of the brain (primary brain involvement) or of the body (physical disease) secondarily affecting the brain function (secondary brain involvement), they are often called *organic mental disorders*. Organically caused mental disorders are able to imitate nearly all "non-organic" mental disorders (e.g., schizophrenia or affective disorder). In other words, the psychopathological features of the syndrome itself do not expose the "organic" nature. A thorough physical assessment has to be part of diagnostic procedures, particularly in persons with a first manifestation of a mental disorder, because clinical manifestations of organic mental disorders may resemble – or are identical with – those of disorders not regarded as "organic."

The ICD-11 [1] terms this group of disorders as "secondary mental or behavioural syndromes associated with disorders or diseases classified elsewhere" and includes "syndromes characterised by the presence of prominent psychological or behavioural symptoms judged to be direct pathophysiological consequences of a medical condition not classified under mental and behavioural disorders, based on evidence from the history, physical examination, or laboratory findings." ICD-11 [1] also specifies that symptoms of this group of disorders "are not accounted for by delirium or by another mental and behavioral disorder and are not a psychologically mediated response to a severe medical condition (e.g., adjustment disorder or anxiety symptoms in response to being diagnosed with a life-threatening illness)." The diagnostic categories included in this meta-structure "should be used in addition to the diagnosis for the presumed underlying disorder or disease when the psychological and behavioural symptoms are sufficiently severe to warrant specific clinical attention" [1].

In accordance with current ICD diagnostic criteria [2], a clinical syndrome is to be diagnosed as an "organic" mental disorder if the following conditions are met:

(a) Evidence of cerebral disease, damage, or dysfunction or of systemic physical disease, known to be associated with one of the clinical syndromes.
(b) Temporal relationship between the development of the underlying disease and the onset of the mental symptoms.
(c) Recovery from the mental disorder following removal or improvement of the presumed underlying cause.
(d) Absence of evidence to suggest an alternative cause of the mental syndrome (e.g., family history or precipitating stress).

Criteria (a) and (b) already justify a provisional diagnosis of organic mental disorder. The certainty of diagnosis is significantly increased in the case that all four criteria (a–d) are met.

Various clinical conditions and factors may result in an increased risk of organic mental disorders: for example, epilepsies, head trauma, brain damage due to hypoxia, infectious and parasitic brain diseases, brain tumors or brain metastases, cerebrovascular diseases, endocrine diseases, metabolic disorders, some nonpsychotropic drugs (e.g., propranolol, levodopa, methyldopa, steroids, antihypertensives, antimalarials).

Persons with intellectual disability (ID) or autism spectrum disorder (ASD) are particularly exposed to such diseases or clinical conditions due to the high rate of physical health problems, which is more than double that of the general population [3–5]. Furthermore, some etiologically defined forms of ID and/or ASD are associated with specific risks for certain physical disorders that often determine organic mental disorders. Examples are represented by the elevated prevalence of hypothyroidism in trisomy 21 [6] and the high risk of renal insufficiency in tuberous sclerosis [7].

Epilepsy is also much more common in persons with ID and/or ASD than in the general population and shows higher association with secondary mental or behavioral problems, less favorable prognosis, and consequent more complex diagnosis and management [8–13].

Persons with ID and/or ASD are particularly exposed to mental or behavioral syndromes secondary to physical diseases, due to the high rate of physical health problems in general and the frequent link between specific genetic alterations and specific physical disorders determining psychopathology.

29.2 Specific Clinical Disorders

The general descriptions of organic mental disorders in the following rubrics are guided by the description of the ICD-10 [2].

29.2.1 Organic Hallucinosis

General Description

An organic hallucinosis is characterized by recurrent or persistent visual or auditory hallucinations, occurring in clear consciousness. The hallucinations may or may not be recognized as such by the subject. Delusional elaboration of the hallucinations may exist, but insight is not infrequently preserved.

Organic hallucinations may occur in alcohol dependence [14], in cocaine dependence [15], in tumors or other lesions of the temporal lobe [16–18], or in Parkinson's disease [19]. In addition, anticholinergic or dopaminergic substances may result in organic hallucinations [20]. The relation of hallucinations to epilepsy is highly complex and the administration of neuroleptics in epilepsy-related hallucinations should be handled with care [21, 22].

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Dravet syndrome [23] is a rare encephalopathy due to a gene mutation in most cases. Myoclonic seizures (starting in the first year of life) are the prominent symptoms. Many affected children develop an ID. Persons with Dravet syndrome very often show hallucinations. Martin and collaborators [24] detected hallucinations in 10 out of 41 patients with this syndrome. In some other cases, they assumed the presence of hallucinations basing only on their behavioral and observable aspects.

The 22q11.2 deletion syndrome (synonyms: velo-cardio-facial syndrome, DiGeorge syndrome, Shprintzen syndrome) [25] is associated with ID and with a broad spectrum of physical and psychiatric manifestations [26]. Biswas and Furniss [27] assumed that some of the cognitive difficulties in persons with this syndrome represent risk factors for hallucinations and paranoid delusions often emerging in these persons.

Homocystinuria, a very rare genetic disorder, comprises various forms with different clinical signs and genetic causes. Frequent symptoms are ectopia lentis, myopia, skel-

etal anomalies, and thromboembolic events [28]. Sometimes, homocystinuria includes ID. Acute psychosis is a rare symptom in this disease. Colafrancesco and collaborators described an affected 17-year-old girl presenting visual hallucinations, behavioral perseverance, psychomotor hyperactivity, and affective inappropriateness [29].

17q24.2 microdeletions are a rare genetic condition, which presents with ID, delayed speech development, truncal obesity, epileptic seizures, hearing loss, and a particular physiognomy. Vergult and collaborators [30] reported hallucinations and mood swings in two out of four persons with this condition.

Malm and colleagues [31] described a group of persons with alpha-mannosidosis, a very rare genetic syndrome [32], with psychiatric symptoms including hallucinations.

Organic hallucinations have been reported in some genetic syndromes including ID such as Dravet syndrome, DiGeorge syndrome, homocystinuria, 17q24.2 microdeletion syndrome, and alpha-mannosidosis. In persons with ID and/or ASD, organic hallucinations may also occur in case of substance dependence, tumors or other lesions of the temporal lobe, Parkinson's disease, epilepsy, and anticholinergic or dopaminergic treatments.

29.2.2 Organic Catatonic Disorder

General Description

The main features of organic catatonic disorder are represented by diminished or increased psychomotor activities and catatonic symptoms (e.g., stereotypies, waxy flexibility, and impulsive acts). The extremes of psychomotor disturbance may alternate. Other catatonic phenomena, such as stereotypies, waxy flexibility, or impulsive acts, increase confidence in the diagnosis of this disorder.

There is some terminological confusion between the terms catatonia or catatonic syndrome and catatonic symptoms in the scientific literature. However, to date organic catatonic disorder or catatonic symptomatology is recognized to occur with a broad spectrum of medical and psychiatric illnesses [33]. Infectious, paraneoplastic, or autoimmune encephalitis and carbon monoxide poisoning may be etiological factors of catatonic symptoms [34, 35]. In addition, metabolic disorders, endocrine conditions, and intoxications may result in an organic catatonic disorder [36, 37]. In most cases, catatonic symptoms are part of a broader array of psychiatric symptoms.

Kiani and colleagues [38] published two cases of anti-N-methyl-d-aspartate (NMDA) receptor encephalitis presenting with neuropsychiatric symptoms of catatonia and neuroleptic malignant syndrome in two patients with ID and ASD.

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In a study on the aspects of catatonia through the copy number variation analysis, Breckpot and collaborators [39] investigated persons with ID and catatonic symptoms. Psychiatric diagnoses in these patients were ASD, psychotic or mood disorders. The authors identified copy number variants, 22q13.3 deletions and 14q11.2 duplications, as relevant causes.

Serret and collaborators [40] presented two patients with a similar psychiatric and genetic diagnosis as well as similar histories and evolutions. They were diagnosed with ASD in childhood and showed regression with catatonic features and behavioral disorders after stressful events during adolescence. Both patients had mutations or microdeletions of the SHANK3 gene. Only lithium therapy reversed clinical regression, whereas other psychopharmacological therapies failed. These observations emphasize the relevance of genetic investigations with respect to effective therapeutic options.

Catatonia has also been described in a female adolescent with velo-cardio-facial syndrome (VCFS). The authors indicate catatonia to be relatively common in persons with VCFS and treatment-refractory psychiatric manifestations [41].

Winarni and collaborators published a case report on a 25-year-old fragile X syndrome premutation carrier who developed ASD, psychosis, and catatonia [42].

Organic catatonic disorder has been reported in persons with velo-cardio-facial syndrome and fragile X syndrome as well as in persons with copy number variations of 22q13.3, 14q11.2, and SHANK3 genes. In persons with ID and/or ASD, organic catatonic disorder or catatonic symptoms may also occur in the course of infectious, paraneoplastic, or autoimmune encephalitis as well as metabolic disorders, endocrine conditions, and intoxications.

29.2.3 Organic Delusional [Schizophrenia-Like] Disorder

29

General Description

Persistent or recurrent delusions dominate the clinical picture of the organic delusional disorder. Hallucinations may accompany the delusions. However, the delusions are not confined to the content of the hallucinations. Schizophrenia-like symptoms, such as bizarre delusions, hallucinations, catatonic symptoms, or thought disorders, may also be present.

Organic delusional disorders share some etiological factors with organic catatonic disorders. Therefore, delusions are often part of a broader spectrum of mental symptoms in medical conditions. Cocaine, alcohol, amphetamines, mechanical trauma, and inflammatory cerebral disease may lead to organic delusional disorders; temporal lobe epilepsies are also often associated with organic delusional disorders [37].

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Prader-Willi syndrome (PWS) is a condition caused by the lack of expression of genes on the paternally inherited chromosome 15q11.2-q13 region [43]. There are three genetic subtypes: paternal 15q11-q13 deletion, maternal uniparental disomy 15, and imprinting defect. Young adults with PWS show high rates of physical diseases and psychiatric disorders, which include delusions [44].

The 2q37 microdeletion syndrome is a complex genetic disorder consisting of diverse mental (including ID and ASD) and somatic symptoms [45]. Lally and collaborators described a 21-year-old woman with moderate ID showing nihilistic delusions, incessant pressure of speech, and worsening of sleep and eating [46]. Other authors reported on a person with 2q37 microdeletion syndrome presenting schizophrenic psychosis including delusion, in line with previous findings of an association between schizophrenia susceptibility and unknown genes within the chromosomal region 2q37 [47].

Recent genetic studies are increasingly indicating vulnerability to psychotic disorders to have a polygenic origin, with a complex array of contributing risk loci across the allelic frequency spectrum that are shared with ID and ASD as well as bipolar disorder and major depressive disorder [48, 49].

Organic delusional disorder has been reported in Prader-Willi syndrome and 2q37 microdeletion syndrome. Cocaine, alcohol, amphetamines, mechanical trauma, temporal lobe epilepsies, and inflammatory cerebral disease may also lead to organic delusional disorders. Delusions are often part of a broader spectrum of mental symptoms in many medical conditions.

29.2.4 Organic Mood [Affective] Disorder

General Description

Organic mood disorders or organic affective disorders are characterized by changes in mood (depressed or elevated), usually accompanied by changes in the level of activity (increased or decreased). Organic mood disorders are often triggered due to endocrine disorders like hypothyroidism, hyperthyroidism, Cushing's syndrome, and Addison's disease [50]. Some drugs (e.g., clonidine, reserpine, beta-blockers, or antibiotics) may act as causes of organic depres-

sions, whereas other medications (e.g., corticosteroids, levodopa) used to treat cerebrovascular disorders, cerebral tumors, or hyperglycemic states in diabetes mellitus may result in organic mania [37].

Rundell and Wise [51] analyzed the records of 755 patients seen by consultation-liaison psychiatrists in a general hospital and found that 87% of manic patients and 38% of depressed patients had organic mood disorders. The most frequent precipitants of organic mania were corticosteroids, HIV infection, and epilepsy, while the most frequent precipitants of organic depression were stroke, Parkinson's disease, and HIV infection.

▪ Reports on Persons with Intellectual Disability and/or Autism Spectrum Disorders

There are many studies describing affective disorders in persons with ID in general. However, convincing studies on etiologically specific syndromes are very scarce. Rodriguez-Revenga and collaborators found fragile-X premutation carrying mothers of children with fragile X syndrome to have a higher tendency to depression than mothers of children with ID but without fragile X syndrome and mothers from the general population. The authors conclude that this premutation could represent a genetic risk factor for mood disorders, both primary and secondary to physical disorders or diseases [52].

- Organic mood disorders can be determined by endocrine disorders like hypo- or hyperthyroidism, Cushing's syndrome, Addison's disease, and hyperglycemic states in diabetes mellitus. A broad spectrum of drugs, cerebrovascular disorders, and cerebral tumors are other frequent causes.

- High vulnerability to primary and secondary depressive disorders has been found in fragile-X premutation carrying mothers of children with fragile X syndrome.

29.2.5 Organic Anxiety Disorder

▪ General Description

The organic anxiety disorder is characterized by the essential features of a generalized anxiety disorder or a panic disorder or a combination of both, but arising as a consequence of an organic disorder capable of causing cerebral dysfunction (e.g., temporal lobe epilepsy, thyrotoxicosis, or pheochromocytoma) [53]. Organic anxiety disorder and organic affective disorder share many clinical aspects and represent the most common organic mental disorders [37]. Wise and Rundell emphasized the frequent occurrence of pathological anxiety in neurological diseases [54].

▪ Reports on Persons with Intellectual Disability and/or Autism Spectrum Disorders

The prevalence of anxiety disorders in persons with ID and/or ASD is high, with generalized anxiety disorder being the most common, followed by agoraphobia [55].

Fragile X syndrome is a clinical condition caused by mutations in the FMR1 gene of the X chromosome, which includes ID, ASD, and many physical problems [56]. Cordeiro and collaborators assessed the prevalence of anxiety disorders in 58 males and 39 females with fragile X syndrome. Despite a high rate of psychopharmacological treatment, 86.2% of males and 76.9% of females showed anxiety disorder, particularly social phobia and specific phobia. The authors concluded that the mutation of the FMR1 gene is associated with a high risk of anxiety disorders [57]. The importance of anxiety disorders in the complex physical and mental vulnerability of persons with fragile X syndrome has been identified or emphasized also by other researchers [58, 59].

Women with FMR1 premutation who are mothers of children with fragile X syndrome have also been found to present high rate of pathological anxiety associated with early ovarian aging and predicted in severity by polymorphisms of the corticotropin releasing

hormone receptor 1 (CRHR1) gene, which associate with differences in hypothalamic-pituitary-adrenal (HPA) axis function [60, 61].

Persons with 7q11.23 duplication syndrome show ID, ASD, distinctive physiognomic features, cardiovascular disease, hypotonia, adventitious movements, gait abnormalities, speech sound disorders, behavioral problems, anxiety disorders, selective mutism, attention-deficit/hyperactivity disorders, oppositional disorders, and physically aggressive behavior [62]. In children with 7q11.23 duplication syndrome, anxiety disorders are common, especially social phobia, specific phobia, selective mutism, and separation anxiety disorder.

Williams syndrome is a microdeletion syndrome due to deletion of genetic material from the region q11.23 of one of the two chromosomes 7. The clinical pictures are characterized by a broad spectrum of mental and physical features. Anxiety and specific phobias are common symptoms in Williams syndrome [63]. The data from a systematic literature review on Williams syndrome [64] revealed that the affected individuals were four times more likely to experience anxiety than individuals with ID due to other causes.

Rett syndrome [65] is a rare genetic syndrome, occurring almost exclusively in girls. The Rett syndrome affects the daily activities. Repetitive hand movements are typical behavioral symptoms of Rett syndrome. Based on comprehensive investigations of anxious behavior in 74 girls with Rett syndrome, Barnes and colleagues [66] interpreted anxiety-like behavior as a prominent component of the behavioral phenotype in Rett syndrome, affecting predominantly children with less severe neurologic impairment.

Treadwell-Deering and collaborators [67] found in the majority of patients with Potocki-Lupski syndrome (duplication 17p11.2) [68] besides ID, many behavioral difficulties such as withdrawal, anxiety, and inattention. Many patients presented autistic symptoms.

Organic anxiety disorder has been reported in some genetic syndromes including ID and/or ASD such as fragile X syndrome, 7q11.23 duplication syndrome, Williams syndrome, and Rett syndrome. In persons with ID and/or ASD, organic anxiety disorder may also occur in the case of temporal lobe epilepsy, thyrotoxicosis, or pheochromocytoma.

29.2.6 Organic Dissociative Disorder

General Description

The organic dissociative disorder has to meet the criteria for one of the conversion disorders. In addition, general criteria for organic etiology are also fulfilled. This diagnostic category is very seldom used. In addition, its nosological position is controversial.

Reports on Persons with Intellectual Disability and/or Autism Spectrum Disorders

There is no information in the scientific literature regarding the occurrence of organic dissociative disorders in persons with ID and/or ASD.

29.2.7 Organic Emotionally Labile [Asthenic] Disorder

General Description

Organic emotionally labile disorder (alternatively: organic asthenic disorder) is characterized by marked and persistent emotional incontinence or lability, fatigability, or a variety of unpleasant physical sensations (e.g., dizziness) and pains regarded as being due to the presence of an organic disorder. Various etiological factors may play a role, for example, cerebrovascular diseases, trauma, and encephalitis [37].

Reports on Persons with Intellectual Disability and/or Autism Spectrum Disorders

The literature contains no information concerning specific occurrence of organic emotionally labile disorder in persons with intellectual disability and/or autism spectrum disorder.

29.2.8 Organic Personality Disorder

General Description

Organic personality disorder is characterized by significant alterations of the habitual patterns compared with the premorbid behavior. In particular, the emotions, needs, and impulses are affected. In addition, cognitive functions in the areas of planning and anticipating probable personal and social consequences may be influenced, for example, in the so-called frontal lobe syndrome. There is a spectrum of etiological factors, mainly primary brain damages or dysfunctions. In Parkinson's disease or in multiple sclerosis, organic personality changes may occur, too [37, 69].

Reports on Persons with Intellectual Disability and/or Autism Spectrum Disorders

There is no specific literature on organic personality disorders in persons with intellectual disability and/or autism spectrum disorders. Of course, some aspects of behavior in persons with intellectual disability due to brain lesions could be interpreted as components of an organic personality disorder.

29.2.9 Postencephalitic Syndrome

General Description

The etiological basis of the postencephalitic syndrome is an inflammatory disease of the brain. Infectious agents cause most cases. The disorder includes residual behavioral change after recovery from encephalitis. The spectrum of nonspecific symptoms depends on the infectious agent, on individual dispositions, and on the age of the individual at the time of infection. Unlike the organic personality disorder, the postencephalitic syndrome is often reversible.

With respect to this syndrome, two aspects are important. First, encephalitis during childhood may be a relatively rare cause of intellectual disability or autism spectrum disorder. Any person can get an encephalitis. Of course, persons with intellectual disability and autism can be affected too.

Reports on Persons with Intellectual Disability and/or Autism Spectrum Disorders

There are no relevant reports on postencephalitic syndromes in intellectual disability or autism spectrum disorders.

29.2.10 Post-Concussion Syndrome

General Description

The post-concussion syndrome is caused by mechanical brain trauma, for example, in traffic accidents or by a fall during an epileptic seizure. Its clinical phenomenology is heterogeneous. It may consist of a number of symptoms and complaints such as headache, dizziness, fatigue, irritability, concentration problems, difficulty in performing mental tasks, memory impairment, insomnia, reduced stress tolerance, emotional excitement, or alcohol. Sometimes, these symptoms and complaints are accompanied by depression or anxiety, resulting from the loss of self-esteem and fear of permanent brain damage. With respect to the last-mentioned symptoms, organic as well as psychological factors may play a role.

Of course, brain trauma are accidental events without any specific relation to intellectual disability or autism.

Reports on Persons with Intellectual Disability and/or Autism Spectrum Disorders

There is no significant literature regarding post-concussion syndrome in intellectual disability or autism spectrum disorder.

29.3 Conclusive Considerations

The field of organic mental disorders is highly complex and connected with many open questions. Some aspects, especially aspects of epidemiology or the relations between certain

clinical pictures and specific etiogical factors, are unclear. In principle, persons with ID and/or ASD are confronted with a higher risk for organic mental disorders than the general population. Moreover, for some specific etiological conditions, specific risks of organic mental disorders are known and can be identified. Mainly genetically caused syndromes play an outstanding role in this field. It is to be expected that knowledge in this area will increase over the next years and decades.

29.4 Prognosis

The prognosis for organic mental disorders is not determined by the specific psychiatric manifestation, but by the underlying mechanism, its natural and therapeutically modified course.

29.5 Therapy

The therapeutic options are twofold. The most important option is the so-called causal therapy, adequately treating the underlying medical condition, if possible. Should the causal treatment of the underlying medical condition be impossible or insufficient, a symptomatic therapy of mental symptoms by psychotropics (e.g., neuroleptics, antidepressant, and anxiolytics) is necessary. Particularly in the case of epilepsy-related organic mental disorders, certain psychotropic medications (e.g., certain neuroleptics or antidepressants) may be contraindicated. Some organic hallucinations can represent an adverse event of anticholinergic or dopaminergic substances. Therefore, options of targeted antiepileptic treatment are to be considered. In other cases, the recognition of the genetic background of mental symptoms may be helpful for an adequate therapeutic decision.

For organic hallucinations, treatment with antiepileptic drugs is often the first choice.

Due to multimorbidity and frailty, persons with and/or ASD are confronted with a higher risk for organic mental disorders than the general population. Some specific genetic syndromes are associated with specific risks.

The most important therapeutic option is the treatment of underlying medical condition, when impossible or insufficient, a symptomatic therapy of mental symptoms by psychotropic drugs is necessary.

29.6 Epilepsy in Intellectual Disability and Autism Spectrum Disorder

29.6.1 Introduction

Approximately 50 million people worldwide are estimated to suffer from epilepsy [70]. The average incidence rate of unprovoked recurrent epileptic seizures in children has been reported from 35/100,000/year [71] to 88.8/100,000/year [72]. The cumulative incidence rate of epilepsy by the age of 15 years is about 0.8% [73]. The average prevalence in developed countries is between 3.5 and 5 per 1000 children; the incidence and prevalence of epilepsy are higher in developing countries [74]. Comorbidities, defined as disorders coexisting with or preceding epilepsy, or else compounded or directly attributed to epilepsy or to its treatment, are common, especially parasitic and infectious diseases (44%), somatic comorbidities (37%), and psychiatric/psychosocial problems (19%), of which neurocysticercosis, head trauma, malnutrition, and stroke are the most frequently studied worldwide [75].

According to the most recent Classification of the Epilepsies proposed by the International League Against Epilepsy (ILAE), seizure types, epilepsy types (focal, generalized, combined generalized and focal, unknown), and epilepsy syndromes should be defined at a diagnostic level. Furthermore, an etiologic diagnosis should be considered, taking in count at least six different groups: structural, genetic, infectious, metabolic, immune, and unknown causes [76].

Prognosis for patients with epilepsy is usually good, but up to 30% of them present are drug-resistant to adequate treatments with antiepileptic drugs [77]. Furthermore, co-occurrence of epilepsy with intellectual disability (ID) or autism spectrum disorder (ASD) makes prognosis less favorable.

29.6.2 Epilepsy and ID

There are several biases that limit the epidemiological estimate of epilepsy in the population of patients with ID: difficulties in reporting their seizures; sensory impairments, which may restrict communication and, thus, self-report of seizures; changing of caretakers over time, thereby creating communication problems; different inclusion criteria which may select different subpopulations; occurrence of subclinical seizures or nonepileptic events.

However, in the "institutional group" of subjects with ID, the prevalence of epilepsy has been estimated as 33.1%, and in the "community group" 16.1% [11]. Other authors reported even higher prevalence rates, from 32% [8], to 40% [10], or 65% [9], in institutional settings.

The prevalence of epilepsy seems not to be constant over the life span and varies with etiology: it increases from age 20 to >60 years for those without ID and for those with Down syndrome or ASD; the prevalence of epilepsy declines for other adults with ID, cerebral palsy, or a combination of these comorbidities [78].

In a meta-analysis by Robertson and colleagues [79], in general samples of people with ID, the pooled estimate from 38 studies was 22.2%. Prevalence increased with increasing level of ID.

The relationship between epilepsy and cognition is hypothesized as a two-way condition in which cognitive factors and epilepsy interact, by influencing the individual's development [80]. Some studies report cognitive impairments prior to the onset of seizures that could, along with the onset of epilepsy, be explained by a common factor determining both conditions [81, 82].

Many factors correlated with epilepsy may determine cognitive dysfunction, such as etiology, seizure activity (onset, type, duration, and frequency), type of electroencephalogram (EEG) abnormalities, neurological side effects of antiepileptic drugs (AEDs), concomitant disability and neurophysiological impairment, environmental factors and parental attitudes, emotional and personality characteristics [83, 84].

The age of onset. Several authors agree that epilepsy severity depends on early seizure onset [85–88].

A longer duration of seizures (longer disease/epilepsy duration) leads to increased probabilities of developing cognitive deficits [89–91]; vice versa, the effects of active persistent seizures do not appear completely clear [92].

Seizure frequency represents an increased risk of developing cognitive problems, too [93–95].

As for memory processes, nonhomogeneous results were found in the comparison studies between "Localization vs generalized" [96–98].

The effects of intercritical EEGraphic activity (EEG findings: focal, generalized, normal) on cognitive development are the subject of debate [99, 100]. Japaridze and colleagues [101] compared two groups of patients with and without ID, both with pathologic intercritical EEGraphic activity. The study did not show significant neuropsychological impacts either on the two groups, or on the control group, except for an increased difficulty in the ID sample with regard to reaction times, selective attention, and cognitive flexibility.

Furthermore, the use of a large number of AEDs is a significant predictor of cognitive performance in samples of children with epilepsy [95]. Thompson and collaborators [102] underlined how the side effects of AEDs are often underestimated in populations with ID and epilepsy. According to Kerr and colleagues [103], most of this population has difficulty in self-reporting their health and quality of life, and in fact the most significant side effects (such as vomiting and weight loss) are reported by caregivers. Studies – performed in English until 2017 – on the impact of AEDs and side effects in adults with ID

and epilepsy have been collected in a review by Copeland and colleagues [104]. The authors concluded that it is more complex to evaluate AEDs' side effects in people with ID because they present with greater comorbidities (e.g., ASD) and it is more difficult to get feedback directly from patients. In order to assess the efficacy of AEDs in this population, Kerr and colleagues [103], while drafting the guidelines for the management of adults with epilepsy and ID, underline that both standardized cognitive and behavioral assessment procedures are needed in baseline as well as in postpharmacological treatment.

Furthermore, there are some specific childhood epileptic conditions, classified as epileptic encephalopathies, usually resistant to antiepileptic treatment, in which the epileptiform abnormalities may contribute to progressive neuropsychological dysfunction and ID: early myoclonic encephalopathy, Ohtahara syndrome, West syndrome, Dravet syndrome (previously known as severe myoclonic epilepsy in infancy), myoclonic status in nonprogressive encephalopathies, Lennox-Gastaut syndrome, Landau-Kleffner syndrome, and epilepsy with continuous spike-waves during slow-wave sleep [105–109].

For these reasons, and on the basis of our experience, diagnostic workup of patients with ID and epilepsy should have a multidisciplinary approach based on the clinical history, with an emphasis on witness description of seizures; physical and neurological examinations; dysmorphology evaluation; assessment of the individual's level of ability; psychiatric examination and tests; video-EEG or ambulatory EEG; MRI, often with sedation or general anesthesia, because subjects with ID may have difficulties in cooperating; blood AED monitoring; metabolic investigations; genetic analyses (i.e., karyotype, FISH, MLPA, array-CGH, gene sequencing, next-generation sequencing, and whole exome sequencing); and other specific investigations.

Moreover, the assessment of cognitive and neuropsychological processes is very important, through the use of appropriate scales such as Wechsler, Leiter, Nepsy, etc., as well as the detection of social and practical adaptive-conceptual skills by means of scales like Vineland or Adaptive Behavior Assessment System (ABAS).

Prognosis of epilepsy in patients with ID appears obviously correlated with the underlying etiology [110, 111]. However, some studies have demonstrated that outcome in this special population is not so unfavorable as one can believe. In a residential group of 215 subjects with ID and epilepsy, over a 5-year period, 18% became seizure-free on one or two drugs; of these, 60% were on monotherapy; of the group remaining uncontrolled, most were improved and only 8% required three drugs to get optimal control. Twenty-two percent of subjects were followed without recurrence once off medication for intervals between 1 and 20 years [9].

29.6.3 Epilepsy, ID, and Psychopathology

The connection between epilepsy and psychiatric disorders is complex and still not fully clarified, determining frequent issues of overlapping symptoms and mistaken diagnoses in clinical practice, especially in persons with ID and/or ASD. Some symptoms of seizures, especially focal seizure, such as altered consciousness or feeling of unreality, are also frequently found in some psychiatric disorders, conversely some psychiatric symptoms may also resemble epilepsy and some psychopathological conditions (e.g., conversion disorder) can produce symptoms that are quite similar to those of generalized seizures that are called psychogenic nonepileptic seizures (PNES). Furthermore, persons with true epilepsy may also have PNES, especially persons with ID and/or ASD [112]. Diagnostic errors between epilepsy and PNES need careful consideration also when reviewing studies showing associations between psychiatric disorders and epilepsy or indicating lower need of seizure control in epileptic patients with co-occurrence of psychiatric conditions [113]. A history of emotional disturbance or a pattern of inadequate coping may help in making the distinction, although continuous electroencephalogram (EEG) and video

monitoring are the best methods to achieve a correct diagnosis.

In the general population, 20–30% of people with epilepsy have psychopathological symptoms that require treatment, especially in pharmacoresistant cases [114–116]. Anxiety, especially agoraphobia, and depression are the most common conditions, with an important role in the premature mortality evidenced in epileptic patients, which includes suicide rate that is reported to be four-to-five times higher than in healthy population [117]. Adults with a history of seizures are two-to-three times more likely than average to develop a psychotic disorder and a history of attention deficit hyperactivity disorder is two and one-half times more common than average in children with seizures [118–120].

A continuum between emotional reaction to persistent seizures and psychopathological features is observable in many clinical cases. For example, long-lasting frustration and discouragement can lead to depression as well as increasing fear of having a seizure in public can give rise to agoraphobia. Nevertheless, there is also evidence that brain alterations and dysfunctions determining seizures are also responsible for significant changes of psychic functioning, especially of mood and thought. Two psychopathological conditions called interictal dysphoric disorder and interictal behavior syndrome occur in some patients with epilepsy [121, 122]. The former includes depression (or more rarely euphoria), lack of energy, pain, irritability, anxiety, and fear, while the latter comprises sadness, humorlessness, interest reduction (including sex), obsession with details, compulsive writing or scribbling, anger, and social withdrawal [123, 124].

The chronological relationship between psychiatric disorders and epilepsy is yet to be cleared, although some findings indicating unprovoked seizures (not caused by brain disease or injury) to follow psychiatric conditions, especially major depressive disorder. Through the twentieth century, many researchers in the field of psychiatry described an "epileptic personality," with a specific reference to affective viscosity (the tendency to prolong interactions with others), slowness in thinking and acting, and dramatic features, including PNES, as a way to gain others' attention [125, 126]. Within ICD-11, this condition is comprised in the chapter of secondary personality change, which refers to "syndromes characterized by a persistent personality disturbance that represents a change from the individual's previous characteristic personality pattern" and "is judged to be a direct pathophysiological consequence of a health condition not classified under mental and behavioral disorders, based on evidence from the history, physical examination, or laboratory findings." "This category should be used in addition to the diagnosis for the presumed underlying disorder or disease when the personality symptoms are sufficiently severe to warrant specific clinical attention" [1].

Besides, many psychoactive drugs are prescribed for both epilepsy and psychiatric disorders. For example, antiepileptic drugs are used in the treatment of mood disorders and benzodiazepines are used for anxiety disorders. Comorbidity between psychiatric disorders and epilepsy is particularly frequent and complex in persons with ID [127–133]. The question of whether individuals with epilepsy and ID have a higher rate of psychopathological comorbidity than people with only epilepsy or ID has been examined in several studies. The results of such researches have highlighted conflicting data.

Some results indicate that the most severe forms of epilepsy, including generalized seizures characterized by higher severity, frequency, and number of episodes, represent risk factors for the development of behavioral problems and psychopathological disorders [134–136].

A significant association between the level of ID severity and the presence of psychiatric disorders, especially ASD and psychosis, in subjects with epilepsy has been identified [129–136]. In particular, the level of ID seems to be linked to the presence of behavioral problems, the more severe the ID, the more the behavioral problems that people with epilepsy show [134, 137]. In contrast, other authors have found no differences regarding the presence of psychological disorders between people presenting only with epilepsy or ID, or affected by both conditions [138, 139].

The results of a review conducted by Jans and collaborators [140] show that the presence of epilepsy does not represent a clear determinant of neuropsychiatric comorbidities in patients with ID, although a tendency toward negative mood symptoms is identified. Other studies found epilepsy to represent a risk factor for self-injurious behavior, either at short- or long-term follow-up [141, 142].

29.6.4 Epilepsy and ASD

A great amount of epidemiological data has been produced on association between ASD and epilepsy. Prevalence of epilepsy in the population of patients with ASD varies from approximately 2.4–46% in different studies [143–145]. These variable figures substantially depend on several factors, such as the study design (i.e., retrospective or prospective), the clinical setting (i.e., primary, secondary, or tertiary centers), the definition of autism which changed over time according to the editions of DSM, the inclusion criteria for epilepsy (i.e., one single seizure, two seizures, febrile or nonfebrile), the age at the recruitment, and the presence of ID or other comorbidities (i.e., cerebral palsy).

The prevalence of epilepsy in the different forms of autistic syndromes, as classified at the time of DSM-IV TR [146], has been estimated: more than one third of cases in autistic disorder, 5–10% in Asperger syndrome, about 31% in pervasive developmental disorders not otherwise specified, 70% in childhood disintegrative disorder, and 90% in Rett syndrome [147].

On the other hand, the prevalence of ASD in populations of patients with epilepsy has been reported from 5% to 21% [129, 148]. The reported rates of epilepsy in all of these studies are greater than general population prevalence estimates and raise the possibility of a direct association between epilepsy and ASD [149].

Familiarity for epilepsy is associated with an increased risk to present epilepsy in subjects with ASD [150]. Furthermore, the analysis of multiplex ASD families showed an epilepsy rate of 12.8% in children with ASD versus a rate of 2.3% in the siblings without ASD. This would suggest a genetic predisposition not only for ASD, but also for epilepsy [151].

The risk to present epilepsy in subjects with ASD seems higher in females than in males (respectively, 34.5% and 18.5%) [152, 153]. On the other hand, some studies did not find any difference between gender and epilepsy prevalence [144, 154, 155].

Other studies showed that people with idiopathic ASD seem to have a lower risk of developing epilepsy than those with syndromic ASD or concomitant neurological conditions [156, 157].

Another relevant risk factor for epilepsy is represented by ID. In fact, prevalence of epilepsy is 21.4% in patients with ASD and ID, and 8% in those with only ASD [153].

Epileptiform abnormalities are also reported as most likely in children with low-functioning autism [158] or with more severe forms of autism [159, 160].

A meta-analysis performed in 2012, including sole follow-up studies, reported rates of 23.7% in patients with ASD and ID, and 8% in patients with ASD without ID [161], while little is known about the relationship between epilepsy and ASD in the absence of ID [162].

The age at onset of seizures has a bimodal distribution in ASD, with a first peak during childhood and a second during adolescence. However, approximately 20% of patients with ASD present their first seizure after the age of 20 [163].

Evaluation and classification of seizures in ASD may become difficult, because avoiding of eye contact, or motor block, typical of ASD, may be confused with absences, and generalized or segmental stereotyped movements may be interpreted as motor features of focal and generalized seizures. In a continuous video-EEG telemetry study performed at a tertiary care referral center, 32 children with ASD were monitored. Of these patients, 22 were primarily referred for seizure evaluation and 15 of them had recorded events which were nonepileptic seizures; the other 7 patients had no recorded events. Interestingly, 11 (73%) of the 15 patients with nonepileptic events had interictal epileptiform EEG abnormalities [164]. There are not peculiar epileptic seizures or syndromes which are

prevalent in the population of patients with ASD [152]. Focal seizures with unawareness are more frequent of the generalized ones (respectively, 73% and 27%) [165]. Among the special epileptic syndromes which have been reported in association with ASD, there are febrile seizures, benign childhood epilepsy with centro-temporal spikes, West syndrome, Lennox-Gastaut syndrome, and Landau-Kleffner syndrome [166]. Recently, typical behavioral characteristics of ASD have been described in 18/30 patients (60%) with Dravet syndrome [167].

Also, interictal EEG pattern is not homogeneous in ASD. Paroxysmal abnormalities are mostly focal, more frequently localized over the frontal, temporal, or centro-temporal regions [166].

The presence of epilepsy in people with ASD may be determined by several structural alterations, genetic conditions, or metabolic dysfunctions, known to play a role in the emergence of both epilepsy and autism. Genetic etiologies include isodicentric chromosome 15 syndrome, Phelan-McDermid syndrome, fragile X syndrome, tuberous sclerosis complex, Rett syndrome, encephalopathies due to mutations of PTEN, CDKL5, FOXG1, MEF2C, CASK, and SCN2A genes [168, 169]. Metabolic conditions, such as mitochondrial diseases, abnormalities in cerebral folate metabolism, disorders of creatine, cholesterol, pyridoxine, biotin, carnitine, γ-aminobutyric acid, purine, pyrimidine, and amino acid metabolism and urea cycle disorders, have also been associated with ASD and epilepsy [170].

The pathophysiology of epilepsy in ASD has not been completely clarified. Three theories, not mutually exclusive, have been proposed to explain the relationships between brain development, epilepsy, and ASD. First, ASD and epilepsy might represent distinct conditions without causal relationship. Second, a common neurobiological dysfunction might lead to abnormal brain development that results in both epilepsy and ASD. Third, epilepsy could lead to ASD or conversely, abnormal brain circuitry underlying ASD could predispose the brain to seizures [171]. More recently, on the basis of neurophysiological and neuroimaging studies (diffusion MRI), the "underconnectivity" model has been proposed, hypothesizing that ASD and epilepsy might be the result of a diffuse dysfunction of neural networks involving different cortical and subcortical regions [172].

Concerning the diagnostic workup for epilepsy, the same recommendations formulated for patients with ID are valid also for those with ASD (see above).

We would like to stress here the importance of video-EEG and sleep EEG for a correct interpretation and classification of interictal and ictal events. Recording a good quality EEG is often an arduous task in patients with ASD who may not tolerate the procedure, and sometimes drug sedation (i.e., with promazine) is needed to minimize artifacts arising from movement in these patients.

According to a meta-analysis, in the absence of clinical seizures, children with ASD and developmental regression have EEG findings similar to those with nonregressive autism. Therefore, no evidence suggests to perform an EEG to rule out underlying subclinical epilepsy in a child with ASD and developmental regression without clinical seizures [173].

As for the consequences of epilepsy on the functioning of people with ASD, the DSM-5 [174], points out that in this population, the presence of epilepsy is associated with a higher severity of ID and a lower verbal communication skill.

In 2015, Shubrata and collaborators observed that subjects with ASD and epilepsy presented with a more severe form of ASD [175].

In a study carried out by Matson and colleagues [176], children with seizure disorders achieved significantly lower scores in the cognitive, personal-social, and communication areas than children without convulsive disorders.

Greater deficits in adaptive functioning and social skills in people with ASD and epilepsy have also been reported, compared to those with only ASD [158, 176–179].

Other studies have not shown differences in behaviors or severity of autistic characteristics in individuals with ASD and epileptiform abnormalities, although they have been found

in subjects with ASD and convulsions, major behavioral disorders, stereotypies, hyperactivity and irritability, sleep disorders, and attention deficit [159, 180, 181].

In a study performed by Smith and Matson [178], it was found that the adult ASD group showed a higher number of self-injurious behaviors than those with ID, and the additional diagnosis of epilepsy did not add to this. Those with ASD and epilepsy were more impaired on measures of disruptive behavior than those with ID alone, ASD alone, or epilepsy alone.

These results suggest that most of the effects of epilepsy on the behavioral phenotype of people with ASD are due to the presence of ID [182, 183].

Ko and collaborators [184], using the Social Responsiveness Scale (SRS), showed that ASD participants with epilepsy were significantly more impaired than ASD participants on some measures of social functioning such as social awareness and social communication. Participants were more impaired than ASD participants on social security measures.

Also for subjects with ASD, the outcome of epilepsy strictly depends on the related etiology; however, in the cases of epilepsy without demonstrable etiology prognosis can be rather favorable [152]. Data about epilepsy outcome are not univocal. While some studies have reported a good control of seizures with one or two AEDs [163, 185], others have observed pharmacoresistance in about a third of cases [186, 187].

The association between subclinical epileptiform EEG abnormalities and autistic regression is not significant, except for some epileptic encephalopathies, such as electric status epilepticus in sleep or Landau-Kleffner syndrome, which may determine psychomotor and behavioral regression [166, 188].

However, there are clear evidences, based on long-term analyses, that epilepsy has a relevant impact on behavioral, psychomotor, and cognitive outcome of subjects with ASD, determining disturbances in gross and fine motor skills, in daily life activities, and in many autistic behaviors [177].

The prevalence of epilepsy is higher in people with ID and/or ASD. Etiology is a very relevant factor for prognosis and treatment of epilepsy in these special populations. Epileptic encephalopathies strongly influence cognitive functions and adaptive behavior in persons with ID and/or ASD.

29.6.5 Pharmacological Treatment for ID and ASD

The right choice of an AED for the patients with ID or ASD and epilepsy often represents a serious challenge. Evidence-based recommendations or guidelines concerning the antiepileptic treatment in this special population are not yet available. When the treatment has been started, the EEG, clinical, and neuropsychological picture of the patient should be periodically reevaluated [189].

The choice of the most appropriate AED is driven by the type of epilepsy and tolerability, which cannot be expected a priori, and should be evaluated case by case. Some AEDs, such as phenobarbitone, ethosuximide, and vigabatrin, may worsen the behavioral disturbances. Other AEDs may present negative pharmacokinetic or pharmacodynamic interactions with the psychotropic drugs used for the behavioral disturbances often presented by subjects with ID or ASD. Regarding pharmacokinetic interactions, carbamazepine reduces plasma concentrations of clozapine, olanzapine, risperidone, quetiapine, and ziprasidone; valproic acid reduces the plasma concentration of aripiprazole, and increases that of quetiapine; phenobarbitone and phenytoin, respectively, reduce plasma levels of clozapine and quetiapine [190]. An important pharmacodynamic interaction occurs between carbamazepine and clozapine, potentiating hematologic side effects, such as thrombocythemia; the associations valproate-clozapine or valproate-olanzapine may show additive side effects, such as weight increase, sedation, and neurotoxicity; carbamazepine and lithium may determine neurotoxicity; valproate and lithium may cause weight increase,

sedation, tremor, and gastrointestinal disturbances [166].

In an open observational add-on study design carried out on 46 patients with severe drug-resistant epilepsy and different degrees of ID, levetiracetam became an effective and generally well-tolerated drug [191]. In another retrospective evaluation of 32 inpatients with drug-resistant epilepsy and ID, pregabalin was only moderately effective [192].

Furthermore, some AEDs have a psychotropic activity and are helpful in the management of behavioral disturbances: valproic acid [193], topiramate [194], and lamotrigine [195].

In a series of 85 patients out of a resident population with epilepsy and ID, an attempt was made to taper barbiturates in order to improve the cognitive and psychological state, and to reduce polypharmacy while avoiding seizure exacerbation. The number of barbiturate drugs has been considerably reduced, but only in one out of four patients barbiturate side effects have been relieved [196].

Patients with ID or ASD who show pharmacoresistance may undergo surgical treatment, should they present a focal epileptogenic lesion at the presurgical evaluation. Vagus nerve stimulation (VNS) may result in an alternative option for subjects who are not candidate to surgery, reducing seizure frequency and consequently improving the quality of life [197].

29.6.6 Psychosocial Treatment for ID and ASD

The clinical management of epilepsy in people with ID is complex; compared to the general population, in this population seizures are unpredictable, atypical, more frequent, and often drug resistant. A reduced epilepsy control can have a significant effect on the quality of life and mortality. However, even when epilepsy is well controlled by drugs, many people with ID and their caregivers report difficulties in managing the disease and a negative impact on well-being [198].

Kerr and collaborators [103] believe that the management of epilepsy in people with an ID demands high professional standards across a range of clinical domains.

On the whole, the guidelines highlight the need for people with epilepsy and ID to receive appropriate information and training in order to increase their capacity to manage their condition [199, 200].

However, there is a lack of appropriate training and support activities for people with ID and their caregivers, as well as a lack of easily accessible information [198]. It is not clear how many interventions exist, which are their characteristics and their impact. In fact, there are no methodologically "relevant" studies aimed at assessing the quality of the intervention [201]. However, specific trainings addressed to the professionals involved in the care of these patients [201], caregivers [201] (and the same persons with ID [202]) are needed, being considered as important factors for a positive outcome.

The treatment of the child with ASD and epilepsy should aim not only at reducing seizures but also at improving cognitive and social development [203]. Interventions in seizure management, along with the development of cognitive and social skills, are probably the best solution to maximize the outcomes of neuropsychological development in children with ASD and epilepsy.

Although the effectiveness of the interventions aimed at enhancing social cognition skills in children with ASD and epilepsy has been underestimated [204, 205], there is, however, clear evidence that interventions aimed at increasing social cognition skills can improve cognitive functioning, adaptive behavior, and social behavior, with results maintained for at least 2 years after the intervention [206–208]. Rodenburg and colleagues [209] believe that parents can benefit from parent-training programs in order to receive psychological support and improve coping strategies for the management of their children both in childhood and adolescence. Early interventions applied by parents to their children with epilepsy at risk of ASD have not only produced improvements in social behavior, but may also have an impact on epilepsy [210].

On the basis of our experience, the rehabilitative work with patients with ID or ASD and epilepsy should have a multidisciplinary

approach based on a multidimensional vision that requires the involvement of different operators, family members, and caregivers specially trained in the management of these complex clinical conditions.

Pharmacokinetic or pharmacodynamic interactions of AEDs with the psychotropic drugs used in people with ID and/or ASD are important factors which could determine behavioral impairments. On the other hand, some AEDs show a psychotropic action and could be helpful in the management of behavioral disturbances. Specific trainings for persons with ID and/or ASD, professionals, or caregivers are needed, being considered as important factors for a positive outcome. Interventions aimed at increasing social cognition skills could improve cognitive functioning, adaptive and social behavior.

29

Tip

- There are several biases that limit the epidemiological estimate of epilepsy in people with ID or ASD: sensory impairments, which may reduce self-report of seizures; changing of caretakers over time, creating communication problems; different inclusion criteria; occurrence of subclinical seizures or nonepileptic events.
- Many factors correlated with epilepsy may determine cognitive dysfunction, such as etiology, seizure activity, type of EEG abnormalities, neurological side effects of AEDs, concomitant disability or neurophysiological impairment, environmental factors and parental attitudes, emotional and personality characteristics.
- The choice of the most appropriate AED is driven by the type of epilepsy and tolerability which cannot be expected a priori and should be evaluated case by case.
- The multidimensional nature of epilepsy, ID, and ASD needs a multidisciplinary approach in the field of diagnosis and treatment.

Key Point

- Persons with ID and/or ASD are particularly exposed to mental or behavioral syndromes secondary to physical diseases, due to the high rate of physical health problems in general and the frequent link between specific genetic alterations and specific physical disorders determining psychopathology.
- Organic anxiety disorder and organic affective disorder share many clinical aspects and represent the most common organic mental disorders.
- The most important therapeutic option for organic mental disorders is the treatment of underlying medical condition, when impossible or insufficient, a symptomatic therapy of mental symptoms by psychotropic drugs is necessary.
- Prevalence of epilepsy is higher in the population with ID and/or ASD than in general population.
- Clinical management of epilepsy in people with ID and/or ASD is complex; compared to the general population seizures are unpredictable, atypical, more frequent, and often drug resistant.
- Action of AEDs on behavioral disturbances is strictly correlated with their pharmacokinetic or pharmacodynamic interactions with psychotropic drugs used in people with ID and/or ASD.
- Rehabilitative work with patients with ID or ASD and epilepsy should have a multidisciplinary approach requiring the involvement of different operators, family members, and caregivers specially trained in the management of these complex clinical conditions.

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Substance-Related and Addictive Disorders in Intellectual Disability

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Contents

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Learning Objectives

The primary objective of this chapter is to raise awareness on the extent of problematic substance consumption in people with ID, a neglected issue in both research and clinical practice. This chapter is developed in order to resume current epidemiological data and to explore the specific diagnostic issues and clinical characteristics of SAD in ID people. Finally, assessment options and treatment approaches for SAD in ID population will be discussed.

30.1 Introduction

Substance use (SU), substance use disorders (SUD), and substance-related and addictive disorders (SAD) among people with intellectual disability (ID) are some of the less investigated issues both from a clinical and from a research point of view. Thus, the main purpose of the first part of this chapter is to clarify the terminology related to this area. Then, an overview of available epidemiologic and clinical data will follow. Some awareness on this topic has been raised over the past two decades with an increasing amount of literature, especially regarding prevalence rates and risk factors [1], whereas there is a substantial paucity of studies regarding treatment strategies. The generalizability of such data to the overall ID population is limited. That is why, the appropriate interpretation of such heterogeneous data requires a foreword regarding the limitations of research methodologies.

30.1.1 Terminology

There is a great variety of terms regarding SU and SUD (e.g., abuse, misuse, dependence), which creates confusion. As regards the term "abuse" for example, the World Health Organization (WHO) defines it as a "persistent or sporadic excessive drug use inconsistent with or unrelated to medical practice" but the employment of the term is not recommended because of its perceived negative meaning [2]; the US Food and Drug Administration (FDA) definition stresses that the substance, regardless of the type, is taken specifically for the positive psychoactive effects produced, "to get high" [3]; and the Diagnostic and Statistical Manual of Mental Disorders in its second-last edition (DSM-IV-TR) defined abuse as "a maladaptive pattern of substance use leading to clinically significant impairment or distress," a residual diagnosis when dependence criteria are not met [4]. The International Classification of Diseases (ICD) counterpart diagnosis is "harmful use" of substances [5], in both its tenth (ICD-10) [5] and eleventh (ICD-11) [6] editions. The term "misuse" instead, according to the WHO definition describes "the use of a substance for a purpose not consistent with legal or medical guidelines, as in the non-medical use of prescription medications" [2]. In this meaning, it is frequently used in the literature on SUD in ID as interchangeable with "abuse." Other definitions of misuse are more restrictive: FDA, for example, considers "misuse" exclusively as the use of medications outside prescription or guide of a healthcare practitioner. This includes patients using a drug for a condition different from that for which it has been prescribed, patients having higher dosages or more frequent assumptions than those prescribed, and persons using medications not prescribed for them [3]. DSM too attributes a similar meaning to misuse. Talking about the use of the term "dependence," it is usually associated to chronic and regular use of the substance. It should be noted that it includes both physical and psychological dependence. There is wide agreement among different international classifications on the definition of "physical dependence" as a cluster of physiological, behavioral, and cognitive symptoms related to the substance withdrawal or to the development of tolerance [3, 5, 7]. Whereas, the "psychological dependence" includes different characteristics ranging from the compulsive assumption of the substance, the experience of impaired control over drug use to the emotional state of craving [2, 5, 7]. The term "craving" has been introduced as a diagnostic criterion in the DSM-5 and it is described as "an intense desire or urge for the drug that may occur at any time but is more likely when in an environment where the drug

previously was obtained or used. Craving has also been shown to involve classical conditioning" [7].

The definition of "craving" by the WHO adds that craving may be also provoked by any kind of physiological arousal state resembling withdrawal symptoms [2]. Finally, the term "addiction" is not used as a classificatory label. It may have different meanings and can be used to comprehend all the abovementioned terms. Indeed, it includes the compulsive use of the substance in spite of the awareness of potential or present harm and negative consequences, the concept of SUD as a chronic illness involving impaired control, craving, and genetic, psychosocial and environmental factors and implying specific neurobiological dysfunctions [8]. The term "Substance-related and Addictive Disorders" (SAD) has been introduced by the DSM-5 to define a new chapter including substance use disorders and the behavioural addiction of gambling disorder [7]. The ICD-11 followed suit, with a name that is extremely similar: Disorders due to substance use or addictive behaviours [6].

30.1.2 Research Limitations

In ID research also, there is a great variability of terms which causes inconsistencies in the application of diagnostic terminology. This, in research settings, makes difficult to compare findings from different studies impeding clear prevalence estimations [9]. In fact, not surprisingly, prevalence rates across studies are broad-ranged. For example, a recent Iranian study in non-ID people, compared DSM and ICD diagnostic criteria finding good levels of consistency among dependence diagnoses (0.81), but very low levels of consistency for abuse and harmful use (0.41) [10]. This is a source of heterogeneity among different studies and may affect the generalizability of results together with other methodological issues. Moreover, this specific population presents peculiar limitations intrinsic to the condition of ID. Factors that contribute to the paucity of scientific and reliable data include the following:

1. ID patients are usually excluded from SUD studies, because of possible atypical manifestations considered as a confounding factor or milder forms of intellectual impairment, as borderline intellectual functioning (BIF), may be overlooked in non-vocational settings [9]. Furthermore, in those studies including ID patients, there are concerns about how ID has been appropriately assessed and scored, and this, in turn, could impair the validity and generalizability of the results.
2. Large epidemiological studies and surveys, for example, those examining utilization and attitudes toward substances among students, often use self-administered tools. In ID people, the limitations in cognitive skills may weaken the validity of collected data [11].
3. ID population is quite heterogeneous by itself for several biological, genetic, psychosocial, and psychiatric reasons. For example, it is well known that brain circuits presiding to different cognitive and executive skills may be variously involved in different genetic syndromes: such a variance may imply different behavioral patterns and a discrepancy in the vulnerability to develop SUD. Even the presence of specific psychiatric comorbidities may influence the proneness to SUD, for example, attention deficit/hyperactivity disorder (ADHD) [12]. In this perspective, a more rigorous identification of clinical phenotypes may allow a better understanding of the problem.
4. Variations in prevalence rates over time may contribute to increase heterogeneity as well as cultural, policy, and social differences across countries [1, 13].
5. The probability to develop SUD according to different ID degree is partly linked to point 3 and partly related to the complex interaction with psychosocial risk factors. In fact, most of the studies on SUD in ID included people with BIF or mild ID. This is comprehensible considering the higher chances for these subpopulations to meet substances, especially in the last two decades after the empowerment of social inclusion policies [1, 14]. However, it could be argued that lower functioning individu-

als might have even higher biological and psychopathological predisposing factors to addiction. From a merely clinical perspective, it is not infrequent to identify behaviors resembling SAD in moderate-to-severe ID patients, such as overeating or misuse and dependence from medications, behavioral dependences, and others [14]. However, until now, the awareness toward alternative addictive behaviors in lower functioning individuals who do not have access to the majority of substances of abuse, is very poor.

Based on these limitations, data on prevalence rates should be critically interpreted, keeping in mind that they should be considered exploratory and that they cannot be generalized to the overall ID population.

The research about addiction and substance use in ID persons is very limited and both epidemiological, clinical, and treatment data have to be interpreted carefully. The validity and reliability of results are influenced by the exclusion of ID persons from most studies about SUD, the large use of self-assessment tools in this field, great heterogeneity of ID persons and across different countries, and different possibility to access drugs according to different ID levels.

30.2 Prevalence and Risk Factors

There are very few good and methodologically strong prevalence estimations for substance use and SUD among ID population [1, 15]. The methodological quality of most articles on this topic has been described as poor or moderate concerning reliability, description of procedures, and designs [16, 17]. Currently available literature, recruiting mainly BIF or mild ID patients, suggests a trend of lower overall licit and illicit drug consumption compared to general population, but an increased risk to develop SUD [9, 18–22] and other negative consequences on functioning [19, 23–25] among ID. Current literature reports a prevalence of total substance-related problems in ID population ranging from 0.5% [26] to 21% [20]. Such a variability could be related to the factors discussed above, particularly the sample characteristics and the studies' setting. For example, in a large population-based survey including almost 10,000 people, 2.6% of the sample met criteria for both ID and substance-related problems [27]. The ID prevalence among treatment-seeking SUD patients ranged from 5.2% to 6.2% [22, 28], two- to three-fold higher than in general population. However, these data are not conclusive. VanDerNagel and colleagues [28] found SUD to co-occur with ID in 4% of the cases as measured in vocational facilities for ID, whereas the prevalence of ID among patients receiving treatments in addiction services was 5.2%. Via capture-recapture analysis, they demonstrated a substantial underestimation of the overall prevalence due to a minimal overlap between the two subsamples. In another study, Cooper and colleagues reported differential prevalence rates according to ID level and gender. As expected, they found alcohol and substance use disorders to be more common in mild ID men (2.5%), whereas the prevalence in moderate-to-profound ID patients was 0.5% (0.8% in men, 0% in women) [29]. Another more recent research found even higher prevalence rates of SUD and indicated tobacco as the most commonly used and abused substance among intellectually disabled persons living independently [15]. Almost half of the 123 study partecipants with mild-to-moderate ID was found to use substances (48% tobacco and 45.5% alcohol) [15]. The authors of the paper also provided data regarding different patterns of substances' use, reporting that 32.2% of the sample showed tobacco-use related problems. As regards alcohol use, 17.8% of the probands reported hazardous drinking, 4.4% harmful drinking, and 11.1% alcohol dependence. These results are in line with previous evidence by Taggart and colleagues [30] who found that 75% of BIF and mild ID patients living independently were at risk of alcohol misuse over a 5-year period.

Therefore, higher cognitive level, ranging from mild ID to BIF, seems to be one of the main risk factors for the development of SUD [15, 29–32]. As for the general population,

young age, and male gender are considered independent risk factors for substance use and SUD [1, 21, 29–33]. Moreover, early onset of substance use may predict a later development of substance-related problems even in ID people. Other risk factors are independent community living [15, 30], poor socio-economic status, and living in the poorest neighborhood [31]. A forensic history is biunivocally related to SUD in ID population, both representing a risk factor and a consequence. In fact, ID people more commonly than normal peers, commit offending behaviors under the influence of alcohol or drugs [34, 35], particularly when substance use takes the form of SUD such as continuous or heavy substance use [36]. On the contrary, ID people with a history of offending behaviors and criminal records have higher rates of SUD than their peers [37]. Finally, psychiatric comorbidity may play a role in increasing the risk of developing SUD [13, 38]. The prevalence of overall psychiatric disorders in patients with co-occurring ID and SUD ranges from 42% to 79% [28]. In a recent study, Lin and colleagues [31] estimated anxiety disorders to be the most common psychiatric comorbidities among ID-SUD patients with a 68% prevalence rate. Affective illness and psychotic disorders followed with prevalence rates of 45% and 36% respectively; more specifically 35.2% of the sample had a diagnosis of depressive disorder, 25.2% of bipolar disorder, and 30.5% of schizophrenia spectrum disorder. Almost a quarter of the sample also had a personality disorder. Moreover, SUD may also be associated with emotional dysregulation and behavioral issues such as aggression [19] and ADHD [12]. In general, psychiatric vulnerability seems to be associated with substance consumption irrespective of the actual development of SUD and substance-related problems [32], but any specific disorder may impact differently on the vulnerability to develop SUD. For example, autism spectrum disorders are inversely associated to both occasional and continuous/heavy substance use, whereas schizophrenia spectrum disorders are strong predictors for substance use and SUD [36]. The presence of personality disorders, other than increasing the risk of SUD, may produce a seven-fold increase of the risk to have had criminal justice system contacts and an 11-fold higher risk for conviction [37].

Substance use and abuse may have similar consequences in people with ID and in general population, although ID population seems to be more vulnerable to negative substance effects [32]. Beyond the already mentioned increased risk to be involved with the justice system, affective instability and unpredictable mood changes, interpersonal conflicts, both verbal and physical aggressiveness toward others, suicidal ideation, poorer mental and physical health, and greater likelihood to hospitalization are common consequences of substance consumption in ID [19, 23, 24, 26, 30, 32]. As expected, the more severe the substance utilization, the more severe are the consequences. In fact, substance abuse impacts more than substance use, particularly on daily functioning and interpersonal relationships [32].

Tobacco Studies on lifetime prevalence of tobacco use in ID found that rates range from 2.6% [39] to 77.2% [15]. The majority of studies showed a prevalence of 15–30% [1, 33, 40–42] with fewer surveys reporting rates lower than 10% [43, 44]. Differences in sampling may account for variable rates. For example, those studies including students with ID reported smoking rates of 23–27% [33, 40, 45]. Higher rates have been detected in people with dual diagnosis (ID plus a psychiatric disorder), reaching over 30%, with the highest rate in those patients with mild ID and BIF [41, 42, 44]. The co-occurrence of alcohol abuse may further increase cigarette smoking [20]. The comparison of different living arrangements confirmed a larger daily consumption of cigarettes in people living independently in group homes (8.5 ± 14.1), with similar lower levels for those living in residential facilities or at home with relatives (1.4 ± 4.4 and 2.2 ± 8.4, respectively) [44].

Alcohol According to some studies, alcohol would be, together with tobacco, the most frequently used substance in ID people. The rate of alcohol use varies largely across studies reaching 77.9% in a study by To and colleagues which includes mild-to-severe ID individuals

[32]. Other studies reported lower but similar prevalence rates: a study including BIF and mild ID people found a 66.1% rate [28] and two studies regarding mild and moderate ID probands found 21% [20] and 45.5% [15] respectively. A recent French survey involving 700 disabled students compared to more than 7000 neurotypical junior high school students, examined the attitude toward drugs through self-administered questionnaires; 62.9% of disabled students reported using alcohol; alcohol use was more common in males, and 16.4% had experienced being drunk: similar rates were found in non-disabled students [33]. Alcohol misuse or abuse rate was about 20% [15, 20, 39]. Compared to other substances, alcohol consumption resulted to be significantly more associated with abuse and use-related problems among people with mild-to-moderate ID and with higher skills to live independently [32]. Similarly, in a sample of 123 individuals with mild-to-moderate ID living independently, prevalence rates for hazardous drinking, harmful drinking, and alcohol dependence resulted to be 17.8%, 4.4%, and 11.1% respectively [15]. These findings confirmed the observation that in people with ID, the line between use and abuse/misuse is fine: indeed, ID people often show lower tolerance to alcohol than non-ID peers with easy-to-appear changes in behavior, mood and attitude, reduced control over the substance, and possibly higher proneness to develop addiction [22]. Prevalence rates may vary according to the level of ID in a case-registered study involving 411 ID-mental health patients, SUD rate was estimated as 12.2% in mild ID, 5.3% in moderate ID, and 2.9% in severe ID [37]. The use of alcohol, particularly in people with BIF and mild ID, is associated with the use of other substances, mostly cannabis: almost half of both samples included in the studies by VanDerNagel et al. and To et al. abused cannabis [28, 32].

Illicit Drugs As regard other substances of abuse in ID literature, cannabis, cocaine, and opiates are the most investigated, whereas data about amphetamines and derivatives, hallucinogens, and inhalants are limited and not fully reliable. Nevertheless, rates for drugs' use in ID patients are apparently higher than in general population and vary between 14.5%–15% [1, 36] and more than 50% [28].

Cannabis Cannabis emerged as one of the most frequently used and abused substance. Data regarding cannabis use in ID people are heterogeneous across studies. Four studies examined cannabis consumption among disabled high-school students finding rates ranging from 9.1% to 26% [33, 40, 45, 46]. The large ACHA-NCHA II biannual survey included 60,940 college students across the United States: authors found that 44.2–49.5% of disabled students (not only ID but also physical and social disability) reported having used cannabis in the previous 30 days, with an odds ratio of 1.36–1.67 compared to non-disabled students [47]. Curiously, antipodal data emerged from two other samples, similar in their compositions (they both included mild-to-severe ID), examined by Chaplin et al. and by To et al. [32, 37], who found 8.8% and 39.4% cannabis use prevalence rates, respectively. Interestingly, in the study by To et al., more than half of cannabis users also developed misuse/abuse conducts, 31.3% of the sample consumed cannabis on a daily basis, and 80% had used it for more than 5 years [32]. When considering the subpopulation of people with mild ID or BIF even a higher rate emerge: VanDerNagel and colleagues found in fact that 8% of the sample used cannabis exclusively, whereas 34.8% used it in combination with alcohol [28]. Finally, the highest rates of cannabis consumption have been measured in ID prisoners reaching 51.2% in a 2011 survey. Comparing to their average IQ peers, ID probands were more prone to develop cannabis dependence (51.2% vs. 42.1%, $p = 0.01$) and the co-occurrence of ID and cannabis dependence doubled the risk of psychosis [48]. Moreover, 52.9% of incarcerated ID individuals used cannabis in the 12 months prior to commit the crime and imprisonment [49].

Cocaine Cocaine resulted to be used by a percentage of 2.7–12.5% of ID people living in the community [15, 20, 32, 37]. It is probable that more than 60% of cocaine users develop addiction and cocaine-related problems [32]. The studies regarding ID people with an involve-

ment with the criminal justice system showed a higher but similar cocaine use rate (12.6%) [49] with a 6% prevalence of misuse/abuse [50]. Prevalence seems to be lower among ID students (3%) [40].

Opiates According to available data, the use of heroin and other opiates seems to be lower than those of other drugs, with rates of 1.2–1.6% [15, 28] to 9.6% [32] in ID and BIF individuals living in the community and 11–17.9% in ID criminal offenders [49, 50]. The rates of abuse, dependence, and opiates-related problems are similar to those found for cocaine [32]. For example, a large cross-sectional clinical study including Medicaid care data of about 9484 ID patients with a comorbid SUD diagnosis, highlighted a prevalence of opiate dependence of 16.4%. Furthermore, as in general population, ID people would misuse opiate replacement medications prescribed for the treatment of opioid addiction. For example, referring to the 5 patients treated with methadone or buprenorphine in the sample by To et al., 80% of them misused the medication [32]. Given the small dimension of this subsample, this result is not generalizable but interesting anyway. In 180 ID individuals evaluated for substance consumption and adherence to programs for the treatment of addiction during their conviction, 30% abused opiates, 3% reported the misuse of methadone in the year prior the arrest, and 47% was treated with buprenorphine in prison [51].

Amphetamines The use of amphetamines and derivatives in ID has been less extensively explored and precise prevalence data are substantially lacking because most studies combined amphetamines' rates together with rates of other substances and abused stimulant medications. For example, Chaplin and colleagues reported an overall rate of 3.2% including opiate, solvents, and amphetamines use [37]. Mild-to-moderate ID subjects who live independently reported a lifetime use of amphetamines in 4.1% of the cases [15] whereas, in a study including severe ID also, the prevalence was 2.9% with two third of the users also showing problems related to amphetamine abuse [32]. In another study on substance and medication use and abuse, 4.7% of the overall sample composed of 86 mild ID and BIF individuals was prescribed with stimulants; the rate of misuse was 15.1% for the entire sample, 23.8% for BIF, and 6.8% for mild ID [28]. The authors did not specify whether the category "stimulants" included only medications (e.g., mixed salts of amphetamines, methylphenidate) or not. Plant and colleagues reported similar rates in a forensic sample (8%) [50]. The co-occurrence of ADHD may favor misuse and diversion of prescribed stimulant medications [52].

Sedatives To our knowledge, there are no systematic data regarding sedative misuse and dependence in ID people even though sedatives especially benzodiazepines (BDZ) are extensively prescribed in clinical practice to treat both psychiatric and neurological conditions frequently co-occurring in this population. In fact, the abuse of BDZ could be clinically relevant given the easy accessibility also to people with higher severity of ID. Further, it has to be taken into account that BDZ use in ID patients is associated with behavioral side effects more frequently than in general population, thus prescribing BDZ requires caution [53].

Food and Drinks Caffeine and other beverages or food with abuse potential (e.g., tea and derivatives) would require similar considerations. In clinical practice, it is not rare to meet ID individuals who show compulsive assumption and addiction-related behaviors, independently of IQ level.

Summarizing risk factors for substance use and SUD in ID, young age, male gender, and milder severity degrees of ID are quite unanimously reputed to be factors independently associated to SUD in ID population [15, 28, 30, 32, 36, 54]. Other factors associated to the development of SUD are the earlier age of first substance use [30], hazardous drinking in the previous 5 years [1, 30], and a diagnosis of conduct disorders, ADHD, and antisocial personality disorders during adolescence [12, 55]. More in general, the co-occurrence of mental illness, particularly mood and psychotic disorders may represent a ground for SUD and

related behavioral complications [19, 30–32, 36, 54]. Moreover, also specific temperamental and personality traits have been reported to be associated with SUD in ID population similarly to what happens in people with an average IQ: difficulties in emotional control [19], negative thinking, impulsivity, and sensation seeking [38] are considered predisposing factors. Even environmental factors may play a role: living independently with low levels of supervision [1, 15, 28, 31, 32, 55], poor socio-economic status including scarce educational and employment opportunities, living in a poor neighborhood, belonging to ethnic minority groups, and isolation. All of these are well-known risk factors in general population, however, few data are available for ID people [31]. Finally, also negative life events are frequently reported (history of trauma and physical or sexual abuse in childhood and/or adulthood) as risk factors [1].

30.3 Criteria and Clinical Features

The Diagnostic Manual for Intellectual Disability, Second edition, (DM-ID-2) [56] provides suggestions and revisions to adapt DSM criteria to people with ID. Such adaptations were highly needed considering that the clinical presentation of psychiatric disorders in ID patients often shows atypical features making the diagnostic process difficult. This, in turn, often leads to under- or misdiagnosis with a negative impact on therapeutic interventions. With regard to substance-related and addictive disorders (SAD) according to DSM-5 description [7], DM-ID-2 does not provide particular suggestions for adaptations of criteria. This is probably related to the relatively low-to-moderate quality of the evidence currently available, as already discussed. In this perspective, it is possible that individuals with mild-to-moderate ID and BIF could satisfy SUD criteria, whereas "it is almost impossible to apply the diagnostic criteria for substance abuse to individuals with severe-to-profound degrees of ID" [57].

DSM-5 SAD category includes several diagnoses that have in common the excessive use of a psychoactive substance. Ten classes of substances have been identified (e.g., alcohol, caffeine, cannabis, hallucinogens, inhalants, opioids, sedatives, hypnotics and anxiolytics, stimulants including amphetamine-type substances, cocaine and other stimulants, tobacco, and other or unknown substances). The excessive use of such substances causes a direct activation of the brain reward system, so intense that normal activities may result impaired: the assumption of the substance may produce feelings of pleasure as a result of the initial activation of the reward system usually described as "to be/get high." Individuals with congenital or acquired impairment of inhibitory brain mechanisms, as some intellectually disabled are, are considered at higher risk to develop a substance-related disorder. SAD are divided in the two major groups: *Substance Use Disorders* and *Substance-Induced Disorders*. The last two decades of research on addiction has disclosed that a pathological activation of reward/motivation brain circuits similar to that provoked by substances of abuse, may be also caused by a range of behaviors that are not pathological in their nature but they become it, assuming an "addictive" quality. In this regard, DSM-5 and, subsequently, DM-ID-2 have separately included in SAD section also gambling disorder, moving it from impulse control disorders where it is located in DSM-IV-TR. Other excessive, repetitive, and impairing behaviors labeled as *behavioral addictions,* such as sex addiction, exercise addiction, and shopping addiction have not been included since current evidence is not sufficient to establish valid and reliable diagnostic criteria, psychopathological, and course descriptions [7], in spite of their clinical relevance in some cases. Internet gaming has raised clinical interest in the last few years hence it has been included in the DSM-5 "Conditions for further study" section.

Substance Use Disorders Regarding SUD, the pathological pattern of *substance use* is defined by four major symptomatic areas: impaired control over quantity and duration of substance use, social impairment caused by substance use, risky use of the substance, and pharmacological indicators. The impaired control over the sub-

stance use is described by the first four criteria: (1) Use of larger amount of substance or for a larger period than how desired. (2) Persistent unsuccessful desire to discontinue or reduce the use of the substance. (3) Great deal of time spent to obtain or use the substance or to recover from their effects. (4) Craving. Social impairment is explored by the following three criteria: (1) Failure to fulfil important activities at school, work, or home caused by the substance use. (2) The use of the substance persists even after the evidence of continuous or recurrent social or interpersonal problems caused or exacerbated by substance effects. (3) Using the substance causes the drop out or reduction of important social, occupational, or recreational activities. Risky use is described by two criteria referring to the use in physically hazardous situations and the persistence in use despite knowing that the substance impacts on physical or mental health, evidenced by continuous or recurrent physical or psychological disturbances. Finally, pharmacological criteria encompass tolerance and withdrawal symptoms. Tolerance is described as the need to markedly increase the dose of the substance to obtain the desired effect, or markedly reduced effects when the usual dose is consumed; whereas withdrawal refers to blood and tissue substance concentration decline causing a constellation of symptoms, different for each class of substances, after a heavy and continuous use. During withdrawal, the consumption of the substance or an agonist may reduce or completely relieve symptoms. DSM-5 provides also the diagnosis of SUD in presence of tolerance and withdrawal symptoms in the context of medical treatment (e.g., opioid analgesics, sedatives, and stimulants), only if prescribed medications are used inappropriately as demonstrated by SUD criteria fulfilment. To make a diagnosis of SUD, at least two of the above-mentioned criteria must be present (Table 30.1). There are some minor differences between substance categories. For example, withdrawal criterion is not present in the list of symptoms for tobacco use disorder [7].

Further, DSM-5 provides some course and severity specifiers. In particular, course specifiers include "in early remission" and "in sus-

Table 30.1 Summary of DSM-5 Criterion A for substance use disorder (adapted from DSM-5 and DM-ID-2) [7, 56]

A problematic pattern of substance use: at least two of the following within a 12-month period		
Impaired control over the substance		**Description of the symptom and adaptation for ID**
1	Use of larger amount of substance or for a larger period than how desired	–√
2	Persistent unsuccessful desire to discontinue or reduce the use of the substance	Poorly applicable in individuals with severe-to-profound ID due to self-control and language/communication impairment
3	Great deal of time spent to obtain or use the substance or to recover from their effects	Reliable in severe-to-profound ID as it is observable. Behavioral equivalents may include repetitiveness and compulsivity with reduced involvement and focus on usual and pleasant activities
4	Craving	Strong desire or urge of the substance provoked by environmental and inner stimuli, even those resembling withdrawal. Difficult to be explored in patients with low verbal abilities. Behavioral equivalents might include problematic and challenging behavior chronologically related to abstinence, stressful events, and places/moments of the day when/where they were used to consume the substance

(continued)

Table 30.1 (continued)

A problematic pattern of substance use: at least two of the following within a 12-month period		
Social impairment		
5	Important activities at school, work, or home failure caused by the substance use	Minor adaptations have to be made to correspond to the individual's functioning level and with respect to premorbid functioning, variations have to be detected. Behavioral equivalents may include refuse to participate or reduced involvement in usual activities in individuals with more severe degrees of ID due to the search or consumption of the substance
6	Continuous or recurrent social or interpersonal problems caused or exacerbated by substance effects	–√
7	Drop out/reduction of important social, occupational, and recreational activities	Reliable in severe-to-profound ID as it is observable
Risky use		
8	Use in physically hazardous situations	Poorly reliable in people with severe-to-profound ID spending much time under surveillance and with basically reduced sense of danger and poor inhibitory control
9	Use despite knowledge of continuous or recurrent physical or psychological disturbances caused by the substance	Only moderately reliable in people with severe-to-profound ID because cognitive difficulties may severely reduce self-awareness of psychological state as well as difficulties to understand the consequential relationship between substance consumption and physical problems
Pharmacological		
10	Tolerance	Need to markedly increase the dose of the substance to obtain the desired effect, or markedly reduced effects when the usual dose is consumed. Reliable as it is observable, for example, through the reduction of the span of time between one consumption and the following
11	Withdrawal	Blood and tissue substance concentration decline causing a constellation of symptoms, different for each class of substances, after a heavy and continuous use. Symptoms are observable (e.g. psychomotor agitation or problem behaviors) and chronologically correlated to abstinence

tained remission" according to the period of abstinence, lasting for at least 3 months but less than or more than 12 months, respectively. An additional specifier for remission (in a controlled environment) is used for those individuals living in an environment with restricted access to the substance. The three severity specifiers depend on the number of diagnostic criteria fulfilled: mild for 2–3 criteria, moderate for 4–5 criteria, and severe for 6 or more criteria).

A dissertation regarding the specific features of each category of SUD and the effects of each substance goes beyond the purpose of this chapter. In fact, considering BIF and mild-to-moderate ID, the physiological effects of substances, the development, and course of SUD is usually comparable to those in non-ID patients. We will limit to describe general and associated features peculiar to ID.

Epidemiological findings showed substance use to begin in early-to-late adoles-

cence in ID individuals as in non-ID peers [1]. Substance use rate is higher in non-ID than in ID population: the main reason could be the easier access to substances for average IQ individuals. However, ID people using substances showed increased risk to develop SUD comparing the non-ID people [9, 26, 36]. Moreover, substance use seems to impact more negatively on both mental and physical health in ID compared to non-ID population [32, 58]. In spite of expectations, all types of substances are used by individuals with ID [28, 32], alcohol [1, 13, 30, 59], and tobacco [15] being the most frequently used, at least in people with mild ID and BIF. There are no systematic data regarding readily accessible substances such as caffeine, medications, food, and beverages, which could have even higher abuse rates. Finally, there is no agreement regarding poly-use to be more common than single substance use [15, 28, 32, 36], however, poly-users are usually younger than 30 years [28].

SUD and its complications might be considered as risk factors for needing specialist treatments in psychiatric, addiction, and more in general, medical services earlier in life. In this vein, in a large survey considering Medicaid records in US [54], ID substance users were younger compared to ID-only individuals seeking treatment for a variety of medical conditions. For example, the survey evidenced that ID people suffering from SUD might be more vulnerable to develop organic substance-related conditions with an earlier onset: ID-SUD patients treated for alcoholic liver diseases were on average in their mid-30s, at least 10 years younger than both their ID without SUD and average IQ-SUD counterparts. As it concerns both mental and physical health, it has to be taken into account that also direct physiological effects of substances may be amplified or paradoxical in ID. Additional cognitive deficits [60], induction or precipitation of seizures [14, 30], worsening of gross motor function [21], confusion, sedation, and coma, especially when medications are associated [61], are common neurological effects. The negative impact on pre-existing somatic problems is also frequent, for example, cardiovascular, respiratory, and gastrointestinal illnesses [14, 60]. It is possible that the sum of these factors may increase the mortality risk in ID compared to average IQ substance users [61]. Moreover, substance use may cause psychopathological states with acute and sub-chronic development and course, such as affective instability, mood dysregulation, inappropriate emotional responses, extreme anxiety and panic attacks, depressive symptoms and despair, suicidal ideation, odd or paranoid thinking, and bizarre behaviors [17, 19, 24, 32, 54]. Usually, in spite of similar effects on mental states and substance-induced altered behaviors, heavy disturbance on functioning in different life domains are related to SUD more than to occasional substance use and intensification of affective instability, unpredictable mood changes, and the appearance of suicidal ideation would be specifically related to SUD [32] and may represent indices of greater severity. The relationship between SUD and psychiatric disorders is biunivocal. Interpreting data from current literature, it can be hypothesized that psychiatric illness is a risk and anticipating factor for the development of a SUD in ID as well as in average IQ population [62, 63]. On the other hand, SUD might represent a trigger or causal factor for some psychiatric diagnoses. Epidemiological data demonstrated that mental health problems are the most common additional diagnosis in ID-SUD patients, with five-fold increase of the relative risk to have any psychiatric diagnosis comparing to ID people without SUD [54]. On the other hand, Pezzoni and colleagues found that in 40 mild ID patients referring to psychiatric services and screened for alcohol abuse, about 20% presented alcohol abuse, 87% of them have had also a lifetime psychiatric illness, and 75% were also smokers [14]. Depressive and bipolar disorders resulted together the most frequent co-occurring diagnosis, with 35% and 25% prevalence rates respectively. Anxiety also is frequently associated with SUD in ID people [19, 31]. Psychotic and personality disorders are common too (30% and 23% prevalence, respectively) [31]. Finally, schizophrenia spectrum disorders are associated to continuous and heavy substance use [36] whereas the impact of co-occurrent

autism spectrum disorder on substance use and development of SUD still needs to be clarified [36]. A recent review explored epidemiology and risk factors associated to SUD in autism spectrum disorder. This subpopulation shares most risk and protective factors for SUD of ID persons such as co-occurrence of externalizing, mood and anxiety disorders, perceived social deficits, peculiar social motivation, psychological distress, weak executive functioning and maladaptive coping styles, familial history of SUD, and poor social conditions. Autism spectrum disorder seems to be related to higher risk for the development of SUD and greater consumption [64].

Risky behaviors, loss of cooperativeness, poor social judgment, hostility, and aggressiveness ranging from being argumentative and confrontational to physical aggressions are the most common behavioral disturbances, and usually, represent the main cause for interpersonal, familial, and daily functioning impairment, or for vulnerability to exploitation and involvement with justice systems [1, 12, 17, 24, 30, 32, 36]. Some authors suggested that SUD would have a particularly relevant role in offending behaviors committed by ID individuals, in fact many of those referred to specialist services or convicted for criminal acts had committed offending behaviors under the influence of alcohol or substances [12, 34–36]. Hassiotis and colleagues recently found a significant correlation among psychosis, cannabis dependence, and attempted suicide in ID offenders [48]. In this study, cannabis use resulted a probable mediator between ID and psychosis.

The course of SUD in ID population is usually alike to that in general population with an early onset usually in late adolescence, even if DM-ID-2 underlines that in ID individuals the age of first substance consumption could be delayed compared to average IQ peers [56]. This delay could be related to the parents and teachers' higher "vigilance" on ID adolescents during their school years. In fact, the period following the end of the studies, especially when specific services and employment integration plans are not provided, should be considered as the most critical time with the highest level of vulnerability. SUD has often a chronic and recurring course as the persistent use of the substance may induce enduring physiological modifications in the motivation/reward, inhibitory control, and salience interrelated brain systems. Such changes may last for years even after the suspension of the substance use making reason for relapses. In ID substance users, two factors may worsen this aspect: pre-existing neurological and cognitive deficits basically reducing self-control and inhibition capacities can accelerate the transition from substance use to SUD and severe addiction [56]. However, it has not been conclusively ascertained whether or not and what kind of specific cognitive and executive deficits would be mediators between ID, addiction, and its development. In fact, a Dutch group investigating on this topic in the last 5 years did not confirm previous speculations regarding the involvement of attentional bias toward the substance and alterations in working memory and inhibitory control [65–67]. Secondly, cognitive deficits may also interfere with capacity to understand and participate in standard rehabilitative programs. In this perspective, it has to be taken into account that addiction services are often poorly accessible for ID addicted people and special informative and rehabilitative programs are not available. This last point will be more extensively discussed in the Treatment section.

Substance-Induced Disorders These include intoxication, withdrawal, and substance/medication-induced mental disorders. Intoxication is generally characterized by the development of a substance-specific syndrome linked to the recent consumption of the substance. The appearing symptoms must have a causal physiological relationship to the substance. Intoxication criteria are not available only for tobacco. Given that each substance induces a specific symptomatic pattern, more common intoxication symptoms include "disturbances in perception, wakefulness, attention, thinking, judgment, psychomotor, and interpersonal behavior" [7]. It is likely that the diagnosis of substance intoxication of mild severity or with chronic course in ID individuals may be more complicated than in neurotypical peers, as most of them may have some degree of impairment

in cognitive and executive functions with some overlapping features (overshadowing phenomenon; see ▶ chapter 5): mild intoxication symptoms, for example, might be attributed to the underlying neurodevelopmental condition. Also, withdrawal syndrome may assume substance-specific features, being the strict link between onset of symptoms and blood levels' decrease of the substance in long-term users the common denominator. Withdrawal criteria are not available for hallucinogens and inhalants.

There are ten *Substance-Induced Mental Disorders* according to DSM-5: psychotic disorder, depressive bipolar and related disorders, anxiety, obsessive-compulsive and related disorders, sleep disorders, sexual dysfunction, delirium, and neurocognitive disorder. A chronological and causal relationship with substance intoxication or withdrawal has to be established based on history, laboratory findings, and physical examination (Criterion B). A diagnosis of substance-induced mental disorder instead of a diagnosis of substance-induced disorder only can be formulated when the syndromic presentation of one of the ten above disorders is clinically relevant and needs independent attention (Criterion A). Obviously, the presence of another mental disorder justifying the symptomatology has to be excluded (Criterion C). Moreover, this criterion indicates as an exclusionary condition the persistence of the symptoms for a long time after the cessation of acute intoxication, withdrawal or the exposure to the substance. In fact, the course of substance-induced mental disorders, by definition, should be transitional and would resolve after the cessation of the causal factor, usually improving within 1 month. However, some disorders, mostly substance-induced neurocognitive disorders or hallucinogen persisting perception disorder may continue as the result of permanent brain damage. An interesting example in ID patients would be alcohol-induced neurocognitive disorder, given the high prevalence of alcohol abuse in this population. Unfortunately, studies specifically addressing this topic are lacking. When the constellation of symptoms (e.g., hallucinations, anxiety, and rapid mood shifts) occurs in the context of delirium, for example, the correct diagnosis should be delirium only. DSM-5 provides also some generalizations regarding the predictability of substance-specific syndromes: it is expected that more sedating compounds (e.g., sedatives, hypnotics or anxiolytics, and alcohol) produce prominently depressive disorders or confusion states during intoxication; on the other hand, intoxication due to excitatory drugs such as stimulants, amphetamines, and cocaine may cause a variety of conditions ranging from anxiety to mania and psychosis [7]. These elements must be weighed when considering ID individuals; this population indeed, as children and elderly, may present paradoxical reactions to substances and medications in the measure that, for example, mild-to-moderate sedatives' intoxication may occur with anxiety, psychomotor agitation, and manic symptoms.

Gambling disorder (GD) is the only behavioral addiction included among SAD in DSM-5 [7]. The diagnosis was moved to this category from impulse-control disorders where it was placed in DSM-IV-TR. GD Criterion A describes nine symptoms or pathological behaviors that cause clinically significant impairment or distress during a 12-month period. At least four of them must be fulfilled to make a diagnosis of GD. ◘ Table 30.2 summarizes GD Criterion A. The second criterion requires that gambling behavior has not to be exclusively present in or explained by a manic episode. Specifiers are the same as for SUD, with two additional ones regarding the *episodic* or *persistent* course of disorder. DM-ID-2 do not provide adaptations but only suggestions regarding the limitations that ID people may show to access money, resources, and treatments [56]. These limitations, similar to those described for the access to substances, may mislead the clinician to underestimate the relevance and the severity of gambling behavior, sometimes overlooking the diagnosis. GD literature in ID is scarce. A 2% prevalence rate of GD in mild-to-moderate ID and BIF people was found and was similar to that of general population. It has also been estimated that an additional 6% may meet criteria

Table 30.2 DSM-5 Criterion A for gambling disorder [7]

Problematic gambling behavior: at least four of the following within a 12-month period	
1	Needs to gamble with increasing amounts of money in order to achieve the desired excitement
2	Is restless or irritable when attempting to cut down or stop gambling
3	Has made repeated unsuccessful efforts to control, cut back, or stop gambling
4	Is often preoccupied with gambling (e.g., having persistent thoughts of reliving past gambling experiences, handicapping or planning the next venture, thinking of ways to get money with which to gamble)
5	Often gambles when feeling distressed (e.g., helpless, guilty, anxious, and depressed)
6	After losing money gambling, often returns another day to get even ("chasing" one's losses)
7	Lies to conceal the extent of involvement with gambling
8	Has jeopardized or lost a significant relationship, job, or educational or career opportunity because of gambling
9	Relies on others to provide money to relieve desperate financial situations caused by gambling

for problematic gambling. Furthermore, GD seems to be associated to living independently and co-occurring anxiety and mood disorders [68]. Unfortunately, reliable studies regarding development, course, and prognosis of GD in ID are lacking.

The textbooks aimed to give descriptions of mental disorders in ID and based on DSM-5 and ICD-10 do not provide significant adaptations for SAD. Thus, diagnostic criteria are the same as for the general population. This raises some issues, especially that hypothetically the diagnosis of SAD would be limited to only BIF and mild ID persons as it is not probable that lower functioning persons may present and communicate symptoms in a way that is similar to that of the general population. Furthermore, in persons with ID the boundaries of addiction should be enlarged to cover any kind of behavioral addictions.

30.4 Specific Assessment

The assessment of SUD requires a multidisciplinary evaluation regarding substance consumption, physical, and mental health and psychosocial factors associated to the illness. Current guidelines [69] indicate as a priority a correct anamnestic assessment on substance use accounting for historical and recent patterns of substance use. Obtaining collateral information and the use of diaries may add objective data to the history and may be used as monitoring instruments. A correct framework of the clinical picture is very important to set up appropriate treatment and the estimation of the severity of the substance use is required: the clinician should collect information and observations regarding the modality, circumstances, and the amount of substance consumed. Consequences in terms of behavioral alterations and emergence of psychiatric symptoms have to be evaluated. The identification of dependence and withdrawal symptoms is crucial to provide adequate pharmacological management. A complete physical examination and appropriate supporting tests are due to identify pre-existing or substance-related physical illness or problems. Checking cardiovascular, liver, and renal functioning, a variety of hematic parameters as full blood count and electrolytes, nutritional state, and eventual infectious illnesses (e.g., hepatitis B and C viruses and HIV) would be performed. Moreover, toxicological blood and urinary tests may help to monitor substance consumption and actual abstinence. A psychiatric diagnostic process to detect mental illness is equally indispensable as the presence of an unbalanced psychiatric disorder, apart from representing a risk factor for the development and retention in substance abuse, may cause failure of all the other interventions. The assessment would include psychological and social problems, as

they represent important targets of intervention and rehabilitation. At the beginning of taking charge of the patient, where not available, the clinician should plan a basal cognitive evaluation with multiple aims: the confirmation of ID and its staging, and the evaluation of specific cognitive dysfunction useful to choose the most appropriate intervention strategy. This is also useful as a baseline parameter to compare with further cognitive evaluation to monitor any positive change related to abstinence or, conversely, any worsening of deficits caused by SUD. Finally, the evaluation of motivation to change and quit the substance or addictive behavior is required. Overall, the diagnostic and follow-up process may be helped by the use of specific assessing tools.

In this regard, a variety of assessing tools for the screening and diagnosis of substance use, SUD, and tools specific for alcohol or other substances are available for the general population. Unfortunately, assessment instruments adjusted for ID are lacking [13]. Adaptations, especially regarding self-administered questionnaire should consist of simplifications of questions and, eventually, visual supports to overcome cognitive and communicative difficulties. The lack of specific assessment tools for ID is a matter of concern since this can lead to the under-identification of SUD in ID population.

Below a brief list of some tools commonly used in research and clinical practice for the general population:

1. The Alcohol Use Disorders Identification Test (AUDIT) is the screening tool for alcohol use and abuse developed by the WHO [70, 71]. It is a 10-item questionnaire exploring alcohol consumption, drinking behaviors, and problems related to alcohol. It is available in the clinician-administered and the self-report versions. AUDIT is based on the quantitative evaluation of drinking habits to differentiate drinking problems, hazardous and harmful alcohol use, or dependence. AUDIT has not been specifically validated for ID population, but its simplicity could make it suitable also for BIF and mild ID.

   The Drug Use Disorders Identification Test (DUDIT) is the screening tool version for substances complementary to AUDIT [72]. It has similar structure and approach to explore substance consumption and related problems. There is also an extended version (DUDIT-E) with additional questions about consequences of substance abuse and treatment readiness [73].

   The strength of AUDIT and DUDIT seems to be the use of familial language and free from criticism: the information obtained can be the starting point for the clinician to further explore substance use and attitudes of the patient. The main weakness is that they are self-administered and, especially with higher degrees of ID, results could not be completely valid and reliable.
2. CAGE is another simple screening test to detect lifetime excessive alcohol consumption and related problems. CAGE-AID is the version adapted to explore also problematic drug use. The acronym CAGE refers to the first four fundamental questions of the test (need to Cut down drinking/drugs, Annoyed by people's critiques on drinking/substance use, feeling of Guilt about drinking/drug use, Eye-opener as the need to drink or assume drugs soon in the morning to avoid hangover). The test is positive when at least one answer is "yes." This tool has several limitations because, even if specific, it is not completely sensitive because questions are interpretable and this is particularly true in ID population [74].
3. The Michigan Alcoholism Screening Test (MAST) is a 25-item questionnaire developed to rapidly screen lifetime problematic drinking and alcohol-related consequences, being able to provide a gross estimation of the severity of the disorder. It is available both as self-questionnaire and clinical interview [75]. In ID people, it is recommended that the clinician administer the test in order to ensure the correct understanding of the questions.

One example of a tool adapted to ID is the Substance Use and Misuse in Intellectual Disability Questionnaire (SumID-Q) developed and validated by VanDerNagel and colleagues of the Dutch SumID group [76]. It is

a structured interview specifically developed for mild ID and BIF individuals to assess risk factors for SUD, substance use, and their consequences. Adaptations consisted in the use of a simplified language and pictures in order to facilitate the proband to understand questions. It can be used to assess lifetime, last month, and current use of a variety of substances including tobacco, alcohol, cannabis, and stimulants. This allows a complete overview on alcohol and substance use across the life and can also be used as a follow-up instrument.

Assessing tools for SAD specifically addressed to ID people are lacking; instruments used for the general population are usually employed also for ID individuals. It has to be remembered that addictive behavior has to be carefully explored and the diagnosis requires a multidisciplinary and complex evaluation.

30.5 Treatment

Available literature points out the lack of treatment options and services for substance-related problems specifically addressed to the ID population [1, 13, 26, 27, 77, 78]. There are several reasons explaining the low access to treatments for SUD and the consequent reduced development of high-standard care measures for ID people. In the past, it was estimated that one third of community treatment services for addiction did not provide specific treatments for patients with ID, and one fifth of services overtly did not accept ID people in their treatment programs [79]. More recent surveys demonstrate not so discouraging data. For example, some research in forensic settings enlightened that ID individuals received treatment for drug addiction and drug education even if significantly less compared to their average IQ counterparts during conviction period [48].

A discussion about social and health policies in this field goes beyond the purpose of this chapter (for more details cfr. [27, 61, 80]): we will limit to mention the main issues preventing ID-SUD population to access adequate interventions. For example, the analysis of Medicaid records by Slayter [80] showed that young individuals with ID and SUD, compared to those with SUD only, had fewer opportunities to access and engage specific treatments and presented higher rates of dropout. As suggested by the author, a variety of factors may influence access and retention rates such as inefficiency of transports, fragmentation of health services for ID, SUD and mental illness, physical, language and communication barriers, and the lack of treatment programs entailed to meet special needs of ID people [80]. The inappropriate framework for these patients, due to deficiency of adapted and reliable diagnostic criteria and the substantial lack of screening and assessing tools, would be considered the primary link of a chain of clinical, organizational, and structural issues. Moreover, in addiction treatment services, professionals are usually not trained for the implications that ID have on clinical course, adherence, and response to treatment, outcome, and specific multi-professional interventions recommended. This shortcoming may affect professionals' capability to receive, interact, and manage ID patients and, more in general, the suitability of the overall service for them. Furthermore, it has to be taken into account that ID individuals admitted to addiction treatment programs have cognitive impairments usually associated to poor verbal skills, memory limitations with scarce capacity to learn treatment information, and behavioral strategies besides discrimination difficulties [18]. These limitations preclude the full inclusion and participation to standard programs, the beneficial use of counseling procedures, and self-help groups.

There are few data regarding treatment strategies for alcohol and tobacco use in ID, and the few available regard only BIF and mild ID subjects. No data are available on the treatment of other drugs abuse [17]. Experts in this field agree that there is a need to develop specialized and integrated treatment approaches. Current literature reported the use of psychoeducation, motivational interviewing, and cognitive behavioral strategies with the aim to reduce substance consumption [17]. Interventions have to be adapted, for example, through the use of simplified

language and clearly understood concepts, pictorial stimuli, and role playing [16–18]. Motivational interviews are aimed to implement motivation to engage and adhere to treatment and to deal with ambivalence using cyclical model encompassing pros and cons of substance consumption and treatment; this is very helpful in anticipating relapsing and in increasing self-efficacy and determination to change substance-related problem behavior [69]. A variety of cognitive-behavioral techniques have been employed in addiction settings with the target of quitting the use of substances (e.g., stimulus control strategies) [13, 16, 18]. Interpersonal abilities, implicated in the development of SUD in ID, might be an intervention approach in some cases through the use of verbal rehearsal strategies, social skills, relaxation, and problem-solving skills training [18]. Educational interventions are aimed to expand the patient's knowledge about short- and long-term substance adverse effects on physical and mental health, possible legal consequences, and motivations for substance consumption. Psychoeducation is usually performed in small groups and, beyond the adaptation of communication techniques, it could be useful to deliver a higher number of short duration sessions [16, 17]. Psychoeducation has been found to be effective in reducing substance consumption in a small proportion of the cases [13] and it could be more effective in primary prevention programs. Some authors suggested that educational approaches would be less effective than contingency contracting and close supervision [22]. Otherwise, it is possible that a multimodal treatment including different strategies might result more effective but, to our knowledge, experimental confirmations are not currently available. More recent findings indicate that abstinence through restriction would be the best choice compared to rewarding behavioral strategies [28, 32]. Restriction is a well-known paradigm in addiction services for non-ID substance users and its application even for ID patients makes sense considering that living independently, isolation, and lack of supervision are among the most important independent risk factors for the development of SUD in this population. However, such an approach opens some ethical and practical concerns because the safeguard of self-determination and empowerment represent fundamental objectives in developing rehabilitative projects in ID services [61]. The conflicts of the two opposite paradigms may result in low cooperation between ID and addiction services with potential harmful consequences on the health of ID addicted patients [32]. Another important issue is the treatment of co-occurring psychiatric disorders, particularly mood disorders: achieving the psycho-affective balance can be in some cases the fundamental prerequisite to proceed to further treatment steps. The emerging perspective of individualized and personalized medicine is going in this direction and could represent the happy medium in clinical practice: in fact, when a correct framework of the clinical picture has been performed, a hierarchical approach can be adopted independently from IQ. Further, methodologically well-conducted studies are needed to identify risk and prognostic factors and treatment outcomes to correctly stratify interventions.

The aim of treatment may vary considerably depending on the type of substance (licit or illicit) and the severity of abuse. For example, in presence of mild severity alcohol use without significant comorbidities, reducing the consumption of alcohol to more acceptable levels can be considered an adequate and feasible goal. On the other hand, the diagnosis of dependence and/or the co-occurrence of relevant physical or psychiatric disorders or severe complications would require to rapidly achieve abstinence [69]. Similar considerations are also valid for the majority of other substances. Current management guidelines for ID, following guidelines for average IQ substance users, identified a range of good practices. Brief verbal support must be the first step with the double aim to inform the patient and his caregivers and to obtain more in-depth information regarding the quantity, modality, and duration of substance consumptions and eventual consequences. When a SUD has been identified, the patient should be evaluated and followed in specific addiction services and a cooperation should be estab-

lished. To achieve abstinence, it is very important to manage withdrawal. Mild withdrawal may not require medical interventions but only monitoring and support, whereas severe withdrawal needs high and complex medical examination and care. For example, it has to be remembered that alcohol withdrawal may cause death in the 8% of people hospitalized for a withdrawal syndrome [81]. Emerging complications as well as seizures should be monitored or better prevented via the administration of anticonvulsants. The identification and treatment of emerging psychiatric symptoms (as insomnia, anxiety, depressive mood, etc.) or comorbid mental illness are considered an essential step. The treatment of co-occurring ADHD represents a particularly great management challenge, requiring a multidisciplinary evaluation and intensive monitoring. All these recommendations, valid for any person suffering from SUD, are even much relevant for ID population considering their particular neurovegetative, neurological, and physical vulnerability. Some medications used to treat acute withdrawal may be useful in the long-term to reduce craving and secondary abstinence. Unfortunately, substitute medications are not available for all the substances: benzodiazepine, gamma hydroxybutyric acid, and acamprosate are currently used to manage withdrawal and both primary and secondary alcohol abstinence. Similarly, the opioid agonist methadone and the partial agonist buprenorphine (with or without the addition of naloxone) are successfully employed to treat opioids and particularly heroin addiction. It has to be taken into account that the scarce control of craving may lead all other treatment strategies to fail. On the other hand, a careful risk-benefit analysis and the adaptation of standard dosages of medications are needed as ID patients are at higher risk to develop side effects. Adversative agents, such as disulfiram, acamprosate, and anticonvulsants in alcohol dependence, or naltrexone and naloxone in opioids dependence, are often used in average IQ cooperative patients to increase the motivation to abstinence and reduce the perceived effects of the substance of abuse. In ID patients, all these should be valid options, except for disulfiram, that is not recommended. Disulfiram mechanism of action and effectiveness require full cooperativeness and a complete understanding that all alcohol must be avoided. No studies are available regarding psychopharmacological treatment outcomes of specific categories of SUD in ID population.

Cognitive limitations often preclude the full inclusion, participation, and adherence to standard treatment programs. There is a paucity of studies on treatment strategies specifically addressed to ID persons. Psychoeducation, motivational interviewing, and cognitive behavioral strategies with the aim to reduce substance consumption are the most studied. They require adaptations. Results are promising but not conclusive. Psychopharmacology would help as in the general population. Unfortunately, no studies relative to this field are available.

30.6 Future Perspectives and Conclusion

SUD is a chronic and often severely impairing illness, with a relapsing course. This trend is mostly linked to the neurobiological general modifications commonly shared by all the substances of abuse and even behavioral dependence, regardless of the pharmacodynamic characteristics of each substance. Addiction involves the disruption of those neural circuitries implicated in reward, motivation, inhibitory control, and other executive functioning [82] such as limbic system, prefrontal, temporal, and cerebellar cortices [83–85]. As shown by epidemiological data, the prevalence of substance-related and addictive disorders is lower in ID population compared to the general one. However, considering only those ID patients with easier access to substances, the risk to develop a full-blown SUD after substance use is far higher than for average IQ substance user. These findings highlight that the presence of ID has to be considered an important vulnerability factor for SUD. It is probable that the neurobiological correlates of ID in single individuals with basic impairment of specific brain networks and cognitive functions may be

responsible of such a vulnerability (e.g., basic hypofrontality may be related to poor inhibition control, impulsivity, attentional biases). This aspect of addiction is of great interest in the field of ID. In fact, studies found that SUD involve in most cases individuals with BIF and mild-to-moderate ID. The analysis of risk factors suggests that this is, at least in part, due to the higher accessibility to substances that people with some independence and with lower cognitive and pragmatic impairments have. However, considering the neurobiological basis of addiction, it is possible to hypothesize that even those with higher severity degrees of ID may develop addictive behaviors. This consideration could be supported by neuroimaging and neurofunctional studies that have demonstrated that the alterations of brain networks commonly found in "classical" addictions are found even in other pathological conditions. This is the example of uncontrolled eating and obesity. There is a great vulnerability and neural disruption overlap between SUD and uncontrolled eating similarly involving dopaminergic modulation of dorsal and ventral striatum, and the above-mentioned brain regions, specifically anterior cingulum, supplementary motor area, medial orbito-frontal cortex, hippocampus, and parahippocampal gyrus. Inborn or acquired alterations impair top-down control that is necessary for self-regulation [86–88]. Indeed, uncontrolled eating also shows wide overlapping clinical areas, especially considering behavioral profiles of affected persons. It is also possible that addiction may manifest in severely ID individuals in an atypical way including a wide variety of behaviors and behavioral equivalents of craving would be the most important parameter to make diagnosis of addiction in ID population. In this perspective, addiction could be more frequent than expected in ID people.

SUD is not an outlying issue in ID psychiatry because, when co-occurring, it has a destroying impact on cognitive, health, behavioral, and functional outcome. By now, in both clinical and research fields, this issue has been neglected with negative consequences on the validity and reliability of diagnosis and interventions for this vulnerable subpopulation. Further studies in this direction are needed.

Tip

The evaluation of addiction in ID as well as in average IQ persons should also include a full psychopathological evaluation. Co-occurring psychiatric disorders or psychopathological dimensions may increase the vulnerability to SUD, the difficulties to treat the SUD and relapses of SUD. Specific interventions should have double purposes and should consider multiple outcomes not only to ameliorate the general mental health but also to indirectly reduce and contain the substance consumption and more in general addiction.

Key Points

1. Substance-related and addictive disorders have been reported to reach 20% prevalence rate in ID population, with the highest rates in BIF and mild ID.
2. ID people seem to be particularly vulnerable to develop substance-related and addictive disorders and severe physical, mental health, and behavioral consequences after substance consumption.
3. The co-occurrence of psychiatric disorders may increase the risk, severity, and chronicity of substance-related and addictive disorders in this population.
4. Currently, there is a wide fragmentation of ID, addiction, and psychiatric services. Often, ID substance users do not receive a correct diagnosis neither correct interventions for substance-related and addictive disorders.
5. Assessing tools and treatment strategies for substance-related and addictive disorders should be adapted to meet the needs of ID population.

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Personality Disorder in People with Intellectual Disability or Those with Intellectual Disability and Autism Spectrum Disorder

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Contents

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Learning Objectives

1. Personality disorders (PDs) are important moderators of mental state and physical disorders, with significant impact on psycho-social functioning and ability to live productive lives in the community.
2. The diagnosis of PDs in persons with intellectual disability (PwID) or persons with intellectual disability and autism spectrum disorder (PwID-ASD) is important because it may affect the person's acceptance into community placements, predict subsequent psychiatric disorders, determine the rate of referrals to psychiatric services, and significantly influence the mode of management.
3. The diagnosis of PDs in PwID or those PwID-ASD is complex due to difficulties in teasing out maladaptive patterns of cognition, emotion and behaviour from those which are integral to the intellectual disability or autism.
4. The diagnosis can be facilitated by greater use of behavioural observation together with informant information.
5. In spite of a growing evidence base, there is a dearth of good quality studies in this area. Treatment strategies are still largely derived from the research evidence in the general population.

31.1 Introduction

Personality is characterised as the set of habitual behaviours, cognitions and emotional patterns that evolve from biological and environmental factors and so define an individual [1]. Due to wide variation in these factors, an individual's personality is unique and cannot be adequately summarised by any classification system. As a consequence, all systems are found wanting in some way. This is even more of an issue in people with intellectual disability (PwID) or those with both intellectual disability and autism spectrum disorder (PwID-ASD).

Personality disorder (PD) is usually defined as an enduring and pervasive disturbance in how individuals experience and interpret themselves, others and the world, that results in maladaptive patterns of cognition, emotional experience, emotional expression and behaviour. These maladaptive patterns are relatively inflexible and are associated with significant problems in psychosocial functioning, which are particularly evident in interpersonal relationships. The disturbance is manifest across a range of personal and social situations and is thought of as being of long duration, typically lasting at least several years. Its features are usually shown first in adolescence but may appear later in life [2].

In the last two decades, the utility of the categorical diagnosis for PD has been debated at length, sometimes quite robustly. Thus, the DSM-5 while retaining the categorical approach to diagnosing personality disorders also proposes an alternative diagnostic system based on a dimensional approach and organised into four stages [3]. The first two stages consist of identification and severity assessment of the deficits in self (identity and self-direction) and interpersonal functioning (empathy and intimacy). The third stage compares the personality traits of the person under evaluation with those of six PD types (antisocial, borderline, narcissistic, schizotypal, avoidant and obsessive-compulsive), which have been retained from the original ten in DSM-IV for the highest availability of research data. The fourth stage includes the identification of maladaptive personality traits, which differ from symptoms for far less fluctuation across time and for continuity with healthy and adaptive characteristics of personality. A trait is considered maladaptive to the extent it interferes with individual functioning and wellbeing, which may depend on cultural and societal factors. Nonetheless, research has consistently validated and replicated five broad personality domains, sometimes called 'The Big Five' Model of personality [4]. The heritability estimates of these Big Five personality traits based on common genetic variants have been described [5], and their pathological aspect has been adopted by the DSM-5 task force for the identification of PD maladaptive traits (see Table 31.1).

Table 31.1 Pathological personality traits

Healthy polarity	Pathological polarity	Pathological facets
Emotional stability	Negative affect	Emotional lability Anxiousness Separation anxiety Submissiveness Hostility Perseveration
Extraversion	Detachment	Withdrawal Intimacy avoidance Anhedonia (lack of enjoyment) Depressivity Restricted affect (limited emotional range) suspiciousness
Agreeableness	Antagonism	Manipulativeness Deceitfulness Grandiosity Attention seeking Callousness Hostility
Conscientiousness	Disinhibition	Irresponsibility Impulsivity Distractibility Risk taking Rigid perfectionism
Lucidity	Psychoticism	Unusual beliefs and experiences Eccentricity Cognitive and perceptual dysregulation

Utilising recent advances in molecular biology and epigenetics, Svrakic and Cloninger [7] proposed a simpler definition of personality disorders as 'maladaptive syndromes developed through person-environment interaction' whereby maladaptation is a failure of integrative functions of personality (i.e. those that carry out adaptive processes) caused by strong biogenetic dispositions or by pathological environmental effects, or both. However, no biomarkers of any sort have been linked clearly to personality status. Despite being common and universal in all medical settings, and an important moderator of mental state and physical disorders, with high societal costs [8, 9], personality disorders often remain undetected or ignored in clinical practice [2, 10, 11].

Knowledge about personality disorders in PwID is sparse, significantly hampered by methodological shortcomings and poor-quality data. This reflects the lack of conceptual clarity relating to the fundamental validity of constructs of PD in intellectual disability (ID) [6, 12]. Teasing out which of the 'maladaptive patterns' of cognition, emotion and behaviour are those of personality disorder and which are of ID is a challenge that has not been met satisfactorily by any classification. Torr [6] concluded that there is 'a lack of conceptual clarity about the fundamental constructs of personality disorder in ID and the blurring of the boundaries between personality, psychiatric and behaviour disorders'.

The current research available on the impact of PD in PwID or PwID-ASD suggests

that its effects are likely to be wide ranging and significant. A community-based study carried out by Lidher et al. [13] demonstrated that compared with controls, intellectually disabled people with a PD were significantly more likely to have a maladaptive pattern of cognition, emotion and behaviour.

31.2 Diagnosis and Classification

As is evident from the preceding discussion, the classification of personality disorder is complex. A key issue, perhaps not unique in clinical psychiatry, relates to the diagnosis of a long-term condition in the absence of biological markers. Further, while a categorical classificatory system provides clear, vivid descriptions of discrete personality types, these categories are seldom 'pure' and most patients in real life clinical practice have elements of several personality disorder categories. Finally, personality evolves with time and requires longitudinal evaluation. All these make clear why the diagnosis of a PD is one of the most difficult tasks in clinical practice [2].

In PwID or PwID-ASD, the diagnosis and classification of PD becomes even more challenging and complex. Unlike those with average ability, where lasting personality characteristics develop by adolescence, the personality development phase in PwID can be prolonged. It is recommended that a diagnosis of PD should not be made before the age of 21 years in this population [14]. Nonetheless, many aspects of a person's personality structure are obvious during the developmental period, and therefore, in the proposed ICD-11 classification, the diagnosis of 'PD in development' can be made in childhood or adolescence.

In people without ID, the diagnosis of PD requires subjective information about thoughts and emotions and therefore demands an ability to communicate adequately. Such information is difficult to elicit in those with severe ID. Physical and sensory problems may compound this difficulty [15]. Consequently, a particular pattern of behaviour diagnosed as 'personality disorder' in those with mild or moderate ID could be perceived as a 'behavioural disorder' in those with severe or profound disability [12]. Guidance specific to ID emphasises that the diagnosis of personality disorders is unlikely to be accurate in those with severe or profound ID [14]. In a comparative study of ICD-10 and ICD-11 personality status in PwID, those with challenging behaviour were more likely to be diagnosed as having personality disorder than others [16].

PwID or PwID-ASD often display other behaviours that overlap with features of personality disorder (see Table 31.2). Zigler and Burack [17] noted that personality development in PwID mirrors the general population, but negative life experiences common to this group can lead to the development of personality traits such as dependency on others, fear of failure and 'outer-directed' problem-solving styles, considered among the several features of PD. PwID also experience numerous failures and negative self-worth compared with their non-disabled peers. It is this which led to Zigler and colleagues to suggest seven personality dimensions, based on motivation to interact with the environment for PwID: positive reaction tendency (a heightened motivation to both interact with and be dependent upon a supportive adult), negative reaction tendency (initial wariness shown when interacting with strange adults), expectancy of success (the degree to which one expects to succeed or fail when presented with a new task), outer directedness (tendency to look to others for the cues to solutions of difficult or ambiguous tasks), effectance motivation (the pleasure derived from tackling and solving difficult problems), obedience (individual follows directions) and curiosity/creativity.

Furthermore, the psychiatric diagnostic manuals specific for ID give only partial indications on the phenomenology of PD, and generally they lack adaptation of the diagnostic criteria for the general population. A recent development in this regard is the Diagnostic Manual–Intellectual Disability (DM-ID-2) which contains notes that help professionals to consider some important variables at the time of diagnosis and the differentiation

Table 31.2 PwID/PwID-ASD and PD: overlapping features

Symptoms	Characteristics often associated with ID/ID-ASD	PD	DSM-5 alternative model for PD
Suspiciousness and/or inhibition and/or restricted range of expressions in relationships	Consequence of the many restrictions, exclusions and frustrations to which many persons with ID are exposed in daily life	Paranoid Schizoid Schizotypal Antisocial Avoidant	Negative affectivity (hostility) Detachment (suspiciousness)
Fear or avoidance of certain situations	Emotional vulnerability 'Outer directed' problem-solving styles Repeated negative experiences Learned helplessness	Avoidant Dependent	Negative affectivity (separation insecurity)
Lack of empathic skills or difficulties in understanding societal rules, indifference, or lack of repentance	Specific form of ID and severity levels Co-occurrence of ASD	Schizoid Antisocial	Negative affectivity (hostility) Detachment (withdrawal; intimacy avoidance; suspiciousness) Antagonism (callousness) Psychoticism (eccentricity)
Dependency on others	Inability to do things independently Poor assumption of personal responsibility Lack of practise and experience in planning and decision-making Overestimation of the level of intimacy of a relationship, connected to a lack of awareness of the type of relationship and of the real emotional investment by the other person. Highly negative impact of the end of an important relationship, because of narrowness of the circle of friendships	Dependent Borderline	Negative affectivity (submissiveness; separation insecurity; submissiveness)
Self-harm or accesses of anger, rancour and aggression	Communication difficulties Poor introspection Low levels of Theory of Mind development Alteration of executive functions, such as inhibition	Antisocial Borderline	Negative affectivity (emotional lability; hostility) Detachment (depressivity) Antagonism (callousness) Disinhibition (impulsivity)
Anomalies of form and content in verbal and non-verbal communication	Very particular, and idiosyncratic communication style Atypical development of language Tendency to exaggerate or underestimate one's talents and results due to deficits of self-assessment, self-awareness and metacognition	Schizotypal Histrionic Narcissistic	Psychoticism (eccentricity) Antagonism (grandiosity; attention seeking)

between personality traits and characteristics attributable to cognitive and relational disability [18]. A list of behavioural and observable symptoms for the three PD clusters has been developed by Bertelli as part of a wider system to help clinicians in the psychopathological diagnostic screening [19, 20]. The main items of this list are reported in Table 31.3.

Alexander and Cooray [12] summarised the problems pertaining to the diagnosis of PD in PwID. These include differences between ICD-10 [21] and DSM-IV classifications [22], the confusion of definitions and different personality theories [23], the difficulties in distinguishing personality disorders from late effects of childhood psychosis [24],

Table 31.3 Main behavioural and observable symptoms of cluster A, B and C personality disorders in PwID or PwID-ASD

PD cluster	Main characteristics/pervasive patterns of behaviour
A. Odd/eccentric Paranoid Schizoid Schizotypal	Absence of adequate reactions to people's suffering or to the emotional climate of the environment Lack of interactive attitudes albeit in a socially adequate environment Aggressive and other non-aggressive but nonetheless harmful behaviour Avoidance or escape from specific objects, people or situations. Excessive self-esteem, belief of being capable of extraordinary performance compared to the real possibilities Distrust of others and/or supporters of unjustified hostility Limited emotional responses from the previous level Expiration of abstraction and logic skills No manifestation of pleasure for the activity or objects previously appreciated Irritation, opposition or fear in circumstances that do not justify such reactions Act as if she/he was another person or an object
B. Dramatic/erratic Antisocial/dissocial Borderline/emotionally unstable Histrionic Narcissistic	Irritation, opposition or fear in circumstances that do not justify such reactions Aggressive and other non-aggressive but nonetheless harmful behaviour Self-injurious or self-mutilating behaviours Decline of logical skills Loss of confidence with the known environment associated with fear or agitation Excessive self-esteem, belief of being capable of extraordinary performances compared to real possibilities Behaviours potentially capable of causing own death Absence of adequate reactions to the suffering of people or to the emotional climate of the environment Pervasive tendency to irritability Only temporary signs of satisfaction in relation to gratification Continuous requests Limited emotional responses from the previous level Interactions aimed primarily at deriving a secondary advantage The same confidence with all people Binge eating Act as if she/he was another person or an object
C. Anxious/fearful Avoidant/anxious Obsessive-compulsive Dependent	Irritation, opposition or fear in circumstances that do not justify such reactions Reduction in frequency or determination in moving away from habitual residence Avoidance or escape from certain objects, people or situations Limited emotional responses, lack of interactive attitudes albeit in a socially adequate environment Excessive indecision in choices and rigidity with respect to the placement of objects or the way of doing things

and the concern about adding another potentially stigmatising label of personality disorder to an already marginalised group cannot be overstated too. Alexander and Cooray [12] advocated strongly for the greater use of behavioural observation together with informant information when making a diagnosis of PD. The limited research available on PwID or PwID-ASD suggests that PD has broad and serious maladaptive patterns of cognition, emotion and behaviour [13, 25]. Despite the difficulties outlined, diagnosis is still significant because it may affect the patient's acceptance into community placements [15], predict subsequent psychiatric disorders [25], determine the rate of referrals to psychiatric services [15] and significantly influence the mode of management [26–30].

Making a diagnosis of PD in PwID or PwID-ASD is more difficult than in the general population. There are many reasons for this including sensory and communication difficulties, psychiatric illnesses, autism spectrum disorder, overlapping clinical features and a prolonged developmental phase for personality characteristics. Taking these, and the particular life experiences of the person, can aid in making a diagnosis.

31.3 Epidemiology

31.3.1 Prevalence

A screening survey in the general population carried out in 2009 across 13 countries by the World Health Organization using DSM-IV criteria yielded an estimated prevalence of around 6% for personality disorders [31]. Demographic and social factors were noted to influence the rate in some instances. Functional impairment was partially explained by comorbid mental disorders. A UK-based national epidemiological study utilising DSM-5 criteria reported that a majority of the study population showed some degree of personality 'difficulties' short of the diagnostic threshold. The estimated prevalence of the most complex and severe cases (including meeting criteria for multiple diagnoses in different clusters) was 1.3%. Personality symptoms even at low levels were noted to be associated with functional problems. In the new ICD-11 classification, the prevalence of personality disorder may be somewhat higher [32].

Research findings on the prevalence of PD in PwID or PwID-ASD can be contradictory. They are limited by the lack of large-scale studies with several works indicating different rates depending on the samples that were studied and the assessment tools that were used [33]. Goldberg and colleagues [23] observed the presence of PD in 90% of 384 Canadians with ID living in the community and in residential facilities, while previous and more recent studies indicate lower rates, between 0.7 and 35% [34, 35]. In a literature review, Alexander and Cooray [12] listed the wide-ranging prevalence rates in community and inpatient settings (1–91% and 22–92%, respectively) and commented that this difference was too large to be attributable to true variability. They highlighted the importance of utilising consensus diagnostic criteria based on developmental level, such as those outlined by the Royal College of Psychiatrists [14], though such criteria currently lack published studies on validity or reliability. Cooper et al. [36] identified PD using four approaches in a population sample of 1023 PwID. The prevalence rates were 1% using a clinical diagnosis, 0.8% using diagnostic criteria-learning disability (DC-LD), 0.7% using ICD-10 diagnostic criteria for research (DCR) and 0.7% using DSM-IV-TR criteria. Tyrer et al. [37] found a prevalence of 35% among 38 persons with borderline to severe ID living in the community. Kiani et al. [38] reported a prevalence of 2% in urban areas and 1.1% in rural areas, in reference to a sample of 2713 persons with mild to profound ID. Tsiouris and colleagues [39] found a prevalence of around 8% in a sample of 4069 persons attending specialised services for ID. Lindsay et al. [40] evaluated the internal consistency and factor structure of PD in PwID, analysing 164 males with ID from three forensic settings who were diag-

nosed with PD using DSM-III criteria. The overall prevalence of PD in this sample was 39.3%. The reliability data for each PD was acceptable, with the exception of schizotypal PD. Within forensic settings, Hogue and colleagues [41] showed that 55% of persons from high secure settings 10% in medium/low secure settings met ICD-10 criteria for PD, while Alexander and colleagues found that up to 50% of those in low and medium secure settings met the diagnostic criteria [29, 42]. The higher prevalence of PD has been noted not just in those with ID, but also in those with borderline intellectual functioning, with a prevalence rate up to 52.8% [43].

- In persons with ID and ID-ASD, the reported rates of PD show a wide range, both in community (1% to 91%) and inpatient settings (22% to 92%). This is perceived as too great to be attributable to true variability.

- A valid diagnosis implies the use of consensus diagnostic criteria based on developmental level, such as those outlined by the Royal College of Psychiatrists, though such criteria currently lack published studies on validity or reliability.

31.3.2 Prevalence of Specific Personality Disorders

The state of knowledge on the prevalence of specific PD is even more uncertain than that of PD overall. In the studies by Lindsay and collaborators in forensic settings [40, 41, 44] and through the use of DSM-IV criteria and the Standardised Assessment of Personality (SAP), antisocial PD was the most frequent PD (22.1 and 62.8%) followed by PD not otherwise specified (NOS) (9.8 and 30.6%), schizotypal (8.6%), paranoid (7.6%), schizoid (7.6%), borderline (7.6%), narcissistic (7.6%), histrionic (3.8%) and avoidant (3.3%). The preponderance of antisocial PD was confirmed by an ICD 10-DCR study on a sample of 430 persons, which found a prevalence of 59%, followed by emotionally unstable PD (28%), a combination of antisocial and emotionally unstable PD (10%) and anxious PD (3%) [45]. On the contrary, a previous study on a community sample using ICD-10 criteria and the SAP [46, 47] found antisocial PD to be one of the less common PD (3%), preceded by schizoid (10%), impulsive (7%) and paranoid (5%), and followed only by dependent (3%), histrionic (1%), anankastic (1%) and anxious 1% [15]. Another relevant DSM-IV-TR study by Wieland and collaborators [43] found PD NOS to be the most commonly diagnosed PD (19.1%), followed by borderline PD (8.7%), which represents together with antisocial PD the most complex PD within the DSM cluster of dramatic PD, also known as cluster B [3]. A higher prevalence of cluster B (antisocial, borderline, histrionic and narcissistic) PD in comparison to cluster A (paranoid, schizoid and schizotypal) and cluster C (avoidant, dependent and obsessive-compulsive) PD has been reported also by Bertelli and collaborators, with rates of 24.0, 18.01 and 15.5 per cent, respectively [19]. With specific reference to mild ID, Wieland and collaborators found cluster B PD to be the most common PD (9.2%) (antisocial 0%, borderline 7.9%, histrionic 0.7%, narcissistic 0.7%), but cluster A to be not diagnosed at all. Cluster C rate stood at 2.6% (avoidant 2.0%, dependent 0.7%, obsessive-compulsive 0%) and PD NOS at 21.7% [43]. In persons with ID and severe challenging behaviours assessed with the SAP, cluster A PD was the most common, followed by cluster B. Specific rates were found as follows: paranoid for 64%, schizoid 50%, dissocial 53%, emotionally unstable 50%, histrionic 33%, anankastic 22%, avoidant 50% and dependent 28% [48]. In persons with borderline intellectual functioning, PD NOS have been identified as the most common diagnosis of PD (28.1%), followed by cluster B (17.4%) (antisocial 1.7%, borderline 15.3%, histrionic 0%, narcissistic 1.3%) and cluster C (8.1%) (avoidant 2.6%, dependent 4.7%, obsessive-compulsive 1.3%), while cluster A PD was not diagnosed at all [43].

As is clear from the account above, there have been a number of contradictory findings in terms of how often the various PD are diagnosed. From a practising clinician's viewpoint,

cluster A PD often overlaps with other developmental conditions like ASD and cluster C PD overlaps with the realistic dependency needs that are often found in PwID. Therefore, it is the recognition and diagnosis of a cluster B PD which often has the most potential to affect treatment plans and therefore have the most clinical utility [45].

Most studies report cluster B PD to be the most common PD in persons with ID, followed by cluster C PD, although the state of knowledge on the prevalence of specific PD in this population is still uncertain. For the practising clinician, the diagnosis of cluster B PD often appears to be the one with most clinical utility.

31.4 Comorbidity

There is ample evidence that in the general population, personality disorders are commonly concurrent with a wide range of other mental disorders, usually impairing both function and outcome [29, 30, 48, 49, 50–52]. A meta-analysis conducted by Friborg et al. [53] to identify the proportions of comorbid PD in the mood disorders, bipolar disorders (BD), major depressive disorders (MDD) and dysthymic disorders (DYS) concluded that the risk of having at least one comorbid PD was high across all three mood disorders (BD = .42, MDD = .45), but highest in dysthymic disorders.

31.5 Assessment/Research Instruments Used in People with ID

A number of assessment instruments have been used in studies of personality disorders in PwID and are described as follows:

Standardised Assessment of Personality (SAP) – The SAP relies on an informant account to establish a diagnosis of personality disorder [25, 46–48, 54, 55]. The presence of three or more durable criteria establishes a personality trait. The diagnosis of personality disorder is established if this causes significant personal distress, or occupational or social impairment.

Reiss screen and Psychopathology Inventory for Mentally Retarded Adults (PIMRA) – The Reiss screen [56] is a screening tool for the detection of psychopathology in mental retardation. For those scoring above a threshold, the PIMRA [57], a checklist of psychopathological behaviour based on DSM-III, is administered.

Temporal-Lobe Personality Behaviour Inventory – This instrument is utilised in identification of abnormal personalities specifically associated with epilepsy [58].

'Clinical' diagnosis based on ICD [21] and DSM [22] – Most studies carried out on PwID have used 'clinical' diagnoses based on either the ICD or DSM systems to identify personality disorders. The standard categories within these classificatory systems have sometimes been supplemented with extra items, for example, immature and impulsive [24].

The International Personality Disorder Examination (IPDE) – The IPDE is a semi-structured clinical interview [59] that is compatible with the ICD and DSM systems. Its manual makes clear that it is not validated for PwID. However, it has been used in PwID [60], particularly when the degree of ID is quite mild.

Diagnostic Criteria in Learning Disability (DC-LD) [14] – The DC-LD which provides operationalised criteria for psychiatric disorders in PwID adopts a multi-axial, hierarchical approach to diagnosis. It suggests that the ICD-10 category of organic PD should not be used purely on the grounds that a person has ID, or a syndrome with an associated behavioural phenotype or epilepsy. It proposes a higher age threshold (over 21 years) for diagnosing PD and does not recommend using the categories of schizoid, dependent and anxious/avoidant PD. It emphasises that the diagnosis of PD in severe or profound ID is likely to be problematic. Initial diagnosis using the criteria for personality disorder-unspecified is suggested and if these are met, further sub-classification should be consid-

ered. It has been used in some studies that compare the prevalence rates based on different diagnostic criteria [36].

The Diagnostic Manual Intellectual Disability 2 (DM-ID-2), [18] which draws on diagnostic criteria from the DSM, allows clinicians to understand the variations in the way diagnostic criteria of the various PDs differ in PwID. It also sets out the alternate dimensional approach to personality disorders. There are few published studies using the DM-ID 2 criteria for PD.

A number of assessment instruments have been used in studies of personality disorders in PwID/PwID-ASD. The main ones are Standardised Assessment of Personality, Reiss screen, Psychopathology Inventory for Mentally Retarded Adults, International Personality Disorder Examination and Clinical Interviews based on standardised classificatory systems.

31.6 Aetiology and Risk Factors

There is no conclusive evidence about the aetiology of PD. As is the case with other psychiatric diagnoses, PDs have a complex aetiology involving genetic and environmental factors and gene-environment interactions. A number of possible causes and known risk factors have been suggested, which vary depending on the disorder, the individual and the circumstances. Flynn et al. [48] found preliminary evidence that the diagnosis is associated with abuse in childhood. Ferguson [61] conducted a meta-analytic review of evidence from behavioural genetics, which supports the conclusion that a significant amount of the variance in antisocial personality and behaviour is due to genetic contributions. Roy et al. [62] studied the factors that shape personality and concluded that understanding how early-life experience and current-life situations give rise to personality traits and taking a developmental perspective, for example, mental age, could clarify the clinical presentation in PwID.

31.7 Treatment

The evidence base relating to the treatment of PD in PwID, though growing, is still rather limited. Johnstone [63] described a four-stage treatment process consisting of (i) assessment and motivational work, (ii) interventions including foundation treatments, offence-specific treatments and personality disorder symptom reduction treatments, (iii) consolidation or relapse prevention and (iv) discharge. This process became the basis for the framework of a comprehensive, multi-disciplinary, ten-point treatment programme [64–67] that could be used both within hospital and community settings (See ► Box 31.1).

Box 31.1 The Ten-Point Treatment Programme

1. A multi-axial diagnostic assessment
   - Degree of learning disability
   - Cause of learning disability
   - Pervasive developmental disorders (autism spectrum disorder)
   - Other developmental disorders (e.g. ADHD)
   - Personality disorders
   - Mental illness
   - Alcohol and/or illicit drug use
   - Physical health conditions
   - Psycho-social disadvantage (e.g. experience of trauma)
   - Types of challenging behaviour
2. A collaboratively developed psychological formulation
3. A behaviour support plan
4. Risk assessments
5. Pharmacotherapy
6. Individual and group psychotherapy
7. Offence-specific therapies, if relevant
8. Education, skills acquisition and occupational/vocational rehabilitation
9. Increasing community participation
10. Preparation for transition and discharge

The treatment of PD in PwID follows the same premise as the general population, with psychological treatment the mainstay of treatment. Most treatments are relatively prolonged and involve group and individual therapy modes, and there is little evidence that any one is superior to another [11]. Dialectical behaviour therapy, mentalisation-based therapy and cognitive behaviour therapy are the most commonly used, but almost all are indicated for borderline personality disorder [68]. Other therapies include person-centred therapy, group treatments and behavioural management strategies. None of these treatments have a robust evidence base in PwID and there is a compelling need for more research to establish whether these benefits extend to the ID population. Past objections have been replaced by innovation to accommodate the needs of people who have ID (see: Commissioning Team for the Faculties for Intellectual Disabilities of the Royal College of Psychiatrists and the Division of Clinical Psychology of the British Psychological Society, 2016) [69]. We list below the highlights regarding some treatment modalities and treatment outcomes.

31.7.1 Dialectical Behaviour Therapy

There are a limited number of studies on Dialectical Behaviour Therapy (DBT) with rather small sample sizes. Sakdalan, Shaw and Collier [70] described a 13-week programme for six patients and reported significant improvements on dynamic risk assessment scores. Morrissey and Ingamells [71] developed a 60-session programme for six people, four of whom showed some improvements.

31.7.2 Schema Therapy

Schema therapy (ST) is an approach deriving from cognitive behavioural therapy but which also has a parenting aspect. It has been used for the management of antisocial and borderline personality problems. ST integrates elements of existing models and techniques into a consistent case conceptualisation as the foundation for understanding and changing coping behaviours that are maladaptive.

31.7.3 Therapeutic Community

Morrissey and Taylor [72] have successfully applied therapeutic community treatment models resulting in significant reductions in antisocial, schizoid and paranoid traits and in schemas relating to entitlement, defectiveness, emotional inhibition and vulnerability in a forensic unit for people with mild IDs within a high secure psychiatric service. However, no randomised studies have been carried out.

31.7.4 Nidotherapy

Nidotherapy is an environmental form of management that systematically analyses the physical, social and personal environment of a patient and collaboratively makes changes that create a better fit [73]. It has been tested in two randomised controlled trials, in people with severe mental illness (schizophrenia with comorbid PD and substance misuse) and PwID and challenging behaviour. Benefits were shown for nidotherapy in symptom reduction, social function and cost in the first trial [74, 75] and in reducing challenging behaviour in the second cluster randomised trial [76]. Nidotherapy is well accepted by patients and can be given by relatively inexperienced practitioners. It may have particular value in PwID who also have PD [77].

31.7.5 Education and Occupational/ Vocational Rehabilitation

Smith and colleagues [78] have described a work-based learning programme designed for patients within a forensic hospital for PwID. They describe the partnership between the hospital, a work place and a further education college which became a crucial part of the patient's treatment programme.

31.7.6 Pharmacotherapy

People with PD receive psychotropic medications much more frequently than any other diagnostic group despite a poor evidence base and the absence of a clear understanding of the neurobiological underpinnings of personality. Most of the available evidence focuses on borderline [79] and schizotypal PD, with some additional evidence concerning the treatment of avoidant and antisocial PD. In general, the evidence supporting pharmacotherapy is weak and the National Institute for Clinical Excellence (NICE) guidelines does not recommend drug treatment except in a crisis, after which the drugs should be stopped [80]. This conclusion has been challenged by Leib et al. [81] who concluded that the evidence from randomised controlled trials suggests that drug treatment, especially with mood stabilisers and second-generation antipsychotics, may be effective for treating a number of core symptoms and associated psychopathology.

In PwID and PD, it is important to have a careful diagnostic evaluation which will clarify the presence of any mental illnesses, many of which may present atypically. This will avoid the risk of diagnostic overshadowing and ensure adequate treatment of comorbid mental illnesses [82]. If clinically indicated, short-term treatment to target four predominant symptom clusters – behaviour dyscontrol, affective dysregulation, anxiety symptoms and psychotic symptoms – has been described. This should be subject to strict monitoring of the following standards: (a) recording all the diagnoses, (b) initiating medication only as part of a multidisciplinary treatment package, (c) clearly identifying the predominant symptom complexes being targeted, (d) identifying and recording in consultation with the patient the expected improvements and behavioural targets, (e) discussing and recording the rationale, effects and potential side effects of the proposed treatment, (f) having regular follow-up appointments to monitor progress on the expected changes and (g) specifying a length of time that the person will be tried on medication with the plan to stop if there is no improvement [64, 83–85]. The development of rational pharmacotherapy necessitates increased understanding of the neurobiological underpinnings of personality disorders and their component dimensions as a prerequisite. Translating personality theory and social cognitive neuroscience into increasingly specific neurobiological substrates may provide more effective targets for pharmacotherapy in the future [86].

31.7.7 Long-Term Treatment Outcomes

There are a limited number of longer term treatment outcome studies. While there are many clinical anecdotes about particular difficulties in this group and suggestions that treatment may need to be more prolonged than in those with average intelligence [87], there is no evidence to show that treatment outcomes are poorer. Alexander et al. [29] reported no significant differences in the clinical outcome of those with a PD treated within a forensic ID hospital. Later in a study comparing three distinct groups within forensic hospitals in the UK [30] (those with ID alone, those with ID and PD and those with PD alone), they found that the ID group and the ID-PD group tended to stay longer in hospital than the PD group, but once discharged into the community had better long-term outcomes in terms of violent re-offending.

Tip

While there is a growing evidence base about treatments and treatment outcomes for PD in PwID, there is a compelling need for methodologically sound research projects to establish whether the benefits identified in the general population can be extended to this group too.

Key Points

- The diagnosis of personality disorders in PwID or PwID-ASD can be complex, but it is clinically relevant for a number of reasons.
- It would be prudent to limit the diagnosis to those with mild rather than severe degrees of intellectual disability.
- The recognition and diagnosis of a cluster B PD (borderline or antisocial) has the most potential to affect treatment plans and therefore the most clinical utility in this group.
- Diagnosis relies on a combination of patient and informant interviews as well as behavioural observations.
- Treatment approaches have to start with a comprehensive diagnostic assessment and psychological formulation and can include a range of psychological and pharmacological interventions, many adapted from those used in the general population.
- While there are many clinical anecdotes about particular difficulties in this group, there is no evidence to show that treatment outcomes are poorer.

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Co-Occurrence and Differential Diagnosis

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Contents

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Learning Objectives

The primary objective of this chapter is to provide a detailed description of the differential diagnosis processes among intellectual disability (ID) and/or autism spectrum disorder (ASD) and psychiatric disorders. This chapter also provides the examination of the current categorical and dimensional approaches to diagnosis and a discussion of the neurodevelopmental perspective as a theoretical framework linking ID, ASD, and psychiatric disorders.

Introduction

The identification, co-occurrence, and differentiation of borderline intellectual functioning (BIF), mild intellectual disability (ID), and autism spectrum disorder (ASD) in adulthood represents a diagnostic challenge for several reasons: (1) the diagnosis has to be formulated in part on behavioral data that are commonly more complicated to be measured compared to the presence/absence of genetic or biochemical data; (2) behavioral manifestation can evolve over time, meaning that the phenomena to be observed could need a time span ranging from infancy to adulthood to fully tackle the entire symptomatology; and (3) the possibility of comorbidity with other psychiatric or neurological conditions could "mask" the clinical background of the patient especially in adult individuals.

32.1 Differential Diagnosis

32.1.1 Issues Specific to ASD

Although studies regarding the clinical course in adults with ASD are still few compared to the ones performed during the last 25 years in children and adolescents [1], it is widely recognized by the scientific community that a first-time diagnosis of ASD in adulthood is frequently characterized by a more subtle presentation, a higher verbal competence compared to the individuals diagnosed during childhood, and a general good or high level of psychosocial functioning [1]. Indeed, the most prominent impairments in first-diagnosed adults with ASD appear to be in non-verbal communication and in some aspects of social reciprocity [2], with less evident repetitive behaviors or stereotyped interests. Accordingly, studies have shown that of all ASD symptoms, repetitive behaviors are the clinical manifestations that tend to decline most strongly with age [3]. To mention other confounding factors that may complicate an ASD diagnosis in adults are the comorbidities often carried by those individuals, considering that several studies have shown that 50–70% of adults receiving a first-time diagnosis of ASD also have or develop a comorbid anxiety disorder, depression, or other psychiatric disorders [1, 4, 5]. Therefore, the differential diagnosis for ASD in adulthood is broad and subtle, encompassing first of all anxiety and mood disorders, personality disorders, psychotic disorders, but also ID and BIF.

In summary, diagnosing ASD in adulthood often represents a challenge in clinical practice due to several aspects linked to the presentation of the symptoms of the condition, to the coping strategies of the individual and to the possible comorbid psychiatric manifestations that risk masking some aspects of the disorder.

32.1.2 Issues Specific to ID and ASD

Both ID and ASD are metasyndromic groups including several and different clinical conditions [6]. The difficulties in defining the diagnostic boundaries between ASD and ID are largely documented [7–9]. Some studies report that a 30–40% of people with ID present also pervasive autistic traits [10–12], whereas a 70% of patients with ASD are also affected by ID [13–19]. The boundaries between ID, ASD, and schizophrenia spectrum disorders (SSD) (i.e., schizoid and schizotypal personality disorders and schizophrenia) are also more difficult to define, and they constitute an important issue considering the high co-occurrence of schizophrenia and ASD [20, 21], especially in people with ID [12, 22]. Such complexity has been associated to their broad phenotypes that overlap over the spectra [23], the similar cognitive dysfunctions [24, 25], alterations of brain regions [26], dysregulated neurotransmission [27, 28] and shared genetic factors [29–31].

Interestingly, both ASD and schizophrenia have been described as common-sense disorders [32, 33]. Specifically, Minkowsky made a distinction between "autisme riche" and "autisme pauvre," indicating with the former an intensive attempt to find meanings in the environment that is also peculiar of what is defined as productive schizophrenia, whereas the latter refers to a prevalent lack of contacts with the external world and that is closest to the current concept of autism.

There is evidence of the higher rate of psychotic disorders in adults with ID in respect with the general population, with some studies reporting a rate of 3% for schizophrenia in ID in comparison to the 1% in people without ID [12, 34, 35], and others suggested a comorbid SSD in 4.4% of people with ID [12].

However, in a context of a dual diagnosis (i.e., a developmental disorder such as ASD or ID and psychiatric disorder), there is evidence of a general tendency to underestimate ASD in people with ID in presence of a diagnosis of schizophrenia [9, 36, 37]. Research showed that about 40% of the items which examined a potential psychosis obtained a high score in presence of autism [38]. Interestingly, in a 20-year follow-up study, authors found that in 20 adults who received as children a diagnosis of "childhood schizophrenia" (infantile autism), their symptoms did not change over time [39]. Moreover, although none of them met Schneider's criteria for schizophrenia (i.e., "first rank" symptoms), they all met Feighner's criteria for schizophrenia [40], which coincide with Kraepelin's dementia praecox, Crow's type II schizophrenia (defect state) and DSM III [41] residual schizophrenia [39].

Data collected from a survey conducted by the National Institute of Mental Health on a sample of subjects with childhood-onset schizophrenia found that a 21% of participants presented a lifetime diagnosis of ASD-not otherwise specified [42, 43], and some authors found that a 26% of adults psychiatric patients had undiagnosed ASD and received instead a diagnosis of schizophrenia [44]. It has been also described a subset of patients affected by a complex neurodevelopmental disorder characterized by impairments crossing the diagnostic categories that received the name of "autism-plus" disorder [45].

It is important to note that the complex diagnosis in people with ASD and ID is essential also in areas where there are concerns about mental-health and the management of psychiatric comorbidity.

It is well known that individuals with ID are at greater risk of hospitalization for behavioral and mental distress, and this has been associated to the difficulties in recognizing co-occurrent ASD or other psychiatric disorders [46]. Problem behaviors (PBs) and autism have been found as the strongest predictors of hospital admission [47, 48], and they resulted frequently associated with mood disorders and aggressive behaviors. [49]

Therefore, there is a considerable amount of data confirming the extreme complexity in the detection of psychiatric illness because of the problematic distinction of psychiatric symptoms from the autism core symptoms and ID [22, 50, 51].

It is now clear that the assessment of mental health problems in individuals with ID and ASD raises several theoretical questions and methodological dilemmas, and this is due to the definition and classification of mental health problems and to the nature of psychiatric assessment [52, 53], in consideration that the mental illness presentation in ID and ASD is often atypical [9].

Data from literature showed that mood disorders [54], anxiety disorders, [55] SSD, and impulse control disorders [56–59] are the most frequent psychiatric conditions in people with ASD and ID, and teens with autism and ID seem to present increased rates of inattention, hyperactivity, and impulsivity compared with people affected by ID alone [60]. Moreover, people with both autism and ID resulted more vulnerable to anxiety, mood, sleep problems, organic syndrome, stereotypies, and tics [56].

In this scenario, the investigation of behavioral phenotypes may help to clarify mental-health issues in some developmental conditions. For example, genetic studies found a statistically significant comorbidity between Fragile X syndrome and ASD, with a prevalence rate ranging from 7% to 25% [61–64]. However, neither the particular

psychiatric features of Fragile X syndrome nor correlations with age or genetics have been investigated. The increased occurrence of psychiatric disorders in individuals with Fragile X syndrome has been confirmed also by the research group of the Weinberg Centre for Child Development at Tel Hashomer in Israel [65]. Indeed, findings showed an occurrence of specific phobias and vocal tics in about half of participants, whereas the criteria for autistic symptoms and schizoid personality were met by one third of them.

It is evident how a more accurate assessment process of both ASD and ID could provide a better understanding of the "psychiatric" presentation and of the associated mental distress [7, 9, 54, 66].

A first-time diagnosis of ASD in adulthood can be challenging, since it is frequently characterized by a more subtle presentation and a general good or high level of psychosocial functioning. The difficulties in defining the diagnostic boundaries between ASD and ID are also largely documented, and the boundaries between ID, ASD, and psychiatric disorders can be even more challenging because of the problematic distinction of psychiatric symptoms from the autism core symptoms and ID.

32.1.3 Issues Specific to ASD and Schizophrenia

Schizophrenia is characterized by both positive and negative symptoms. Positive symptoms include thought content not based on reality, such as paranoid delusions, and perceptual disturbances, such as auditory and visual hallucinations. Therefore, positive symptoms are considered as the expressions of a loss of ability to participate with other people in a shared sense of reality. Negative symptoms include, among others, avolition, or loss of motivation, and blunted affect, or loss of emotional expressivity in facial expressions, gestures, and tone of voice. Negative symptoms are not overtly psychotic. They are evidence of the loss of aspects of the sense of self. A given individual with schizophrenia can display any combination of symptoms – predominantly positive, predominantly negative, or a fairly balanced mix of both.

The presentation of schizophrenia with predominantly negative symptoms can appear similar to ASD. Negative symptoms can be described in a variety of ways, and there is a large body of research devoted to how to best characterize them. According to the most recent National Institute of Mental Health consensus statement in 2005, negative symptoms are most accurately categorized into five symptom domains: blunted affect, alogia (or poverty of speech), asociality (social withdrawal), anhedonia (loss of pleasure in activities), and avolition (loss of motivation) [67]. Studies have shown that although ASD and schizophrenia have different developmental trajectories, their clinical presentations at any given time can overlap [68, 69]. In particular, individuals with schizophrenia with predominantly negative symptoms show many of the same social deficits as individuals with ASD [68].

There are a few approaches that may be taken in trying to differentiate these two diagnoses. For a person to be diagnosed with schizophrenia, at least one of the following symptoms must have persisted for a considerable period of time for at least one month: delusions, hallucinations, or disorganized speech (e.g., "frequent derailment or incoherence") [70]. Delusions or hallucinations persisting for this amount of time do not typically appear in ASD. The disorganized speech in schizophrenia is qualitatively different from the stereotyped and repetitive or idiosyncratic use of language in ASD. If a patient with schizophrenia, however, presents during a period of time when negative symptoms are predominant and the patient's only history of positive symptoms is disorganized speech, it may be difficult to tease out by history-taking the presence of disorganized speech if only alogia, or poverty of speech, is present at the time of the examination. If, however, a clear history of disorganized speech can be provided by a family member or close friend, this information can help to point toward a diagnosis of schizophrenia and not ASD. Of course, it is important, as in every case, to take as detailed

as possible a history of childhood development because a patient with schizophrenia will not display prodromal symptoms as early as the patterns of ASD will appear in a child.

Although ASD and schizophrenia have different developmental trajectories, their clinical presentations at any given time can overlap and in particular, individuals with schizophrenia with predominantly negative symptoms show many of the same social deficits as individuals with ASD. In terms of differential diagnosis, SSD and ASD can be distinguished mainly by the interest toward others, the level of understanding others' meaning and emotions, the manifestation of positive symptoms, and the deficits in global cognition.

32.1.4 Issues Specific to Schizophrenia Spectrum Disorders and ASD in ID

A reliable diagnosis requires a great knowledge of target symptoms and a considerable attention to the details of differential diagnosis. Basically, ID is characterized by significant impairments in cognition, social, and adaptive behavior [71–77] and by the frequent presence of stereotypies, challenging behaviors, and autistic symptoms [78, 79].

The core symptomatology characterizing ASD consists of impairments in social interaction (e.g., inadequate eye contact, inappropriate peer relationships, lack of emotional reciprocity), impaired verbal and nonverbal communication (e.g., language delay, poor conversation skills, inability to play imaginatively), and a pattern of repetitive behaviors and restricted interests (e.g., repetitive motor movements, intense attraction to particular objects or parts of objects) [80–88]. However, some of these difficulties are also found in ID [17, 78].

As mentioned before, there is an increased risk to receive a diagnosis of schizophrenia spectrum and other psychotic disorders in this kind of population, and there is evidence showing that a first diagnosis of schizophrenia or psychosis tends to be confirmed over time [7–9, 89].

In order to provide a reliable and robust diagnosis of a psychotic disorder in individuals with ID, it is of extreme relevance the evaluation of the impact that the ID itself (e.g., cognitive fragmentation associated with stressful life events) and a possible coexisting ASD exert on the individual's clinical presentation [9, 90, 91], even with the consideration of the context in which the psychotic-like symptoms appear [92].

It is important to emphasize that some psychotic-like behaviors such as talking to one's self, regression, as well particular postures that may be assumed in response to stressful events, can be part of the ID symptomatology, also without ASD [93, 94]. Even some particular ways of experiencing the world in ASD that may result in a specific symptomatology, such as bizarre expression or fantastic thinking, may be confused with psychotic symptoms [37].

In this challenging situation, the careful evaluation of the individual's developmental history, the prodromes, the onset, and the course of the condition, the presence of positive symptoms has a central role for determining whether the clinical manifestations belong to the autistic or schizophrenic spectra or are the result of their comorbidity [95, 96].

Data documenting neurological and social development from infancy are of great value, in consideration that the abnormalities in social cognition typically present in ASD can be detected in the first year of age; conversely, in schizophrenia, they are evident at 7–8 years old and more frequently in teenage years.

In terms of differential diagnosis, it has been reported that SSD and ASD can be distinguished by the interest toward others, [97] the level of understanding others' meaning and emotions, the manifestation of positive symptoms, and the deficits in global cognition. As Crespi and Badcock [98] reported, individuals with ASD are characterized by an unusual disinterest in shared sense and others' thought, whereas those with schizophrenia try to interpret the other's thought and intentions, and such effort in understanding can become even an obsession. Furthermore, evidence shows that people with ASD generally lack of contact with other persons from the

infancy, and usually this can be evident in the first years of life; conversely, a progressive loss of contact appears to characterize individuals affected by schizophrenia [99].

According to literature, social cognition refers to a number of abilities encompassing the detection of biological motion, the identification of facial affect and metacognitive skills such as the theory of mind and the attribution of mental states [100]. The impairments in social cognition characterizing people with ASD adversely affect face processing, emotion recognition, eye gaze processing, face scanning processing, biological motion perception, dynamic social interactions, and interpretation of emotionally charged situations [100–107]. It is noteworthy that similar deficits, for example, impaired perception of emotions, theory of mind, perception of social cues, complex social judgments, face scanning processing, and biological motion, have been also reported in people with schizophrenia [108–113].

However, there is evidence that these two conditions can be differentiated considering the strategies that people affected use for evaluating events characterized by social and emotional contents. In fact, in individuals with schizophrenia, it has been described the so-called "jumping to conclusions" bias, when they have to evaluate emotional information [114]; instead, in people with ASD, a poor perceptual integration has been considered the cause of a dysfunctional decoding strategy [115]. Consequently, it has been elaborated a new theory conceptualizing ASD as an hyposocial disorder and schizophrenia as the opposite, because it is characterized by an hyper, even though dysfunctional, development of social attitude [98, 116].

In contrast with what is usually described regarding ASD, in ID social cognition appear less compromised in respect to the general intellectual capacity, and this pattern tends to remain stable during the lifespan. On the other hand, impairments in cognitive and social abilities that characterize schizophrenia appear to gradually increase over time and may present an exacerbation in early adulthood after the first psychotic episode [117]. In addition, delusions and hallucinations are typical symptoms of schizophrenia and generally are not present in ID as well in ASD [9]. Nevertheless, approximately 90% of children with autism have abnormalities in sensory perception affecting smell, taste, and also vision [118], and they can be mistaken for hallucinations. Also the presence of pretend/imaginary friends, relationships with a "transitional object" (when a toy is treated as a real friend), stereotypic preoccupations, concrete externalization of thoughts or conscience and pseudo-hallucinations should not be confused with hallucinations and delusions [119]. Other features that can help to differentiate autism and schizophrenia are the typical lack of flexibility and the disturbance in response to routine changes characterizing autism [97] and the bizarre behavior including stereotypical speech, echolalia, posturing, grimacing, and rigidity characterizing the catatonia spectrum. However, mild forms of catatonia may also occur in autism, since they have been observed in approximately 17% of adolescents and young adults affected [120].

In consideration of the current classification systems, it is very likely that the disorganized subtype of schizophrenia can be confused with autism, because of its core symptomatology represented by disorganized speech and behaviors, and flattened affect. This can be also the case for the residual type, because of the persisting negative symptoms after the psychotic episode [95].

It is possible to observe a mixed form of catatonia, autism and psychosis, when none of the single diagnoses could explain better the clinical presentation. If a single diagnosis is given, such as "schizophrenia," the standard pharmacological interventions usually do not produce the expected response [121].

A recent study revealed the presence of a relationship between autistic symptomatology and schizotypal traits, and interestingly autism symptoms resulted associated with negative, disorganized, and positive schizotypal ones [122]. It is noteworthy that individuals with an ASD diagnosis present some schizophrenia spectrum traits during the years of adolescence, and such overlap includes not only the negative schizotypal symptomatology, but also the disorganized and positive symptoms [123].

There is an increased risk to receive a diagnosis of SSD in the ID population. In order to provide a reliable diagnosis of a psychotic condition in individuals with ID, it is of extreme relevance the evaluation of the impact that the ID itself (e.g., cognitive fragmentation associated with stressful life events) and a possible coexisting ASD exert on the individual's clinical presentation, with the consideration of the context in which the psychotic-like symptoms appear. Indeed, some psychotic-like behaviors (e.g., talking to one's self, regression, as well particular postures that may be assumed in response to stressful events) can be part of the core ID symptomatology, also without ASD.

32.1.5 Issues Specific to ASD and Social Anxiety Disorder

Social withdrawal and poor socializing in public situations are behaviors that may be exhibited by adults with ASD but also by individuals with social anxiety disorder, and these manifestations may represent the primary reasons for which the diagnosis of either ASD or social anxiety disorder may be missed or delayed [124].

It has to be remembered that social anxiety disorder is characterized by the fear of the judgment by third parts, accompanied by the preoccupation to have an embarrassing or humiliating behavior in social contexts. Accordingly, those individuals choose to avoid personal encounters in order to circumvent intense physical and emotional discomfort frequently correlated to social stressful situations (e.g., fear, trouble concentrating, avoiding eye contact, trembling, sweating, and heart racing) [125]. Nevertheless, in one-on-one settings, the signs and symptoms of social anxiety are expected to abate to a major degree in individuals with social anxiety disorder compared to individuals with ASD, considering also that people with ASD do not always display all the physical signs (e.g., sweating and blushing) of anxiety in social situations that individuals with social anxiety disorder present. Another key point for differential diagnosis is the presence of sensitiveness to verbal and nonverbal signs of judgment that can guide the clinician to consider the presence of social anxiety instead of ASD since interpreting social cues is one of the main areas of deficit in individuals with ASD.

Behaviors that may be exhibited both by adults with ASD and individuals with social anxiety disorder are social withdrawal and poor socializing in public situations. Nevertheless, people with ASD do not always display all the physical signs (e.g., sweating and blushing) of anxiety in social situations that individuals with social anxiety disorder present. Moreover, the interpretation of social cues is more deficient in individuals with ASD compared to the ones that have only a social anxiety disorder.

32.1.6 Issues Specific to ASD and Major Depressive Disorder

Neurovegetative symptoms (changes in sleep, appetite, and energy) together with psychomotor agitation or retardation and lack of mood reactivity with a specific loss of pleasure in almost all activities (anhedonia) typically characterize the presentation of severe depression [70]. As a result, the individual may experience an evident personal and social impairment that resembles the characteristic behavior of individuals with ASD. Indeed, the presence of anhedonia can appear through behavioral conducts such as poor modulation of facial expression, lack of research of social interactions, lack of interests, absence of spontaneous seeking to share personal activities, and all of these manifestations resemble typical conducts of individuals with ASD. Moreover, psychomotor agitation or slowing can be found also in ASD individuals. Nevertheless, depressive illnesses are expected to have a definite time-frame and can be characterized by acute phases and remitting phases. As a consequence, in the differential diagnostic process, also the general functioning profile is expected to have an abrupt

decline linked to the acute phase of the illness, whereas functioning in ASD individuals is almost constant, except for the presence of a psychiatric comorbidity.

Evident personal and social impairment (e.g., poor modulation of facial expression, lack of research of social interactions, lack of interests, psychomotor disturbances) can be present in individuals with ASD and individuals with mood disorders. In the differential diagnosis process, the time-frame of symptom presentation and the general functioning profile are key elements to differentiate ASD from mood disorders presentations.

32.1.7 Issues Specific to ASD and Obsessive-Compulsive Disorder

Differential diagnosis between the presence of an obsessive-compulsive disorder (OCD) and ASD with the typical restricted and repetitive patterns of behavior, interests, and activities can be extremely difficult and lead to overdiagnosis of OCD in people with ASD.

OCD is characterized by the presence of obsessions and compulsions. Obsessions are defined as intrusive and recurrent thoughts or impulses causing severe anxiety, comprising obsession of contamination, ordering/counting, symmetry, intruding forbidden thoughts, harm. Compulsions are defined as rigid ritualistic and repetitive behaviors typically acted in order to relieve obsession-induced anxiety [70]. Such patterns of behavior, when observed by the clinician, may closely resemble the rituals, stereotypes, and adherence to routines of adult individuals with ASD [126]. In the differential diagnostic process, the key to proceed can be found mainly in analyzing the function of the obsessive-compulsive behavior and the emotional valence of thoughts and the compulsions. Indeed, restricted and repetitive behaviors represent pleasurable experiences in ASD individuals, intrinsically motivating and reinforcing the repetition [1]. First of all, generally the actions in ASD individuals lack the "neutralizing" character of true compulsions [127]. Moreover, those experiences are emotionally considered as "ego-syntonic" in this population, meaning that the individual lives the experience in a positive and not conflicting way when generating the behavioral conduct. On the other hand, individuals with OCD live the experiences related to the illness in an "ego-dystonic" manner, admitting that both thoughts and behaviors are irrational and without the possibility to stop them and not wanting to experience them anymore [128]. Finally, social cognition and empathy are mainly preserved in individuals with OCD, making therefore always suitable to explore the general functioning profile of the individual when considering a differential diagnosis with ASD.

There is a trend toward overdiagnosing OCD in individuals with ASD, since both conditions share restricted and repetitive patterns of behavior, interests, and activities. Key elements to proceed in the differential diagnosis procedures can be found mainly in analyzing the function of the obsessive-compulsive behavior and the emotional valence of thoughts and compulsions.

32.1.8 Issues Specific to ASD and Obsessive-Compulsive Personality Disorder

The differential diagnosis of ASD includes also personality disorders. For the case of obsessive-compulsive personality disorder (OCPD), repetitive and rigid patterns of behavior may resemble stereotypies of individuals with ASD. The differential diagnosis is even more complicated since OCPD individuals, unlike OCD ones, have impaired empathy and social cognition and often experience their compulsions in an ego-syntonic way [129]. Moreover, OCPD patients tend to not report the content of their thoughts as intrusive or upsetting [130]. Indeed, OCPD is described in the DSM-5 [70] as a condition involving impairments in self-functioning due

to a rigid obsession with productivity, morality, or other personal or social codes. For these individuals, self-imposed rules are believed to be the best way of behaving and they present lack of flexibility accompanied by a consequent perseverating pattern of conduct with rigid perfectionism [70].

Therefore, adults presenting rigid behaviors, lack in empathy, and social impairment should be necessarily considered by the clinician in the view of a differential diagnostic process between ASD and OCPD, with special attention to be given to the age of onset (adolescence or adulthood in OCPD, childhood in ASD).

Interestingly, researchers have stressed the presence of a significantly increased ability to delay gratification in individuals with OCPD, with people with OCPD exhibiting a hypercapacity for self-inhibition [130], while individuals with ASD tend to show a significantly reduced ability to delay gratification and exercise effortful control [131]. Consequently, as a second step, clinicians may perform tests of effortful control and reward delay to pursue the differential diagnosis, with high performances in these tasks suggesting the presence of OCPD.

Finally, impairments in social functioning confined to intimate or familial context with a well-preserved community general functioning may again suggest the presence of OCPD instead of ASD [132].

32.1.9 Issues Specific to ASD and Avoidant Personality Disorder

Avoidant personality disorder and ASD share similarities in their presentation particularly in the field of social impairment. Avoidant personality disorder is described as a "pervasive pattern of social inhibition, feelings of inadequacy, and hypersensitivity to negative evaluation, beginning by early adulthood and present in a variety of contexts" [70]. Moreover, a diagnosis of avoidant personality disorder in people with generalized social anxiety disorder represents a marker of severity of the condition [133] with "emotional guardedness," assessed primarily by response to the Structured Clinical Interview for the DSM (SCID) [134] scale item of "difficulty being open even with people you are close to," most consistently differentiating avoidant personality disorder symptoms in patients with generalized social anxiety disorder [133]. Starting from these premises, it appears that people with avoidant personality disorder may share with ASD individuals even more features than patients with social anxiety alone.

Apart from the individual report during the clinical interview, it would be of primary importance to observe patients' nonverbal communication in order to find behaviors more likely suggesting the presence of ASD or avoidant personality disorder. In the differential diagnostic process, age of onset of the impairments (adolescence or early adulthood), the absence of stereotyped or idiosyncratic language, and the presence of a certain attitude to social cues with suffering in interpersonal relationship would suggest the presence of the personality disorder instead of ASD.

32.1.10 Issues Specific to ASD and Schizotypal Personality Disorder

ASD and SSD have been at the center of a scientific controversy since Leo Kanner borrowed the term "autism" from Swiss psychiatrist Eugen Bleuler, who had previously coined to describe the socially withdrawn behaviors of individuals with schizophrenia [135]. Over the past years, many researchers have argued about the boundaries between the two diagnostic categories. Studies in the 1970s for the most part suggested that ASD and SSD were mutually exclusive categorical diagnoses with distinct developmental trajectories [123]. More recently, however, studies have shown empirical evidence for conceptual and phenotypic overlap [5, 123, 136].

In clinical practice, when considering personality disorders of the above-mentioned spectrum, severe difficulties emerge in the process of the differential diagnosis. Behaviors

performed by ASD individuals frequently overlap with schizotypal ones [137, 138], with data showing that 40% of ASD subjects can meet formal criteria for schizotypal personality disorder [122].

By definition, schizotypal personality disorder is described as "a pervasive pattern of social and interpersonal deficits marked by acute discomfort with, and reduced capacity for, close relationships, as well as by cognitive or perceptual distortions and eccentricities of behavior" [70]. Criteria for the disorder include odd or eccentric thinking, speech, and behavior, as well as "excessive social anxiety that does not diminish with familiarity and tends to be associated with paranoid fears rather than negative judgments about self." Nevertheless, criteria also include nondelusional ideas of reference, "odd beliefs or magical thinking that is inconsistent with subcultural norms," and suspiciousness or paranoid ideation and the exploration of these dimensions is critical in the differential diagnostic process, since ASD individuals generally do not show paranoid ideation or nondelusional ideas of reference. Indeed, individuals with ASD show impairments in the ability to conceptualize the mental state of other people and therefore might be defined as "hypo-mentalizers," while individuals with schizotypal personality disorder tend to overinterpret the mental state of others in a suspicious or paranoid way and might be called "hyper-mentalizers" [1, 122]. Apart from the presence of an interpretative way of thinking, as with all personality disorder, it will obviously help in differentiating patients with ASD from those with schizotypal personality disorder the exploration of the time-frame in which symptoms emerge, respectively, childhood or adolescence/early adulthood [1].

32.1.11 Issues Specific to ASD and Schizoid Personality Disorder

The term "schizoid," like the term "autism," was coined by Swiss psychiatrist Eugen Bleuler, who first used it in 1908 to describe an individual's exaggerated tendency to direct attention toward his inner life and away from the outside world. However, the existence of schizoid personality disorder as a discrete entity has been controversial since then, with the rising of the hypothesis that it could be seen as a personality trait rather than a personality disorder [139].

By definition, schizoid personality disorder manifests itself as "a pervasive pattern of detachment from social relationships and a restricted range of expression of emotions in interpersonal settings, beginning by early adulthood and present in a variety of contexts" [70]. Criteria for the diagnosis include "neither desires nor enjoys close relationships, including being part of a family"; "almost always chooses solitary activities"; "takes pleasure in few, if any, activities"; and "appears indifferent to the praise or criticism of others". Interestingly, schizoid personality disorder "should not be diagnosed if the pattern of behavior occurs exclusively during the course of schizophrenia, a bipolar or depressive disorder with psychotic features, another psychotic disorder, or autism spectrum disorder" [70].

Consequently, a subject cannot have a diagnosis of both ASD and schizoid personality disorder because the schizoid symptoms are considered to be etiologically rooted in ASD and not a personality disorder. Nevertheless, as with schizotypal personality disorder, studies have showed overlap between schizoid and ASD behavioral traits [137, 138]. In clinical practice, it must be noted that the lack of a paranoid dimension in individuals with schizoid personality disorder represents the main obstacle in the differential diagnostic process. Therefore, the first and principal step in considering the two diagnostic options is to obtain a detailed childhood developmental history of the subject, since behaviors raising during adolescence or early adulthood will be suggestive of the presence a personality disorder rather than ASD.

The differential diagnosis of ASD includes also personality disorders. Particularly, in the differential diagnosis process, it has to be paid special attention to the age of onset of the symptoms (adolescence or adulthood in personality disorders, childhood

in ASD) and the general functioning profile of the individual. Particularly, a subject cannot have a diagnosis of both ASD and schizoid personality disorder because the schizoid symptoms are considered to be etiologically rooted in ASD and not a personality disorder.

32.2 Categorical and Dimensional Approaches

The DSM-5 introduces an integration of a dimensional approach to diagnosis and classification together with the DSM-IV-TR [140] categorical approach [70].

Previous editions of DSM used a categorical model requiring a clinician to determine that a disorder was present or absent. The dimensional approach allows a clinician more possibilities to assess the severity of a condition and does not imply a real threshold from low-functioning forms to forms with very high general skills. All the psychiatric disorders in DSM-5 remain in specific categories, while measures indicating the degree of severity have been added to several combined diagnoses. Indeed, the DSM-5 combines four different categorical disorders in the ASD diagnosis and conceptualizes them as occurring along a single spectrum characterized by dysfunctional social communication and restricted, repetitive behaviors or interests. Using DSM-IV [141], people with such symptoms could be diagnosed with four separate disorders: autistic disorder, Asperger's disorder, childhood disintegrative disorder, or Pervasive Developmental Disorder-Not Otherwise Specified.

Given the phenotypic heterogeneity of this condition, the DSM-5 introduced the concept of neurodevelopmental disorders that include a range of disorders associated with learning difficulties, skills impairment, high reduction in logical-deductive intelligence, and significant difficulty in sharing emotional content [70]. The disorders included in this new metastructure are not primarily associated with a debut in infancy and adolescence, as described in previous classification systems, but from etiopathogenetic, risk, and clinical factors, such as the neuro-evolutive genetic phenotype, neural circuit anomalies, cognitive, learning, communicative and behavioral deficits, early onset, lifelong persistence, and the high mutual comorbidity [142].

The categorical approach and the dimensional approach both present advantages and disadvantages. The categorical approach is based on the kraepelinian and neo-kraepelinian perspective and refers to the process of dividing mental disorders into discrete entities, whereas the dimensional concept of spectrum, often used interchangeably to that of the continuum, includes a set of clinical forms that show a similar appearance or are thought to be caused by the same underlying etiological mechanism, differentiated according to increasing severity. The principal disadvantage of the categorical model is its tendency to encourage a "discrete entity" view of the psychiatric disorders. Conversely, dimensional approach introduces quantitative variation and graded transition between different forms of the disorder, as well as between normality and pathology [143].

Since 2009, a dimensional approach to neural circuits and their connections with psychopathology was promoted. The National Institute of Mental Health (NIMH) instituted the Research Domain Criteria (RDoC) project to develop a research classification system for psychopathology based on neurobiology and observable behavior.

The RDoC is based on three assumptions. First, the RDoC framework conceptualizes the mental illnesses as disorders of brain. While neurological disorders show identifiable lesions, mental disorders were considered as disorders of brain circuits. Second, RDoC assumes that the dysfunction in neural circuits can be detected by some screening and diagnostic tools employed in neuroscience, such as the fMRI. Third, the RDoC framework assumes that data from genetics research and clinical neuroscience will yield biosignatures that will augment clinical symptoms and signs for clinical management. RDoC is intended to support research toward a new classification that encourages investigators to approach to the study of the genetic, neural, and behavioral features of mental disorders [144–146].

In this context, advances in knowledge of early brain development highlighted the role of the neurodevelopmental perspective in theorizing about the etiology of disorders. Distinctive types of psychopathology may have early onset, features in common, and different time courses in symptoms depending on the variations of the same underlying disposition.

The categorical approach to diagnosis and classification of disorders is based on the kraepelinian and neo-kraepelinian perspective and refers to the process of dividing mental disorders into discrete entities, whereas the dimensional concept of spectrum, often used interchangeably to that of the continuum, includes a set of clinical forms that show a similar appearance or are thought to be caused by the same underlying etiological mechanism, differentiated according to increasing severity.

32

32.3 The Neurodevelopmental Perspective

Advances in the development of dimensional models and approaches to research on psychopathology made possible to find that behavioral, cognitive, social, and emotional competencies share common neurobiological bases, evolve across the brain development, and vary across individuals in different traits, abilities, or mental disorders [147].

Research in ASD and other neurodevelopmental disorders is in progress. How a damage in one system may lead to deficit in another or how different brain regions develop and interact with others remain largely unknown. Indeed, disruptions in specific cerebro-cerebellar circuits in ASD might interfere with the specialization of cortical regions involved in motor control, memory, attention, language, and social interaction, leading to deficit in these domains. Extensive connections within the brain provide an anatomical substrate for the broad range of clinical presentations of ASD and others neurodevelopmental disorders [148]. Genetic, anatomical, and functional imaging studies in ASD found an atypical organization of subcortical and cortical areas related to autistic symptoms. A damage occurring early in the brain development may induce a cascade of more complex deficits. Furthermore, neuronal circuits interact with others across the different brain regions. The cumulative consequences of these interactions among complex biological and also environmental systems can alter the course of brain development. Indeed, the international scientific community considers the extreme variability of symptoms as a polymorphism due to a complexity of different causal factors, both biological and environmental. Among the biological factors, genetic type has a predominant role. Association studies on the entire human genome (genome-wide) show that variations in the number of gene copies characterizing some syndromes including ID are also present in many forms of ASD and other major psychiatric disorders such as schizophrenia, bipolar, and major depressive disorder. A recent review conducted by Torres and colleagues [149] found that CNVs of 1q21.1, 3q29, 15q11.2, 15q13.3, 16p11.2, 16p13.1, and 22q11 chromosomal regions result in a wide phenotypic spectrum, ranging from normal development to learning problems, including ID, ASD, and SSD. Waltereit [150] and collaborators also analyzed the functions of genes mutated in these three disorders and selected some shared mechanisms in neurodevelopmental pathways, finding some similarities.

To note, emerging evidence suggests that ID, ASD, and also SSD are part of a single group of neurodevelopmental disorders [151]. Etiologically all the three syndromes are considered to result from the overlap between different combination of specific causative factors, both genetic and environmental that influence brain growth and maturation with subsequent effects on general and specific impairments of cognitive functioning [152].

In summary, the neurodevelopmental perspective is currently recognized as the most valid theoretical framework to explain the relationships between systems and to support multidimensional, transnosographic, interdisciplinary approaches, referring to the whole life, more useful to the understanding of the etiopathogenesis of mental health problems

and the identification of the best interventions [142, 153].

In fact, this perspective, which draws on the tradition of psychology and developmental psychobiology, and that has been enriched by evolutionary and cognitive neuroscience, localizes positive and pathological adaptation in the transactional relationships between the biological mechanisms of the brain, their psychological functions, and the external environment. Moreover, it represents a dimensional approach able to grasp the complexity of the differences between individuals, their evolution over time, the relations between mind and behavior, and the declinations of personal functioning in a better way than the categorical approaches and the relative quantitative diagnostic references.

Indeed, one of the most important advances linked to this perspective is to have recognized that psychopathological conditions traditionally considered distinct share common etiological and risk factors, and they occur simultaneously in the same person or at different times of life (sequential comorbidity), representing changes related to the age of the underlying common provisions.

Also dementia and other disorders traditionally considered to be neurodegenerative in nature – and that are often present in some genetic syndromes including ID – have also been suggested as conditions of late-onset neurodevelopment, as evidenced by an increasing number of genetic and neurobiological studies [154, 155]. Accordingly, a group of American researchers from the University of Wisconsin and Indiana recently found high levels of the beta-amyloid precursor protein, known so far only for its implications in the pathogenesis of Alzheimer's dementia, in people with Fragile X syndrome and with ASD [156].

While the prospect of neurodevelopment could contribute to a better understanding of the multifactorial etiopathogenetic mechanisms underlying many psychiatric and cognitive disorders and provide new intervention strategies, on the other hand it could lead to the premature abandonment of traditional nosology and the emergence of broad spectrum covering the full range of current psychopathology. For example, the trend of recent years to increasingly broaden the number of psychiatric aspects included in the autism spectrum [157–159] should be more appropriately evaluated in relation to the potential negative impact on research and clinical resources dedicated to the autistic syndromes closest to the descriptions of Kanner or associated with ID. In many countries, the ever increasing identification of ASD has been associated with a proportional reduction of ID diagnoses, with significant epidemiological, social, cultural, and economic implications [160]. Research on the differentiation criteria is desirable to undergo a progressive development, including new semeiological references, such as sensory-perceptual or motor disorders, and limiting the spread of conditions of uncertain nosographic validity, such as the high-functioning autistic disorder, the semantic-pragmatic disorder, the evolutionary disorder of the right hemisphere or the multidimensional impairment disorder.

Some members of the Psychiatry Section of the ID of the World Association of Psychiatry (WPA-SPID) have recently proposed a contribution in this sense, with the aim of achieving a better definition of the diagnostic borders between ID, ASD, and SSD; particular attention has been paid to the differentiation of ASD and SSD in people with ID [161].

The prospect of neurodevelopment has also provided the theoretical framework for the recent process of redefining ID, which is abandoning the reduction of the IQ, the deficits in adaptation, and the age of onset as the main diagnostic references. ID is not considered either as a disease or as a handicap, but as a metasyndromic group of health conditions associated with a variety of specific etiologies, which have in common a deficit in cognitive development prior to the acquisition of skills through learning and that significantly interfere with the activities and skills of participation [162]. Although many clinical conditions including ID can be diagnosed before birth or during the neonatal period, other syndromes may occur much later, in adolescence or even in early adulthood. Continuous adaptation throughout life and cognitive skills can develop sequentially over time, especially when optimal environmental supports and

stimuli exist [163, 164]. Much has been done to identify the periods in which the effect of experience has the greatest impact on brain development and the evidence collected shows interesting specificities for language, attachment, and behavioral pattern [165].

The introduction of an evolutionary and longitudinal perspective therefore seems to be able to improve the understanding of how the different presentations of ID and low-functioning ASD, with or without a known etiology, can change and evolve over time and how the different genotypes can determine specific cognitive behaviors and profiles in the different ages of life. For example, children with Down syndrome show better abilities in receptive language earlier than expressive ones [166, 167], while boys with Fragile X syndrome tend to acquire more skills in simultaneous processing with respect to sequential processing during development [168].

Further advances in this research area would allow to deepen the understanding of the dynamics of cognitive development and how to enhance the effectiveness of educational, therapeutic, and rehabilitative interventions, by better regulating their structure and chronology.

The model of intelligence, based on the measurement of IQ, appears to be of limited use compared to the definition of ID recently proposed in the DSM-5 [70] and in the eleventh edition of the ICD [162, 169]; people with ID present in fact an extreme variability of specific cognitive dysfunctions, even with IQ parity, strongly correlated to various bio-psycho-social factors and to the equally wide range of adaptive skills [170]. These impairments of a single cognitive function, or some, may have the effect of a neuropsychological overtone and determine low scores in complex executive performance. People with Down syndrome, for example, usually have deficits in specific areas of language, long-term memory, and motor performance [171], whereas in those with Williams syndrome, areas are more compromised of attention, visuo-spatial construction, short-term memory, and planning [172].

There is much evidence on how the limits in functioning, behavioral problems, and various alterations of instrumental examinations of the central nervous system correlate more with deficits in specific cognitive functions than with IQ or its complex subdivisions [173, 174].

In the literature on ID, the most investigated cognitive functions were the associative memory, the working memory, the orientation response, and the attention switch. Although there is not yet a conceptual map or hierarchy of relevant cognitive functions and domains in ID, some of these are compromised more often and more markedly, such as perceptual reasoning, operational memory, processing speed, and verbal comprehension [170].

The ICF [175] already considers intelligence as a generic concept, which includes cognitive functions, adaptive behavior, and learning and which depends on age and cultural standards. Also the DSM-5 [70] has abandoned part of the over-reliance traditionally placed on the IQ in favor of the expression of executive functions in everyday life, suggesting that the determination of levels of gravity of intellectual functioning, previously based only on the reduction of IQ, should have as main reference the level of adaptive functioning, assessed through conceptual, social, and practical skills [70, 176].

In the perspective of neurodevelopment, the impairment of cognition should be evaluated with the highest possible level of specificity, to precisely identify the dysfunctions that have the greatest impact on individual behavior, skills, adaptation, autonomy, and quality of life, during all of life [176]. It is desirable that specific neuropsychological investigative tools are progressively adapted to all levels of ID severity, spread rapidly in clinical practice and favor the establishment of a new concept of "functional neurocaracterization" [176].

Also, the neurodevelopment perspective offers the condition defined BIF, not yet codified in the ICD, a new consideration by the international scientific community and a large space in the debate on the reconceptualization of the ID. The term BIF refers to a very slight impairment of preschool onset cognitive abilities, characterized by an IQ between 70 and 85 points, with very high prevalence rates, up to 1 in 8 people [177]. The connotation of an

element of continuity between the ID and the normality that is frequently attributed to this condition must be reconsidered in the light of recent research: In fact many more commonalities have been found with people with IQ clearly below 70 than with the general population, for example, the high rates of psychiatric disorders and psychopharmacological treatments, the greater tendency to substance abuse, being more easily victims of exploitation or abuse, the type of communication and social support [177, 178].

On the one hand, in most countries of the world, people with ID still face great difficulties to access mental health services and to receive adequate assessment and treatment, on the other hand the prospect of neurodevelopment is arousing increasing interest among neuroscientists for psychiatry in this sector, both for clinical and research aspects. In recent years, there has been an increase in the production and dissemination of data on epidemiology, symptomatology, therapeutic interventions, and habilitation, even in countries with medium and low income.

Finally, the neurodevelopmental perspective has also favored a multidimensional and polysemic approach to the new classification of ID, which distinguishes between clinical metasyndromes and their declension in terms of dysfunction or disability, with different applications and implications for health and social policies. According to the first WHO ICD-11 working group, the first dimension should find an adequate place in the ICD, with the term "intellectual development disorders," while the second should be included in the International Classification of Operation, of disability and health (ICF) [175], with the term "intellectual disability," already in use for some time in the international scientific community [162]. The proposal of some researchers and clinicians to use the word "disability" also in systems of classification of disorders and diseases could have extremely negative implications for the health needs of people with this condition: The social connotation of the term could in fact offer the opportunities for healthcare providers and organizers to steal resources, already severely limited [6, 162].

The neurodevelopmental approach is currently recognized as the most valid theoretical framework to explain the relationships between genetic and environmental factors that influence brain growth and maturation with subsequent effects on general and specific impairments of functioning and to support multidimensional, transnosographic, interdisciplinary approaches to the study and treatment of neurodevelopmental disorders and psychiatric disorders.

Tip

A systematic approach to differential diagnosis involves a thorough consideration of all candidate diagnoses to yield the most accurate classification, with attention to be primarily given to the age of presentation of symptoms. As a second general step, the most prominent symptom should guide the clinician in the process of considering the possible differential diagnoses. Finally, an accurate evaluation of the general functioning profile of the individual has always to be taken into account.

Key Points

- Symptoms of ASD and/or ID overlap significantly with a wide range of psychiatric conditions and in particular the recognition of ASD across the lifespan represents a clinical challenge. The diagnosis of ASD in clinical practice is often made difficult by the preponderance of differential diagnoses that should be considered, such as neurodevelopmental, anxiety, mood, psychotic, and personality disorders.
- Given the numerous overlapping symptoms among ID, ASD, and psychiatric conditions, it has to be considered that the DSM-5 has introduced an integration of a dimensional approach to diagnosis and classification together with the DSM-IV-TR categorical approach. Indeed, previous editions of the DSM used a categorical model requiring a cli-

nician to determine that a disorder was present or absent. The dimensional approach allows a clinician more possibilities to assess the severity of a condition and does not imply a real threshold from low-functioning forms to forms with very high general skills.

- Advances in the development of dimensional models and approaches to research on psychopathology made possible to find that behavioral, cognitive, social, and emotional competencies share common neurobiological bases, evolve across the brain development, and vary across individuals in different traits, abilities, or mental disorders. In this context, the neurodevelopmental perspective is gaining a central role in theorizing about the etiology of disorders. Indeed, distinctive types of psychopathology may have early onset, features in common, and different time courses in symptoms depending on the variations of the same underlying disposition.

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Intellectual Disability and Sensory Impairment

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Contents

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Learning Objectives

- To become aware of the high prevalence of hearing, visual and dual sensory impairments in the population of people with intellectual disabilities (ID).
- To learn about adequate assessment methods.
- To learn to differentiate key phenomena of autism in people with ID and sensory impairments.
- To learn to consider communication difficulties as possible reasons for challenging behaviour in people with ID and sensory impairments and to differentiate from mental health disorders, especially in deaf individuals who use sign language.
- To learn how to improve environmental conditions with respect to the specific needs of people with hearing, visual and dual sensory impairments and ID.
- To learn how to avoid unnecessary stress for people with sensory impairments by using proper announcement and information strategies.
- To learn to acquire a responsive communication style and knowledge about different communication modes like sign language and tactile communication.
- To learn how to improve communication abilities in individuals with ID and sensory impairments and in all people who are part of their social network like family members, peers and professionals.

33.1 Introduction

Impairments of sensory functions in people with intellectual and multiple disabilities stay frequently unrecognised. Abnormalities and delays in information processing and behaviour are frequently attributed to cognitive dysfunctions only. This 'overshadowing' leaves sensory impairments undiagnosed.

This chapter aims to give an overview on prevalence rates of the combination of sensory impairments and intellectual disabilities. Assessment methods and therapeutic consequences, especially with respect to improving communication, will be highlighted. Challenging behaviour and autism spectrum disorders occurring in people with sensory impairment and intellectual disabilities will be discussed. Practical aspects regarding the design of environments in ways that accommodate the needs of people with sensory impairments and intellectual disabilities will be pointed out. Furthermore, the chapter will close with practical aspects focusing on the improvement of communication.

33.2 Definition and Epidemiology

Various studies have investigated the prevalence rates of sensory impairments and intellectual disabilities in the past. The rates vary depending on the methodology (assessment or questionnaire) and the definition used for sensory impairments. Studies collecting data from questionnaires might be influenced by the subjective information given by staff or family members and, therefore, may not be completely accurate [1].

33.2.1 Visual Impairment and Intellectual Disability

Definition of Visual Impairment

The WHO defines visual impairment as a visual acuity of less than 0.30 but more than 0.05 and/or visual fields less than 30° around a central fixation point. Blindness is defined as a visual acuity of less than 0.05 and/or visual fields of less than 10° [2].

Prevalence of Visual Impairments

Globally, 0.48% of the population is blind, 2.95% are moderately to severely visually impaired, and 2.57% have a mild visual impairment [3].

Prevalence rates of visual impairments in people with intellectual disabilities are varying according to different methodologies used in studies. A study conducted by Warburg in 1994 showed that there was only consensus in 32% of the cases when estimates of caregivers and clinical assessments were compared [4].

A study in Copenhagen, Denmark, looked at a representative group of 778 people, attending therapeutic working programs and day structures. It showed that after the assessment of the visual function, 20% of the participants were attested with a visual impairment. After they received adequate visual aids, 10% of them were still permanently visually impaired. The most common causes identified were excessive myopia, cortical visual impairment, optic atrophy, cataract and keratoconus. The study concludes that the prevalence rate for visual impairment in adults with intellectual disabilities is 10 times higher than would be expected when compared to a sample of the general population [4, 5].

In the Netherlands, a large-scale epidemiological study including 1598 adults with intellectual disabilities was conducted in order to gain more information on the prevalence of visual and hearing impairments within this population. Even though the results found in this study cannot be extrapolated to the entire adult population with intellectual disabilities in the Netherlands, it can still be assumed that a large number of participants portray a reliable reflection of this population. The results show that visual impairments were present in 13.5% and blindness in 5.7% of the study population. Visual impairment had been newly diagnosed in 42.6% and blindness was not diagnosed in 35.9% prior to this study. In total, in 40.6% of the cases, the visual impairment had not been diagnosed before. These results are in line with the numbers other studies have found in the same group of population and are partly traced back to the overshadowing effect [6].

A study conducted in Austria had the aim to investigate the proportion of previously undiagnosed deafblindness in a large sample (224 participants with available data) of persons with intellectual disabilities living in congregate settings. All participants were enrolled at a facility called 'Hartheim', which offers high standard care for people with profound, severe and moderate intellectual disabilities. All people living in this facility are seen by a general practitioner prior to admission, and medical check-ups are done regularly. For this study, all participants had their vision re-assessed. The assessment identified 21.4% of the total sample with a previously undiagnosed visual impairment (see Table 33.1). It was apparent that ophthalmic pathology was more often diagnosed compared to hearing impairment. This could also be observed in the Dutch sample [7].

The Metropolitan Atlanta Developmental Disabilities Surveillance Program (MADDSP) was established in 1991 and the main goal is to estimate the prevalence of children in the metropolitan Atlanta area with one or more

Table 33.1 Visual impairments previously and newly detected by severity of impairment in 224 residents with intellectual disability [7]

	Before assessment *n* (%)	Newly detected *n* (%)	Total after assessment *n* (%)
Visual impairment			
Severe/profound (VA < 0.1)	20 (71.4%)	8 (28.6%)	28 (12.5%)
Moderate (0.1 ≤ VA < 0.3)	18 (31.0%)	40 (69.0%)	58 (25.9%)
Total	38 (17% of 224)	48 (21.4%)	86 (38.4%)

developmental disabilities. The disabilities looked at included cerebral palsy, hearing loss, intellectual disability and vision impairment. In 1996, autism spectrum disorder was added to the list. The method that is used is called 'active record review' and means that, every other year, trained staff get information from various health and educational sources. From the years 1991 to 1994, children between the ages of 3 and 10 were included. Starting in the year 2000, only 8-year-old children were included. By that age, it is assumed that most children with developmental disabilities have been identified for services [8]. This large study found that intellectual disability was the most common co-occurring developmental disability in people with visual impairment, present in 55% of visually impaired people. The prevalence rate was stable between 1991 and 2010 [9].

Van den Broek reported in line with other studies higher prevalence of visual impairments in people with more severe degrees of intellectual disabilities [10].

Evenhuis et al. investigated whether visual impairment causes impact on adaptive skills in people with intellectual disabilities. Research shows that, in the general population, visual impairment causes problems with daily activities. This study found that with an increase in intellectual disabilities and an increase in visual impairment, adaptive behaviour decreases. The study had five hypotheses. Firstly, they hypothesised that independent of the severity of intellectual disability, lower vision was associated with a decrease in independent living skills. Further, they assumed that lower vision increased the difficulties in communication. Thirdly, they associated lower vision with more insecure movements. Additionally, visual impairment was associated with more self-absorbed and withdrawn behaviour. Finally, it was assumed that lower vision was associated with less antisocial and disruptive behaviour. This study finds that, generally, the effects caused by intellectual disability are larger than the effects of the visual impairment. Independent living skills, communication and social skills are mainly affected by the intellectual disability, but as expected, visual impairment does add to this. The study found that, contrary to their hypothesis, visual impairment is not related to a more self-absorbed behaviour. In the subgroup of people with severe intellectual disability, lower vision was associated with less anxiety. The last hypothesis was also not confirmed, disruptive and antisocial behaviour did not decrease with lower vision [11].

Dijkhuizen et al. investigated the impact of visual impairment on the ability to perform daily living activities in a sample of 240 people with intellectual disabilities. The Barthel index was used to measure the ability to perform daily activities and the locomotor functioning was operationalised through measuring the comfortable walking speed. The outcome of the study is in line with the study by Evenhuis et al. and shows that, in people with intellectual disabilities, visual impairment only slightly affects the ability to perform daily activities [12].

Assessment Methods and Refractory Correction

People with intellectual disabilities may have difficulties communicating a worsening of their sensory abilities due to language deficits. Therefore, sensory impairments often become evident when behaviour changes, which in many cases is misinterpreted as the person being non-cooperative [13].

Generally, in this population it is recommended that visual functions are regularly evaluated by the use of more than one assessment method. This increases the reliability of visual acuity measurement [6, 14].

The assessment of visual function often proves to be more difficult in people with intellectual disabilities due to problems in communication and understanding. The assessments rely on subjective responses, which might be difficult to elicit. For the assessment of the visual acuity in people with mild-to-moderate intellectual disabilities, a high-contrast logMAR letter or symbol acuity charts are suitable [15]. For people with more severe intellectual disabilities, the Teller Acuity Cards [16], which are grating cards with contrasting stripes, can be used. This test makes use of a

forced choice procedure based on the tendency of people to preferentially look for contrast. The assessment includes the observation of the patient's direction of gaze. The stripes become progressively narrower, with the size of the smallest stripes seen determining the visual angle and, thus, the visual acuity [7]. The Pacific Acuity Test, originally a test to measure recognition of visual acuities in preverbal children, has also proven to be suitable to use for people with intellectual disabilities [17]. If people are not familiar with letters or shapes, objects that are known from daily life can be used [15].

Restrictions of the visual field might explain certain behaviours or the failure to do certain activities. Usually, it is difficult to do an automated static visual field assessment in people with intellectual disabilities; therefore, modified confrontation testing, such as the Stycar graded balls test, is well suited [18]. Additionally, attention should be put on eye movement and head turns towards peripheral targets [15].

If possible, subjective refraction measurement can be performed, if the intellectual level of the person does not allow this, then retinoscopy should be done to measure the refractive error of the person's eye. In the last part of the assessment, the doctor looks at the ocular health. This can be particularly challenging in people with intellectual disabilities. Due to difficulties in sitting still, it is important to use portable equipment [15].

Findings regarding the treatment were as alarming as the high prevalence rate of undiagnosed visual impairments. In many cases, individuals did not wear glasses. It has been observed that people with intellectual disabilities receive less eye care compared to the general population [13]. Previous studies have also found that the prescription rate for glasses is much lower in people with intellectual disability [19]. It is difficult to predict how a person with intellectual disability will respond to wearing glasses and how big the benefit of it would be, nevertheless, doctors should not decide about the appropriateness basing only on the presenting behaviour, but ideally trial the effectiveness of the glasses and then observe the behaviour [15]. There are various reasons found in the literature to explain this, one of them highlights the misconception that people with intellectual disabilities have less visual demands. It is often argued that people with profound intellectual disabilities do not participate in activities that would require good vision and, therefore, it is said that glasses would be of little use. It is important though to look at this phenomenon the other way around. Could it be possible that the visual impairment is the reason for disengagement? Another explanation could be that physicians are sometimes facing challenges such as limited specialised training, difficulties in communication, inadequate information prior to the assessment and eventual presence of challenging behaviour in persons with intellectual disabilities. The Functional Vision Assessment (FVA) for people with learning disabilities [20] can help to evaluate different aspects of visually related symptoms and behaviour. There are checklists for seven domains: appearance of the eyes, behaviour, central vision, peripheral vision, sensitivity to light, colour vision and contrast sensitivity and poor vision in one eye. These checklists can support the history taking with the patient and, furthermore, can sensitise the carers to look out for visual and ocular health [15].

- About 35% of individuals with intellectual disability have visual impairments.
- Severity of intellectual disability correlates with severity of visual impairment.
- Half of the co-existing visual impairments stay undetected without proper assessments, which should lead to correction of refractive errors.
- Assessment methods that make use of the tendency to preferentially look for contrast are beneficial

33.2.2 Hearing Impairment and Intellectual Disability

Definition of Hearing Impairment

Hearing loss can be categorised into four different groups. Mild hearing loss is present from 20 to 30 dB, and moderate hearing loss is

up to 69 dB. Within this definition, severe hearing loss is up to 94 dB and profound hearing loss is higher than 95 dB [13]. People suffering from mild to severe hearing loss are also referred to as hard of hearing. The World Health Organization uses the definition of disabling hearing loss if the hearing loss is greater than 40 dB in the better ear. Hearing loss can be either due to congenital causes or acquired causes. Congenital means that the hearing loss is already present or acquired soon after birth. Globally, the most prevalent reasons for hearing loss are maternal rubella, low birthweight and birth asphyxia. Acquired hearing loss can occur at any age and the most prevalent factors include infectious diseases (meningitis, measles and mumps), chronic ear infections, excessive noise, and ageing. Among children, chronic otitis media is a common cause of hearing loss [21].

Prevalence of Hearing Impairment

According to the Metropolitan Atlanta registry between the years 1991 and 2010, the prevalence rate of moderate-to-profound hearing loss was 1.4 per 1000 children with an average change of 0.93% per year. Forty-seven per cent of the children have moderate hearing loss, 23% have severe hearing loss and 31% have a profound hearing loss. The prevalence rate was stable throughout the years across all severity levels. The most common co-occurring developmental disability was intellectual disability, present in 23% of the children. Looking at it the other way around, 3% of all 8-year-old children with an intellectual disability have a co-occurring hearing loss. A significant increase could be found for the years 1991 and 2010. The increase ranged from 0.02 to 0.03 per 1000 children, with an average yearly increase of 2.4% [9].

The prevalence rate of hearing impairments in adults with intellectual disabilities varies depending on the methodology of the studies. A Dutch study which investigated visual impairments in 1598 adults with intellectual disabilities also investigated the prevalence of hearing impairments in this population. The results show that 35.8% of the study population have a hearing impairment, and 47.6% out of them did not have a diagnosis prior to the study. There was no significant relationship between the degree of intellectual disability and the hearing impairment. The re-weighted prevalence of hearing impairment to the total population with intellectual disability was 30.3%, which is almost twice as high as the prevalence in the general population (16–17%). The important message of this study is that almost half of the hearing impairments have not been diagnosed before structured assessment [22].

The previously mentioned Austrian study conducted in Hartheim ($N = 224$) found that 46% of the individuals with intellectual disability have a hearing impairment. Prior to the study 72.8% (75 of 103) did not have a diagnosis of hearing impairment (see Table 33.2) [7]. This high number of undiagnosed hearing impairment, both in this and in the study from the Netherlands, shows the importance of proper assessment in populations with intellectual disabilities.

A study by Timehin et al. found a prevalence rate of 9.2% of hearing impairment in people with learning disabilities. Only 12 people out of 50 have ongoing assessments and hearing aid maintenance. Further, only three-quarters of the participants have their hearing checked at some point in their life [23].

Assessment Methods and Hearing Aids

Checking the otoacoustic emissions proves to be an ideal screening method for people with intellectual disability, after cleaning the external auditory canal from cerumen (in many cases necessary) [24, 25]. Depending on compliance, behavioural audiometry, puretone audiometry and speech audiometry can be used. If the target population does not show compliance, the last step in the diagnostic process is brain stem audiometry, which requires general anaesthesia.

Once the diagnosis of hearing impairment is made, hearing aids should be fitted. However, in people with intellectual disabilities, the use of hearing aids is often not tried, or fails. This is because it is assumed that they

Table 33.2 Hearing impairments previously and newly detected by severity of impairment in 224 residents with intellectual disability [7]

	Before assessment *n* (%)	Newly detected *n* (%)	Total after assessment *n* (%)
Hearing impairment			
Profound: hearing loss ≥90 dB	10 (90.9%)	1 (9.1%)	11 (4.9%)
Severe: 70 dB ≤ hearing loss <90 dB	6 (46.1%)	7 (53.9%)	13 (5.8%)
Moderate: 40 dB ≤ hearing loss <70 dB	10 (16.9%)	49 (83.1%)	59 (26.3%)
Mild:25 dB ≤ hearing loss <40 dB	2 (10%)	18 (90%)	20 (8.9%)
Total	28 (12.5% od 224)	75 (33.5% of 224)	103 (46% of 224)

would not easily accept hearing aids [26]. A constant effort and careful process of adaption are needed to encourage people with intellectual disability to use hearing aids [27]. In a study with 16 people with mild-to-moderate intellectual disability, the acceptance of hearing aids was tested. The participants were interviewed before and 6 months after fitting with hearing aids. The questions asked evolved around the topics of benefit, cosmetics and self-image, sound quality/acoustics, comfort and ease of use and service. Most of the participants were aware of their hearing impairment and understood why they were fitted with hearing aids. Furthermore, similar as in the general population, the perceived benefits of hearing aids played an important role in the acceptance. The authors concluded that the people with mild-to-moderate intellectual disability are able to express their opinion and experience with hearing aids, given that they are asked simple questions [28].

The fitting with hearing aids is only one important aspect; central issues such as communication and environment will be discussed in the last chapter 'practical issues'.

- About 40% of individuals with intellectual disability have hearing impairments, and about 75% do not have diagnosis prior to systematic assessment.
- Severity of intellectual disability does not correlate with severity of hearing impairment.
- Otoacoustic emissions are a proper screening method.
- The use of hearing aids requires constant efforts.

33.2.3 Dual Sensory Impairment and Intellectual Disability

Prevalence of Combined Visual and Hearing Impairment (Deafblindness)

In the year 2004, the European Parliament classified 'deafblindness as a distinct disability, that is a combination of both sight and hearing impairments, which results in difficulties having access to information, communication and mobility' [29].

It is important that this declaration not only uses the word 'deafblind' but that it also includes people with moderate impairments of vision and hearing. This takes into consideration that a dual sensory impairment causes a greater degree of impairment than the sum of each impairment individually, which not only affects the communicative and social development but also the quality of life [30]. A person is considered deafblind when the hearing loss in the better ear is more than 35 dB and the vision

is less than 0.3. Evenhuis suggests a more conservative definition of hearing loss greater than 25 dB in people with intellectual disability [25].

The Dutch cross-sectional study, which also included a high number of people with mild intellectual disabilities, identified 5% with a visual and hearing impairment, of which 2.9% were younger than 50 years of age. A specific risk factor identified was Trisomy 21. The more severe the intellectual disability, the more often deafblindness was present [31].

The Austrian study that focused on the detection of deafblindness in people with intellectual disabilities identified 21.4% of the participants with a combined visual and hearing impairment (see Table 33.3) [7]. This number is close to the prevalence rate of 20% that was found in 2001 in a large Dutch Institution in a group of people with severe and profound intellectual disabilities [32]. More severe forms of deafblindness were more likely to be detected. There was no correlation between the severity of deafblindness and the level of intellectual disability. But more severe intellectual disability was related to a higher prevalence of combined sensory impairment. Failure to identify the various degrees of deafblindness is common among those with intellectual disability and is most common in those with profound intellectual disability. This emphasises the need for awareness training for staff members as well as appropriate assessment methods [7].

In this study [7] as well as in a Danish prevalence study [33], Trisomy 21 was identified as a further risk factor.

- Deafblindness is a distinct disability and is defined as a combination of both sight and hearing impairments.
- About 20% of individuals with intellectual disability have dual sensory impairments.
- More severe intellectual disability is related to higher prevalence of dual sensory impairments.
- Especially individuals with Trisomy 21 are at risk for acquiring dual sensory impairments; regular assessments are needed.

33.3 Intellectual Disability, Sensory Impairment and Autism

Prevalence of autism spectrum disorder in people with intellectual disabilities is much higher compared to the normal population. Similarly, the prevalence rates of autism spectrum disorders in people with sensory impairments are also much higher, but there are no studies that give exact numbers. Existing literature suggests two different explanations. Firstly, a relationship between autism spectrum disorder and sensory impairment and intellectual disabilities is

Table 33.3 Hearing impairments and visual impairments previously and newly detected by severity of impairment in 224 residents with intellectual disability [7]

Category of deafblindness	Known before assessment	Newly detected	All deafblind	% newly detected residents
Profound/severe hearing & visual impairment	4	1	5	20%
Moderate hearing & profound/severe visual impairment	2	14	16	87.5%
Profound/severe hearing & moderate visual impairment	1	9	10	90%
Moderate hearing & visual impairment	1	16	17	94.1%
Total n (%)	8 (3.6% of 224)	40 (17.9% of 224)	48 (21.4% of 224)	83.3%

assumed. The second thesis postulates an overlap of symptoms, which lead to false-positive diagnoses, which subsequently are the reason for the high prevalence rate in this population. The risk that autism spectrum disorder is missed or unjustly diagnosed in people with intellectual disabilities and sensory impairments is often due to the similarities in behaviour. The importance of a correct diagnosis cannot be highlighted enough, when thinking of the right treatment plans that differ greatly whether an autism spectrum disorder is the cause or not. Even though the ICD-11 criteria for autism spectrum disorder include qualitative impairment in social interaction, qualitative impairment in communication and restricted patterns of behaviour, interests and activities, these aspects can also be found in people with intellectual and sensory disability, although they are usually expressed in a different way.

Difficulties in the following areas, reciprocity and peer relationships, verbal and non-verbal social behaviours, joint attention and theory of mind, making conversations, language, imitation and make belief or symbolic play, stereotyped use of objects and self-stimulation, occur in people with autism spectrum disorder as well as in people with intellectual disabilities and sensory impairments. The origins differ depending on the impairment. It is important to focus on the small differences in the way the symptoms are expressed in order to make the right diagnosis and decide for the correct treatment. The lack of suitable instruments to diagnose autism spectrum disorder in people with multiple impairments adds to the difficulties to differentiate between all the overlapping symptoms [34].

Deafblindness and Intellectual Disabilities and Autism

Research shows a positive correlation among dual sensory impairments, intellectual disabilities and autism [1, 35, 36]. It is, therefore, assumed that deafblind people have a higher risk for autism spectrum disorder. The same risk of overdiagnosis that appears to be present in people with single sensory impairments also seems to be present in people who have dual sensory impairments [37].

All three of the components of autism mentioned above can also be found in non-autistic people with sensory and intellectual disabilities [34].

People who are deafblind often show similar behaviours with regards to social interaction as people with autism spectrum disorder. In deafblind people, their lack of social interaction is often a result of their dependency on others to start the initiative. For them these situations are often frightening because they miss out on important context-relevant information. Qualitative impairments in communication can also be found in people who are deafblind. They, similar to people who are autistic, have problems with identifying and understanding non-verbal communication. Many people with autism spectrum disorder show a delay or lack of spoken language. In people who are deafblind, spoken language is often absent due to their hearing impairment. Further, they can only communicate to people who are trained in tactile sign language, which restricts the size of their social environment. Both people with autism spectrum disorder as well as deafblind people can show stereotyped and repetitive behaviour. In the latter, the behaviour will decrease with age and with an increase in opportunities to communicate [37].

Differential Diagnosis

The important question that has to be asked is whether the observed behaviour is caused by autism spectrum disorder or by the sensory impairment. For example, when looking at repetitive behaviour, it is assumed that in people with deafblindness it is caused by sensory and social deprivation, whereas in people with autism spectrum disorder it is considered as a way of coping with overstimulation. The different causes of the symptoms highlight the importance of the right diagnosis in order to make the correct treatment plan [37].

Until 2009, it was difficult to differentiate between characteristics in clinical practice because existing tests were not validated or reliable to use with people with deafblindness.

Hoevenaars-van den Boom et al. designed an instrument called 'Observation of characteristics of autism in persons with deafblindness' (O-ADB), which is a semi-standardised observation instrument adapted to identify the competencies and skills of deafblind people. The instrument scores the quality and the frequency of the target behaviours [37].

The instrument was used in a study with 10 deafblind people with intellectual disabilities and with and without autism spectrum disorder. The results showed that the instrument was suitable for the differentiation between deafblind people with and without autism, especially in the domains 'openness for contact', reciprocity in contact including 'joint attention' and 'communicative functions'. The participants with autism were rated significantly lower than the participants without autism. No differences were found in exploration, play and problem-solving behaviour. The same applies for stereotyped behaviour. These categories are not well suited to differentiate because baseline levels are too high, meaning that the prevalence is also very high in people with sensory impairments without autism [37].

- In people with intellectual disability, autism spectrum disorder and dual sensory impairments have overlapping symptoms.
- To differentiate between autism and deafblindness, it is necessary to implement proper treatments.
- The domains 'openness for contact', reciprocity in contact including 'joint attention' and 'communicative functions' are most suitable to establish a proper diagnosis.

33.4 Intellectual Disability, Sensory Impairment and Challenging Behaviour

Limited understanding was associated with the highest relative risk for aggressive-destructive behaviour (RR:3.8 [1.7–8.5]) in a study including the total population of people with intellectual disability in Jersey, UK [38].

A meta-analysis by McClintock identified risk markers associated with challenging behaviour in people with developmental disabilities. Problems in both receptive and expressive communication were significantly correlated with self-injury. Difficulties in expressive communication were additionally significantly linked with aggression towards others [39].

These findings indirectly address the problems of individuals with intellectual disabilities and sensory impairments (especially when hearing is affected), who frequently suffer from communication problems.

Challenging Behaviour in People with Disabilities and Hearing Impairments

In their review with the title 'Risk factors leading to behavioural problems in individuals with hearing impairments and intellectual disabilities', Buskermolen et al. complain about the scarcity of studies on challenging behaviour in people with both hearing impairment and intellectual disability and mention that they found only Timehins' publication to give a prevalence rate [40].

Timehin et al. showed in a sample of 543 residents of homes affiliated to the learning disability trust (Lifecare NHS Trust, locality 4) that 9.2% of the sample had a hearing impairment and 42% out of them were profoundly deaf. The prevalence rate of challenging behaviour was 62% within the sample. Thirty-four per cent of the people showed self-injurious behaviour. They also point out that hearing impairments in people with intellectual disabilities are not always identified and that challenging behaviour can be seen as a form of communication for people with limited language abilities. Therefore, they emphasise the importance of paying attention to the link between a possible undiagnosed hearing impairment and challenging behaviour [23].

Buskermolen et al. designed a new assessment instrument called the 'Individual Behaviour Observation and Recording Scale' to assess challenging behaviour in people with intellectual disability and hearing impairment. After they did a review of the existing instruments, they decided that none of them

were adequate for the target population. Taking into account that every behaviour should be considered as some form of communication, it was important for them to create a scale with an emphasis on non-verbal signals. The authors set the following requirements for the observation and recording scale: 'A variety of non-verbal behavioural aspects must be measureable. There must be room for individual behavioural descriptions. The severity of problem behaviour must be measurable. There should be room for several records per day. It should be filled in by professional carers. It must be efficient and easy to use for every carer' [41 p. 33].

The scale consists of five different levels of behaviour (relaxation, slight restlessness, tension, threat and loss of control) and eight different dimensions of behaviour (making sounds, locomotion, facial expression, object manipulation, body manipulation, concentration, expressive communication and receptive communication). A total of 40 individual behavioural descriptions are needed to fill in the scale. Emphasis was put on the recording of individual behaviour [41].

Buskermolen et al. (2016) used the questionnaire in a sample of 21 people with intellectual disabilities and hearing impairments. The cognitive level of the participants was given in developmental age and ranged from 1.3 years to 8.1 years. The hearing loss was measured with the Fletcher Index and the measurements were between 40 and 110 dB. Furthermore, 10 of the participants were diagnosed with autism spectrum disorder. Additionally, the communication abilities and the level of social independence were measured. During the 1-year period of the study, all participants experienced challenging behaviour at some point of time. In the person showing the least challenging behaviour, it was recorded in 1.8% of the time, and the person with the most challenging behaviour showed it in 75.4% of the time. The average was 28.9%. The authors could find a significant negative correlation among challenging behaviour, age, level of social independence and communicative development. This means that people who were socially more independent and who had higher levels of communication showed challenging behaviour on fewer occasions. Further, autism spectrum disorder as a comorbidity had a significant influence on challenging behaviour and was, therefore, identified as an important risk factor [42].

Challenging Behaviour in People with Intellectual Disabilities and Visual Impairments

In a systematic review on challenging behaviour in people with physical and intellectual disabilities, de Winter found a significant correlation between challenging behaviour and visual impairment. Visual impairments were significantly associated with self-injurious behaviour but not with aggressive behaviour. The three studies that looked at sensory impairment and challenging behaviour were of varying quality. The first study by Evenhuis et al. [11] investigating visual impairment and challenging behaviour did not find a significant relationship. The second study by Wieseler et al. [43] found that people with self-injurious behaviour were more often diagnosed with visual and hearing impairment. The last study also investigated self-injurious behaviour and found a high prevalence of visual impairment in eye-poking children. These children often also experience other self-injurious behaviours [44].

In a large Scottish cohort study, Cooper et al. looked at factors that influence the prevalence of self-injurious behaviour in 1023 people with intellectual disability and found a prevalence rate of 4.9% and a significant association with visual impairments [45].

However, these correlations cannot establish causation. Visual impairment might be the reason for the self-harming behaviour, or the other way around that the tissue damage causes the visual impairment [45, 46].

Challenging Behaviour in People with Intellectual Disabilities and Dual Sensory Impairments

Carvill stated in a narrative review that people with intellectual disabilities combined with sensory impairments are often more at risk for developing behavioural problems/challenging behaviour and mental health problems [1]. In 2002, Carvill conducted a case

series with 18 people with intellectual disabilities and sensory impairments in order to investigate behaviour disorders within this population. Most of the participants had dual sensory impairments due to maternal rubella infections during pregnancy and only a few of them had a single sensory impairment. It was evident that the majority of them got referred because of self-injurious behaviour and aggression. In 10 out of the 18 patients, an ICD-10 diagnosis could not be made. This was either due to the unusual behavioural presentation or their degree of intellectual disability. In 15 cases, pervasive developmental disorders were identified. One of the conclusions of the study was that the assessment of psychiatric disorders in people with intellectual disabilities and sensory impairments is problematic, and the difficulties increase with the degree of intellectual disability. It was recommended to gather detailed information from various sources. For behavioural problems in this population, diagnostic overshadowing was highlighted as a reason for long delays in getting proper diagnoses and treatments [47].

Dammeyer showed that within a group of 95 deafblind people, 75% have a psychiatric or behavioural diagnosis [48].

In order to adequately address the needs of people with intellectual disability and sensory impairments who show challenging behaviour, Carvill [49] and Fellinger et al. [7] mention the importance of secured communication as well as experience with people with sensory impairment and intellectual disability in order to identify even small anomalies. Further, the sensitive usage of external sources of information is emphasised.

- Challenging behaviour is frequently associated with communication problems.
- High rates of challenging behaviour in people with intellectual disability and sensory impairments are reported.
- Mental state examination in signing prelingually deaf individuals requires specific experience.

33.5 Intellectual Disability, Sensory Impairment and Psychiatric Disorders

While it is important to consider challenging behaviour as a consequence of communication problems, as well as a number of associated psychological and/or environmental factors, it is also essential to assess their possible nature of symptoms of psychiatric disorder. Especially in prelingually deaf individuals with language deficiency, a mental state examination is difficult, particularly for clinicians with little experience with signing deaf people [50]. Table 33.4 summarises important aspects of a mental state examination in deaf individuals.

Having detected psychopathology in a deaf person with intellectual disability, the treatment plan has to consider the specific communication needs by involving signing experts or sign language interpreters. Support by professional deaf staff is highly recommended also in deaf psychiatric patients with limited sign language skills [50].

When it comes to medication, one has to be specifically cautious with respect to extrapyramidal side effects that might impact vision or hamper communication in sign language ([52] pp. 249–251). In some countries, psychiatric units for deaf people are also accessible for deaf people with intellectual disability including those who need forensic settings [53].

Studies that indicate whether there are higher prevalence rates for specific psychiatric disorders in individuals with intellectual disability and sensory impairments are scarce and hardly population based.

From studies in deaf populations without intellectual disability, one can deduce that increased rates of depression and somatoform disorders can also be expected in deaf individuals with intellectual disability [51]. As far as psychotic disorders are concerned, especially in individuals with congenital rubella, the likelihood to suffer from non-affective psychosis was reported to be five times higher than healthy controls. Brown could also show in his mixed deaf and hearing sample with normal

Table 33.4 Mental state examination of deaf individuals ideally undertaken by signing specialist

Appearance	Deaf people using visual communication modes (sign language and gestures) might give a misleading impression of being agitated. Nevertheless, some seem to be withdrawn or anxious, potentially because of a reaction to the inability to communicate with medical staff and so this can be a result of the situation and not a symptom of a mental health disorder
Affect	In sign language, facial expressions not only represent emotions but also have specific linguistic functions. Some problems such as low drive can be made clear by the clinician imitating the symptoms – for example, looking listless and apathetic. Judgement of whether the patient shows affect appropriate to the topic being discussed could be hindered by poor communication
Thought	Language dysfluency might be wrongly believed to be a result of thought disorder. There is evidence that thought disorder often manifests itself in sign language in a bizarre quality and a meaningless repetition of signs. Signing to oneself might be a symptom of psychosis
Cognition	Many deaf people have reduced access to information. Poor knowledge should never be attributed to low intelligence without proper assessment. In many cases, information from external sources about behavioural and language functions is helpful, but such outside information should not prevent the patient from being able to express himself or herself [51]

Taken from Fellinger et al. 2012 [51]

IQ and congenital rubella that the fact of infection and not deafness itself was the main factor contributing to these high prevalence rates [54].

Besides rubella infections during the first trimenon causing dual sensory impairment, Usher syndrome is one of the most prevalent genetic etiologies, with prevalence rates of 3–5 per 100,000 in the general population. Different subtypes are characterised by retinitis pigmentosa and sensorineural hearing loss and problems in balance. Hallgren reported an association of Usher syndrome and psychosis in 23% [55], whereas more recent studies suggest prevalence rates of about 4%. This is still four times higher compared to the general population [56–58].

In other syndromes with deafblindness like CHARGE syndrome[1] and Wolfram syndrome,[2] a high likelihood of co-occurring mood disorders and/or psychotic symptoms is also reported.

With respect to specific psychopathology in people with intellectual disability, who are blind, no prevalence studies could be found. Visual hallucinatory symptoms due to sensory deprivation are attributed as Charles Bonet syndrome as long as there is no evidence for persistent paranoid ideation.

33.6 Practical Conclusions

As mentioned above people with intellectual disability and sensory impairments are highly vulnerable to stress with communication problems as a specific risk factor. In the following, measures to reduce unnecessary stress and to improve communication are introduced. It is realistic to suspect sensory impairments in a high number of individuals with intellectual disability. Therefore, all of the following recommendations can be of general value.

33.6.1 Structured Time and Environment

- **Announcement Strategies and Structured Schedules**

Lack of information in advance for people with visual and hearing impairments has to be

1 CHARGE syndrome: C – coloboma of the eye, central nercous system anomalies; H – heart defects; A – atresia of the choanae; R – retardation of growth and/or development; G – genital and/or urinary defects; E – ear anomalies and/or deafness.

2 Wolfram syndrome: diabetes insipidus, childhood-onset diabetes mellitus, a gradual loss of vision caused by optic atrophy and deafness.

acknowledged as a relevant source of distress [59]. Therefore, appropriate announcement targeting the functional senses (for deafblind people, e.g. vibration or draft) is crucial to prevent surprise by external stimuli. Picture boards, if necessary with haptic elements (e.g. magnetic boards), depicting the daily routine, can give an accessible structure and contribute to reducing unnecessary stress reactions. People with intellectual disabilities and sensory impairments can use them independently to plan upcoming events or to deal with challenging activities in advance. In congregate settings, the use of picture cards (haptic symbols) of staff who is on duty has proven to be helpful. Just as important as preparing for future activities is dealing with past events by creating, for example, a visual (haptic) personal diary.

An appropriate design of the environment according to the needs of individuals with sensory impairments is a crucial precondition not only for quality of life but also for stress reduction, communication and mobility.

Acoustical Design of the Environment

Background noise reduction makes the target sound more accessible for people with intellectual disability and sensory impairments. Visits to congregate settings for people with multiple disabilities as well as congregate settings for the elderly have shown that often there is a lot of unnecessary background noise polluting the auditory environment. It is important to explain the consequences of background noise on the communicative atmosphere to the staff and find ways to keep radios and TVs switched off. It might be a good idea to mention in the house rules that undirected constant sonication should be refrained from. Room acoustics are particularly important. For reverberant rooms (glass surface, smooth walls and hard floors), the most effective measures (in this order) are the installation of acoustic ceilings with a high insulation value, the usage of soft materials for the floors (e.g. carpets instead of tiles) as well as curtains and wall hangings. The use of felt gliders on chair and table legs particularly in dining rooms can reduce background noise. When designing new living and working environments for people with intellectual disabilities and sensory impairments, the cooperation with experts with knowledge of room acoustics is advisable. Particularly, people with hearing impairment are easily distracted in their visual attention, therefore, it is crucial to avoid 'visual noise' (e.g. constantly switched on TV).

Adaptation of the Environment with Respect to People with Visual and/or Dual Sensory Impairment

Visual and dual sensory impairment is a high-risk factor for falls and accidents; therefore, it is important to design the environment in a way that supports the person to use touch and smell for orientation [13]. Contrasts can be of additional help for orientation (e.g. door frame, edge of a table and edge of a plate) and can, therefore, support people with visual impairment in their independence. A clear division of the rooms into contact and recreation zones can be helpful. It is especially important that nothing unexpected is put somewhere without informing the person about it. Easy-to-access wardrobes, possibly with haptic symbols on the doors, should be installed.

33.6.2 Interpersonal Communication

Even more important than creating an optimised environment is a constant focus on the quality of interpersonal communication.

Interaction Style That Fosters the Development of Communicative Abilities

Communicative deprivation during childhood is an additional difficulty, especially when parents did not communicate enough using an adequate communication mode. This childhood experience has a strong negative effect on people with an intellectual disability and sensory impairment. In order to improve communicative abilities in this population, it is important to develop a high sensitivity towards the communication partner with intellectual disability and sensory impairment by giving her/him enough opportunity to actively participate in the communication. It

is very important to respond to the signals the communication partner with intellectual disability and sensory impairment is sending. Being the dominant part of the conversation can be avoided by giving the person with sensory impairment enough time to process the information and respond.

Interaction with Deaf Individuals

The development of communicative abilities especially in deaf people with intellectua disability has to be a constant priority. The following bullet points (taken from Fellinger et al. [51]) highlight some practical aspects for successful communication with deaf individuals.

- Engage the person warmly and directly with eye contact as often and for as long as possible. Make it clear when focus needs to shift away.
- Ask the person about his or her preferred communicative approach. If it is sign language, collaborate with a signing professional or with a qualified interpreter.
- Be aware of the restricted effectiveness and fatigue of lip reading. Add clear visual elements to discourse – for example, gestures; writing notes and use of simple, key words and grammar, drawings and many visual aids.
- When speaking, ensure that the person has the best possible view of your face. Do not stand in front of a light source (e.g. window or lamp).
- When speaking, use simple language and short sentences. Speak at a natural speed and volume. Give clear, concrete examples, and avoid vague, general terms and jargon.
- Avoid simultaneous comments. Communicate first and then act.
- Accept that good communication with deaf people takes more time.
- Check for comprehension. Ask the person to summarise essential points. Do not ask whether he or she understood because nodding might not mean comprehension [51].

Use of Sign Language

Sign language is the preferred communication mode of many deaf people and signs are particularly important for deaf people with multiple disabilities even if access to this communication mode was not available in early childhood. Models of residential and work facilities, which provide constant visual communication in sign language for deaf people with intellectual disability, offer new opportunities for social learning and relationships. A recent study by the author of the present chapter shows the changes in adaptive skills profiles of eight deaf individuals with intellectual disability who are living in a fully inclusive therapeutic living community. Over a period of 12 years, particularly social awareness and community living improved in the participants [60]. Deaf professional staff members are of high value as role models and guarantee the constant use of sign language within the team [61].

Interaction with Deafblind Individuals

To identify the preferred sense for communication through an interactive assessment, as introduced by Jan van Dijk, is highly recommended for people with dual sensory impairment [62].

Fellinger et al. suggest a classification for dual sensory impairments that ranges from profound impairments in both senses, to those with profound in one but only moderate in the other and to those with only moderate impairment in both senses. Building on this, the provision of educational and care needs can be unique to each group. People in the dual profound group rely on touch and tend to withdraw into their own world of stereotypes and self-stimulation. Individuals with profound visual impairment and moderate hearing impairment rely mainly on touch. They can be alerted by clear auditory stimuli. Help can be provided with tactile cues and orientation. People with profound hearing impairment and moderate visual impairment can participate in social activities, but they are often having severe difficulties in communication. Some may benefit from simplified sign language and hearing aids. Provided that the hearing and/or visual conditions are optimal, people with dual moderate impairments can participate in social activities and are able to communicate.

Use of Tactile Communication

For not only people with dual sensory impairment but also for people with highly limited speech comprehension and a severe intellectual disability, the usage of tactile communication support is recommended. In this group of people, it is more important to identify the preferred mode of communication. This includes tactile cues, for example, the touching of the bottom lip with a spoon before feeding somebody. It is essential to give the person enough time to react after the tactile signal in order to respond with agreement or defence. Additionally, reference objects, known from everyday life, can be useful to announce imminent activities or changes, such as a sponge indicating bathing time. Reference objects can also be used as a tactile calendar to structure longer time periods and to encourage active participation in the daily life. People with prior knowledge of sign language (before deterioration of sight) benefit from tactile signs of a national sign language which means to perform signs hand over hands (Fig. 33.1). It is important to communicate the signs through leading the hands of the other person from underneath. In this way, the person with impairment has enough space to give an independent answer.

Easy-to-understand signs such as swimming movement of the arms and moving the hand to the mouth for eating are usually also understood by people without prior knowledge of sign language.

Fig. 33.1 Hands over hand communication. (Drawing by the author)

- Announcement of imminent activities and an accessible time schedule avoid unnecessary distress.
- A well-structured physical environment supports independence, especially in people with visual impairments.
- The use of sign language or tactile communication based on a responsive interaction style fosters the development of communicative and social skills.

Tip

An intellectual disability can have an overshadowing effect, especially on milder degrees of sensory impairments, which are not detected without appropriate assessment.

Key Points

- Proper assessments are necessary to detect impairments of sensory functions in people with intellectual disability.
- Awareness of the high prevalence rates of the combinations of intellectual disability and sensory impairments can improve the management of challenging behaviour and the creation of adequate environments.
- Everyone in the social environment of people with intellectual disabilities and sensory impairments should develop an attitude that encourages these individuals to actively participate in communication.
- Competences to use appropriate communication systems according to the needs of the respective individual have to be acquired and constantly implemented.
- Continuous development of communication competences of people with intellectual disabilities and sensory impairments helps to improve their autonomy and quality of life.

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Educational and Training Opportunities

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Contents

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Learning Objectives

- Formal and informal training available for medical and other staff who look after the mental health of people with intellectual disabilities in Europe and other countries.
- How advances in the understanding of mental health issues affecting people with intellectual disabilities in the last few decades changed the demand for training?
- How various national and international organisations developed specific projects for training?
- The need for a European or an international consortium to develop a worldwide training programme to standardise training in this area.

Health provision systems are typically governed and regulated by public authorities and governments and include measures for primary and tertiary health prevention as well as defining and promulgating a consistent health promotion policy. Further, governments frame and regulate the quality of the provision of services in the health system by defining, among others, standards in the basic education, training and continuous education for health professionals and paramedical professions, often in close collaboration with the respective professional guilds (e.g. national medical, psychological or psychiatric associations). With the massive rise in health-based knowledge as well as technical advances in this area, the number of specialists in the health field is steadily growing, acknowledging the scientific advances in the respective fields. The establishment of health specialists and health consultants, regulated generally by standardised and quality-controlled training curricula, contributes to quality assurance of services in the health sector. In the area of mental health, the early professionalism depicted best by the general psychiatrist, formerly taking care of people of all ages and all kind of disorders, often trained on a dual track in neurology and psychiatry, as so-called neuropsychiatrists. Many countries have established mental health professionals as specialists for children and adolescence and for adults and some for older people or for people with intellectual disabilities (ID) (i.e. learning disabilities).

Tip

In most countries in the world, there is a lack of formal structure for training doctors and other professionals who look after the mental health of people with intellectual disabilities.

34.1 Introduction

Awareness of the relevance of education and training professionals in mental health issues in people with intellectual disabilities (ID) has only be growing in the final decades of the twentieth century. Since 1980s the evidence base on mental health issues in ID is growing with a better understanding of its bio-psycho-social aetiology. Until recently mental illnesses and problem behaviours (PBs) were perceived as part of the ID phenotype caused by the underlying brain impairment [1]. Subsequently, as the underlying brain impairment of ID was seen as incurable, so was the co-morbid mental illness and PB. Similarly, psychosocial interventions and various forms of psychotherapies, including cognitive behaviour therapy (CBT), were seen as inapplicable to people with ID. Diagnostic nihilism was followed by therapeutic nihilism, as manifested in the failure to provide a systematic assessment of mental health problems in people with ID and led to the neglect in training for mental health professionals working in the field of ID.

An early textbook on mental health in ID edited in the USA by Menolascino & Stark [2] addressed this issue in a chapter on training mental health professionals. The authors reported that the upcoming de-institutionalisation of people with ID from large-scale psychiatric institutions, as early as the late 1960s, to community-based housing

facilities, had spurred the interest in providing both general and specialised training experiences in ID for mental health professionals [3]. Mental health of many of these people improved once resettled in small community-based accommodations, with more personalised support and more options for self-determination. In the USA, the new disability rights law leading to deinstitutionalisation of people with ID contributed to a better understanding of the bio-psycho-social model of mental illness and PB in this population.

The shift from institutionalised ID services to mainstream community-based provisions for people with ID called for a change in training structure for support staff. The professional skills and competencies of staff should support the concepts of social inclusion and empowerment of people with ID. Rose and colleagues' [4] systematic review of staff training in mental health in people with ID showed that effective training can improve skills, knowledge and attitudes of support staff. The authors discussed the importance of incorporating this specialised training within the generic training framework of support staff training within community services for people with ID.

With the emerging model of self-determination and inclusion, new training options in ID mental health arose. Most of the new training frameworks were implemented in Anglo-Saxon countries initially, followed a decade later by western European countries. The drivers in these training advances were the need for mental health support, as now experienced in community-based settings for people with ID [5]. The need to develop specialised mental health services exposed the need for a fundamental re-evaluation of support staff knowledge and awareness of current models and concepts in supporting people with ID. The idea was to introduce these new training frameworks founded on principles of social inclusion and person-centred care for people with ID both for staff working within the ID as well as mental health services. Educational programmes should always be based on strong evidence for their effectiveness. New training programmes should value the advances in the knowledge of assessment of mental health issues and PBs in people with ID using biological as well as psychological, cognitive behavioural and psychotherapeutic approaches [6].

Finally, two main reasons underpin the relevance of high-quality education and training need in ID mental health:

(a) High rate of mental ill health in people with ID, including PB and co-morbid additional neurodevelopmental disorders (NDDs), such as autism spectrum disorder (ASD) and attention deficit hyperactivity disorder (ADHD) [6–9].
(b) High risk of social exclusion, community placement breakdown and use of restrictive practice including inappropriate use of medication when mental illness and PB in people with ID do not receive appropriate intervention and support [6, 7, 10, 11].

In the following sections, we described how education and training is structured and organised in the field of mental health and ID, with particular reference to European countries, where the main authors of this chapter carried out most of their training activities. Examples include formal professional educational programmes providing continuing education, leading to postgraduate qualifications and online training. We have described the different organisations that are providing good-quality training for staff working with people with ID who have mental illness and PB and differences in the structure of training.

34.2 Challenges for Education and Training in ID Mental Health

Education and training frameworks in mental health and ID show a wide variation among the European countries. The *Declaration of Rome, 2003,* developed and promoted by the European Association for Mental Health and Intellectual Disability (EAMHID) [12], addresses many areas of inequality and discrimination with respect to the mental health of people with ID, with education and train-

ing of health professionals in the area of mental health and ID being one of these. The declaration calls among others for:

- *Policy makers and public administration officers to review and analyse the requirements for adequate mental health-care provision and to stimulate the development of special supports for those with ID and additional mental health problems*
- *Professionals in the care provision system to intensify their professional engagement, with special efforts in interdisciplinary collaboration, and mental health education and training*
- *Researchers to increase the scientific efforts to promote healthy mental development*
- *Finally, recognises that relatives and friends of people with ID claim intensified public awareness of this topic*

Systematic reviews on training of staff in ID and mental health are rare [13, 14]. This highlights the lack of recognition of the relevance and the need for formal training in mental health and ID nationally and internationally. Costello and colleagues [15] identified the factors promoting and hampering delivery of appropriate training for psychiatrists, psychologists and primary care physicians working in the ID field in five countries (the UK, the USA, Canada, Australia and Austria). Of these countries, only the UK has a formal nationally accredited training structure for psychiatrists working with people with ID.

A cross-sectional study, conducted by Dias and colleagues [16] in 2015, on 42 European countries, investigated the availability of ID training and found that only 23 (56%) countries provide postgraduate psychiatric training on ID. Training shows a wide variety in terms of duration and format (lessons, clinical rotations and case discussions) and a frequent exclusive reference to child and adolescent psychiatry (see Table 34.1). Only in Ireland and the UK, ID is recognised as a subspecialty of regular psychiatry training.

There seems to be a substantial correlation between a good-quality service provision and a high-quality training and education for the professionals working in the field of mental health and ID. Any service staff should be adequately trained and regularly updated, with further resources given to those with particular needs [17]. Rose and Gallivan [13] proposed a framework for staff training which identifies areas related to support and prevention. It includes training topics and processes that can be introduced proactively such as supervision and risk assessment but also reactive strategies (i.e. strategies contingent upon maladaptive behaviour, such as appropriate physical skills training, and responsive elements, such as incident analysis and behavioural skills training). A modified version of this framework is provided in Table 34.2.

Supervision (biweekly or monthly) is a methodology for maintaining or reinforcing the training intervention.

Formal standardisation of training and establishment of higher academic posts in psychiatry or psychology at professorial level are missing in most European countries apart from two exceptions, namely, Republic of Ireland and the UK. Similarly, apart from a few exceptions, most countries do not have specialist posts in ID psychiatry or psychology.

Recent advances in training in the ID field are often due to the effort of committed institutions, service providers, private and professional organisations [18]. Furthermore, major players are very often key individuals, collaborating in networks, and pulling together consortiums establishing European Union funded grants with projects producing further education material in this field. In many parts of Europe, mental health professionals working with people with ID and mental health problems have been self-taught, soon realising that they missed out during their formal professional training on gathering substantial knowledge and competencies necessary to provide appropriate support to people with ID who have mental health needs.

In the next sections, we described some good examples of formal nationally accredited mandatory training in the field of ID and mental health.

Table 34.1 Training on intellectual disability within psychiatry training across European countries

Country	Intellectual disability psychiatry as part of psychiatric training	Teaching format
Albania	No	–
Austria	No	–
Azerbaijan	Yes	Lectures
Belarus	Yes	Lectures, clinical rotation and case review
Belgium	Yes	Mandatory lectures and optional clinical rotation
Bosnia Herzegovina	NR	NR
Bulgaria	Yes	Part of CAP clinical rotation
Croatia	No	–
Cyprus	No	–
Czech Republic	Yes	–
Denmark	Yes	Very few lectures
Estonia	Yes	Lecture and CAP training
Finland	Yes	In CAP, it is possible to have clinical rotation in child neurology
France	No	–
Georgia	No	–
Germany	Yes	Lectures and optional rotation
Greece	Yes	Lectures
Hungary	No	–
Ireland	Yes	Learning outcomes for psychiatry of ID must be attained. Three years clinical training in HST is required for certification in ID (or 2 years in dual certification)
Italy	No	–
Israel	Yes	Lectures and case reviews
Latvia	Yes	NR
Lithuania	No	–
Luxembourg	Na	–
North Macedonia	Yes	Lectures

(continued)

Table 34.1 (continued)

Country	Intellectual disability psychiatry as part of psychiatric training	Teaching format
Malta	Yes	Lectures and clinical rotation
Montenegro	Yes	Lectures
Norway	No	–
Poland	No	–
Portugal	No	–
Romania	No	–
Russia	Yes	Lectures, examinations and rotations
Serbia	Yes	Lectures and clinical practice
Slovakia	No	–
Slovenia	No	–
Spain	Yes	–
Sweden	Yes	Lectures and clinical rotation
Switzerland	No	–
The Netherlands	No	–
Turkey	Yes	Lectures, outpatient practice and forensic psychiatry
Ukraine	Yes	Lecture
The UK	Yes	This is taught as part of the compulsory MRCPsych exam preparation course and is tested in the exam Some trainees may complete an ID placement during core training There is also the possibility of completing the 3-year higher specialty training programme in ID

Time of data collection is 2014–2015
Abbreviations: NA not applicable, *NR* no response, *HST* Higher Specialist Training, *MRCPsych* Membership of the Royal College of Psychiatrists, *CAP* Child and Adolescent Psychiatry
Source of information: [14, 16, 35, 73]

34.3 Mandatory Formal Education and Training in Psychiatry and Clinical Psychology

From a European perspective, the cases of Austria and the UK might be examples of ways how an appropriate framework in the field maybe achieved for training psychiatrists and clinical psychologists.

34.3.1 Psychiatrists

In the UK, after leaving the Medical School, doctors spend 2 years in foundation training in different medical specialities, which may include placement in a psychiatric or ID service for 4 months. After that they can choose a medical speciality for further training including psychiatry. The core train-

Table 34.2 Staff training framework

Universal training	Specific training	Responsive training
Person-centred values and attitudes Increasing knowledge Training on the communicative aspects of challenging behaviour Training on attributions of challenging behaviour Training on emotional intelligence Training on risk assessment/risk management culture	Appropriate physical skills training	Nidotherapy Behavioural skills training Incident analysis Formulation

Adapted from Rose and Gallivan [13]

ing in psychiatry is for 3 years which may include a 6-month placement in psychiatry of ID. Within the core training, they will have to complete a Member of the Royal College of Psychiatrists (MRCPsych) UK examination. After that they enter a higher training phase for 3–4 years. For higher training, they can choose from six psychiatry subspecialties, one of which is in psychiatry of ID. During the whole training period, the doctors are in a salaried job seeing patients in various settings including inpatient admission units, outpatients, community homes, etc. For psychiatric training, the doctors work within a multidisciplinary team comprising nurses, clinical psychologists, speech and language therapist, social workers, etc.

During the psychiatric subspecialty, higher training doctors can spend time doing special interest session in neurology, genetics, forensic psychiatry, child psychiatry, neuropsychiatry, etc. Some dual training opportunities exist not only involving psychiatry of ID and forensic psychiatry but also psychiatry of ID and child and adolescent psychiatry. At the end of the subspecialty, higher training psychiatrists receive a certificate of completion of training (CCT), which allows them to apply for a consultant psychiatrist in ID job in the UK National Health Service (NHS) or private sector. In some cases, they can apply for a child ID psychiatrist job or a forensic ID psychiatry job. All trainings are based on a structured curriculum developed by the Royal College of Psychiatrists and accredited by the UK General Medical Council (GMC) that oversees all medical training including training in psychiatry in the UK. The UK universities also have professorial posts available in ID both for psychiatrists and psychologists.

In Austria, training of psychiatrists is regulated by the legal framework of 'Physicians Training System' [19]. Training lasts for 6 years, of which 4 years are spent in psychiatry. They spend one year each in neurology and internal medicine. After completing a final examination, they can apply for a professional title 'consultant in psychiatry and medical psychotherapy'. On-the-job training is provided within the hospital settings, which is a major obstacle in the way of gaining competency to provide community-based psychiatric-psychosocial services. The curriculum for this postgraduate training in psychiatry sometimes involves disability and, specifically, ID. However, subspecialty training in ID or disability is not mandatory. Training in psychiatry has no structure with regard to specific target groups, with the exception of the 'consultant in psychiatry of childhood and youth and medical psychotherapy' defined as a speciality of its own. Austrian universities do not provide professorships in psychiatry and ID. However, medical universities have established professorships in psychiatry of childhood and youth, covering in addition ASD and ID.

34.3.2 Psychologists

In the UK, the Doctorate in Clinical Psychology (ClinPsychD) is a 3-year-long postgraduate course accredited by the British

Psychological Society (BPS) and Health and Care Professions Council (HCPC). Those who hold this qualification can apply for a Consultant Clinical Psychologist post either within the UK NHS or in the private sector. All across the UK, 34 such courses are on offer. King's College in London hosts the UK's oldest programme which is underpinned by a bio-psycho-social model with an emphasis on integration of theory, research and practice in all aspects of training. The programme follows an evidence-based approach and has cognitive-behavioural therapy as its main therapeutic modality [20].

In each year, trainees spend 3 days per week on supervised clinical work and 2 days are dedicated to teaching and research. Trainees undertake six, 4-to-6-month clinical placements. The first year covers four core areas including adult and child mental health. Second year covers modules on older adults and ID, and the third year comprises two electives or specialist placements, with ID being one of the options. The academic curriculum includes clinical psychology as applied to ID as part of 1 of the 12 thematic areas. Other mandatory modules consist of adult mental health, forensic psychology, research methods, race-equality-diversity and leadership.

The curriculum is delivered through a combination of lectures, seminars, workshops, reflective practice meetings and small group tutorials, offering the trainees opportunities to reflect critically on theoretical issues and their application to clinical practice and research. Trainees are allocated clinical supervisors while on clinical placements and a clinical tutor who supervises overall training. Trainees also choose their own supervisors for the literature review and research projects. The available doctoral training posts are full-time positions paid by the UK NHS.

In Austria, post-graduate training is on two tracks, namely clinical psychology and health psychology, which is regulated since 2013 by new Psychologists' law [21], replacing the one of 1991, which consisted of theoretical and practical components. The theoretical part has a common basic trunk for both tracks followed by separate advanced training in either clinical or health psychology. The common trunk has a duration of a minimum of 340 units (one unit = 45 min), with 220 units minimum for the basic and 120 units minimum for the advanced branches. The basic trunk covers taught courses in groups of 15 (maximum) on professional legal and ethical topics, communication, counselling methods and supervision, assessment and diagnostic strategies, service provision system, psychological concepts of health prevention and health promotion as well as crisis intervention. The advanced branches, with a minimum of 120 units, are designed to train transfer of taught knowledge to practical clinical use in order to strengthen professional skills. In this section, trainees have to produce a number of individual case studies. The internship has a total of 1638 h for the health track and 2188 h for the clinical track. The trainee has to take at least 500 h of his or her internship timely in parallel to the theoretical component of the training programme, thus assuring the reflective supervision units (100 units health track and 120 units clinical track) to be offered by the programme certified for postgraduate training.

The internships for both professional psychology degrees follow separate pathways with practical training programmes linked to the health promotion sector or to the clinical prevention sector. The duration is spent in full-time employment for 1.5 years for the health track and for 2 years for the clinical track. The legal framework does not specify any elements mandatory in the theoretical and practical components from the ID field. The theoretical training programmes on offer have to be approved by the National Ministry of Health, and are set up either by professional psychology organisations, such as the Austrian Association of Psychologists, or by postgraduate programmes of public universities. A number of service providers from the ID field are accredited to provide internship placements for the two tracks. Upon successful completion of theoretical and practical components of the programme, the trainees take a final committee-based oral examination, and if successful are awarded the professional title of either Health Psychologist or Psychologist. The next step is the registration in the national

list of clinical psychologists and health psychologists at the National Ministry of Health. The trainee has to cover for the costs of the theoretical part of the training and is supposed to have a salary-based contract for the period of the internship.

34.4 Non-Mandatory Formal Training Options in ID Mental Health

Non-mandatory formal training programmes are currently the main options in a number of European countries to improve the competency and skills of professionals and front-line staff working in the ID field. A comprehensive training in mental health and ID is not only lacking for the psychiatrists and clinical psychologists but also for other professionals such as psychotherapists, social workers, educators and even disability caregivers or personal assistants. Therefore, some academics, researchers and managers in care provider organisations tend to provide training in ID and mental health. Such activities can be grouped under three major categories: (a) University-based postgraduate programmes; (b) Organisations or Associations provided training and (c) Training programmes developed within some European consortium projects.

34.4.1 University Based Programmes in ID Mental Health

Universities would typically offer specific bachelor and master's programmes on the overall topic on disabilities and/or ID in the department of education, special education, psychology and psychiatry. In worldwide, the UK, the USA, Ireland and Australia are examples of countries providing such University-based programme for higher degrees.

Postgraduate studies on a Master of Science level (MSc) in intellectual and developmental disabilities (IDD) offer advanced knowledge of psychological aspects of development in people with IDD, and aim to provide a grounding for students who are interested in studying later for a professional qualification in clinical or educational psychology, or a career in research. Such programs, designed for 1 or 2 years, provide practical knowledge in social psychology of IDD, skills in behaviour analysis, mental health assessment and intervention as well as knowledge and understanding of service issues in IDD. The programmes, frequently subject to charges, are on offer either as traditional in-person programmes or as online and distance-learning formats [22–24].

The University of New South Wales' Department of Developmental Disability neuropsychiatry is providing free online training in ID mental health [25], set up in partnership with other organisations in New South Wales. This ID mental health, e-learning platform provides training tracks tailored for three target groups: (a) carers and family, (b) health professionals, and (c) disability professionals [25]. In the UK, Imperial College London has developed an online training programme aimed at both paid and family carers of people with ID with a special emphasis on medicine review with the view to withdrawing unnecessary psychotropic prescriptions for the management of PB in adults with ID [26, 27].

34.4.2 Associations Promoting Advances in ID Mental Health

In many areas of the social field, non-governmental organisations (NGOs) are the key providers of services for vulnerable and socially marginalised groups, such as people with disabilities. The next section provides some examples of these organisations promoting ID mental health in Europe.

34.4.2.1 EASPD

In Europe, the *European Association of Service providers for Persons with Disabilities* (EASPD) has been promoting, for more than two decades, the views of currently 15,000 social services and their umbrella organisations. Apart from offering day-to-day support

for people with disabilities, the vast majority of these organisation promote equal opportunities for people with disabilities through effective and high-quality service systems [28] (for further information go to ► http://www.easpd.eu/en). However, in the health sector, and especially the mental health sector, equal opportunities are not assured for people with ID, with widespread inequalities for mental health and ID [29]. EASPD's activities regularly include awareness-rising campaigns on the need of appropriate staff qualification as well as European workshops on issues of health and mental health for people with ID.

34.4.2.2 ARFIE

Association de Recherche et de Formation sur l'Insertion en Europe, Association for Research and Training on Integration in Europe (ARFIE) assumes its responsibility by arranging consortiums developing appropriate training and continuous education programmes in the field of ID, in general, and ID mental health, in particular. Two of ARFIE's recent innovative training products relating to mental health and ID are as follows: (a) TRIADD and TRINNODD and (b) AGID project (see below). ARFIE's mixed membership includes from innovative service providers to leading researchers, which offers excellent conditions for setting up training projects to allow transfer of research into practice [30].

34.4.2.3 EAMHID

The *European Association for Mental Health and Intellectual Disability* (EAMHID), founded by leaders from academia, research and the clinical mental health field, contributes significantly with its biannual European conferences and pre-conference training workshops to the advances in research and practice in the field of ID. The purpose of the Association is to facilitate international cooperation and exchange of knowledge and experience in the field of mental health in people with an ID. Special emphasis is placed on the coordination and promotion of scientific activities and improving standards of care and support throughout Europe. EAMHID conferences offer a variety of formats attractive to the various professionals in the field of ID and mental health. Besides traditional formats like keynote presentations, solicited symposia, posters and individual presentations, a conference would typically encompass formats such as round-table discussions, coproductive symposia, a format of gathering up to five stakeholders to debate around an overall theme and half-day pre-congress courses offering training to improve skills based on recent evidence-based advances, for example, mental health assessment. Thus, EAMHID delegates benefit from accreditation, training credit points as required by most professions engaged in the field of mental health. For further information, see [31] ► https://eamhid.eu/. Finally, EAMHID keeps formal collaboration with other similar organisations like *The National Association for the Dually Diagnosed* (NADD) in the USA [32] (► http://thenadd.org/) or the *International Association for the Scientific Study on Intellectual and Developmental Disabilities* (IASSIDD) [33] (► https://www.iassidd.org/), and specifically, IASSIDD's Special Interest Group on Challenging Behaviour & Mental Health (SIRG/CBMH) [34] (► https://www.iassidd.org/content/challenging-behaviour-a-mental-health).

34.4.3 ID Mental Health: European Consortium-Driven Training Programmes

34.4.3.1 TRINNODD

The European training modules for staff supporting individuals with dual diagnosis TRINNODD (TRansfer of INNOvation for Dual Diagnosis) was a 2-year project (2008–2010) funded through the EU Leonardo da Vinci Lifelong Learning Programme [35]. TRINNODD aimed at updating the previous TRIADD (Teletraining Research and Information around Dual Diagnosis) project deliverables [36] and to transfer advanced knowledge on topics related to dual diagnoses (DD) to target groups, with little exposure to these issues in countries with little opportunities for continuous training options in that field. The contents of the training package to

develop were based on a survey of frontline staff and mental health professionals reporting their needs with respect to knowledge and skills to understand and handle mental health issues in people with ID. Findings not only revealed differences and commonalities in the needs between countries but also identified four key areas for training activities and training considerations: emotionality, quality of life, vulnerability and training methodology. Furthermore, modules developed with high priority were derived from the areas of stress prevention, assessment of quality of life of the person with ID as well as family members, complexity of the individual's vulnerability as well as psychosocial and contextual-environmental consideration (individual and community).

The development as well as the implementation of the training modules mainly followed the steps as defined in the knowledge transfer and exchange model (KTE) suggested by Saini and Brown [37]. This model considers, for the development of the training material, questions like: What is the knowledge and skill to transfer? What is the quality and evidence of the knowledge and skill? What are the limitations of the knowledge and skill? Further, considering the training session itself: Who is the audience? How can the message be tailored? How should the message be delivered?

Next, the use of the knowledge and/or skill as transmitted is reflected: Do the trainees use the product as expected? Is the product relevant for the trainees? And finally, the model considers the impact: Does using the product impact change on outcomes relevant to the goals, objectives and purposes of the KTE product? Training modules, with sessions totalling to 30 h, were delivered in Italy, Portugal, Romania and Spain. The training modules took in account a broad primary training background of the audience. Thus, trainees included physicians, psychologists, nurses, paramedical therapists, social workers and educators. The impact of the training was evaluated at two different points in time: First, directly upon delivery of the training, and second, 6 months after the delivery of the course in order to assess the impact on the trainees' daily practice. The training sessions themselves were positively evaluated by the trainees and the trainees reported 6 months later positive effects with regard to their professional life, such as improved competence to handle and/or to prevent stressful daily situations. TRINODD's materials as well as its achievements are located on [36] ► http://www.triadd.lu/index.htm. TRINODD's training material is disseminated through the Association de Recherche et de Formation sur l'Insertion en Europe (ARFIE) [30] (► www.arfie.eu).

34.4.3.2 AGID

Whereas TRINODD had its focus on mental health in general for adults with ID, AGID (Developing Training Modules for staff on Ageing and Disability Issues) focused on a bio-psycho-social perspective on the challenges of older people with ID [38]. The AGID project is funded by the EU Leonardo da Vinci Lifelong Learning Programme and co-funded by the Austrian 'Fonds Gesundes Österreich'. It was set up by mixed consortium of university-based academics and leaders form the service provision system, and aimed at the development of a free-to-use online training platform offered in five languages: English, French, German, Italian and Flemish [39] (► www.agid-project.eu).

The detection of topics with high relevance for continuous training in the area of ageing and ID was based on the appreciative inquiry methodology [40] using an inclusive strategy with regard to main stakeholders (e.g. health professionals, self-advocates, family members and frontline staff). This process leads to the definition and evidence-based development of six training modules:

1. Ageing process: Ageing process in general with a specific focus on people with ID.
2. Person-centred planning: Introducing to the basic conception and methodology of person-centred support aimed at improving participation and self-determination of an ageing person with ID.
3. Social network and communication of elderly people with ID: Strengthening the awareness of trainees for social networks of older people with ID, for example, the preventive function of social networks and

develop skills and techniques for maintaining, improving and initiating social contacts for older people with ID.
4. Emotional regulation for frontline staff: Provide staff with the necessary skills to reflect and systematically explore factors in their professional environment and to develop competencies in the application of strategies reducing stress, thus, contributing to prevent mental health problems in older people with ID as well as to prevent burnout of staff members.
5. Pathological ageing in people with ID: Transfer knowledge of common age-related diseases and disorders, competency in handling emergency, competency in effective communication with older people with ID and competency in creating a supportive environment for people with ID and dementia or depression.
6. Taking care: The main objectives of this module are to develop a professional attitude among staff and health professionals so they can be prepared and equipped to deal with the specific challenges when supporting older people with ID. The modules will train professionals on whom to implement 'good-care' in their everyday work while considering the self-determination approach.

An initial evaluation of AGID's e-learning platform based on 171 users revealed that overall users rated the platform attractive largely due to the multimedia content of the modules (images, interactive exercises and videos). Users reporting experience with e-learning platforms (n = 111), mainly the younger cohort, reported less need of support and training with the platform. In addition to the many positive comments received, there were some criticisms, with readability being one of the most cited. Nonetheless, the attractiveness of the platform had substantial acknowledgement by a large majority of the users, which is of particular importance as usability is considered as a significant factor. AGID's external evaluation focused on a general assessment of the project objectives, progress and outcomes, while pointing to issues like the importance of providing main theoretical reference for understanding the concept and measurement of quality of life or the need for an overview of the recent literature on policies and services, particularly in Europe.

34.5 World Perspectives

In low- and middle-income (LAMI) countries, there is generally a lack of well-trained therapists who could provide specialist interventions to support persons with ID/IDD and their families, especially with reference to the adult age. To have utility for LAMI countries, these programmes would need to be able to be delivered by non-specialist community workers or by family members of the persons with ID. In reference to the developmental age, reviews of parent training (both individual and group) carried out in LAMI countries indicate that the involvement of parents is both effective and cost-effective [41], although scientific rigour of available literature is scarce, especially for methods and outcome measures.

Since parents have frequent and consistent contact with their child across a range of settings, effective intervention, generalisation, maintenance and prevention are all more likely. Cost-effectiveness can be further improved if specialist professionals train paraprofessionals or lay volunteers to inform and support parents in turn [41]. Parent training outcomes, such as enrolment, dropout, consistent attendance and child improvement, have been associated with parental education, socio-economic status, expectations, values, marital relationships and being a single parent [42].

A training system that has already been effectively rolled out in the USA [43] and in Australia [44] and for which cross-cultural adaptations have been developed is represented by the Stepping Stones Triple P Parenting Program [45]. The World Health Organization (WHO) has implemented a caregiver skills training programme in Africa and in other LAMI countries across the world to train parents and key caregivers to improve outcome in children with IDD [46].

Training of professionals in the area of IDD in Nigeria is often incorporated into postgraduate paediatric and psychiatry training and accredited and supervised by National Postgraduate Medical College of Nigeria and West African College of Physicians in the West African sub-region [47]. Many sub-regions in Africa also have postgraduate training in Paediatric and Psychiatry, which contribute to training of professionals to attend to health needs of children with IDD. There is a pocket of mid-level professional occupational and speech therapists training schools across sub-Saharan Africa [48, 49].

In most Asian countries, the medical support for people with IDD focuses on basic physical care rather than meeting their psychiatric needs. It is well recognised that psychiatric disorders are common in both children and adults with ID [50, 51], but this area rarely receives adequate, if any, attention from the health administrators in Asian countries. The lack of specialist staff, particularly psychiatrists, is often offered as the major reason for not starting such services in this part of the world. Due to historical reasons, Hong Kong has adopted a British model for its medical education and specialist training. There are close ties between the Hong Kong College of Psychiatrists and the Royal College of Psychiatrists in the UK. To be an approved training scheme that is accredited by the Royal College of Psychiatrists of UK (so that trainees can sit for their examinations like MRCPsych), Hong Kong established its first formal psychiatric unit for adults (age 16 and above) with ID in 1995.

In Hong Kong, after registration as a psychiatric trainee, he or she has to go through a rotation where different types of clinical experiences are provided. The psychiatry of ID is among one of those specialties, but it is not mandatory. The training curriculum in this specialty consists of management of mental and behavioural problems in people with ID in inpatient, outpatient and community settings. Trainees are engage in multidisciplinary team work to tackle the different problems of this population under supervision. As the trainee goes through his training, he may sit for the Part I, II and III of the Fellowship Examination. He will be qualified as a specialist after completing at least 6 years of professional training and passing all three parts of the examination.

Among Asian countries, financial resources, manpower and expertise are always the most important challenges facing the development of medical and health-care services. Training and service development should go hand in hand and neither one should become a barrier for not providing appropriate mental health services for people with ID. Singapore should be appreciated for following the service development pathway of Hong Kong to set up a specialist psychiatric service for adults with ID and/or ASD. It is named the Adult Neurodevelopmental Service (ANDS) at the Institute of Mental Health (IMH) in Singapore. Singapore had invited a psychiatrist from Hong Kong to provide training and experience sharing before commissioning this specialist service. This is another example of how training and service development will eventually bring significant benefits to improve the total health care and quality of life of people with ID.

The Academy of the International Association for the Scientific Study of Intellectual and Developmental Disabilities (IASSIDD) has an International Visits Program, whereby individual members from any country may arrange to visit an accredited community agency, university or hospital for a period of approximately 2 weeks for the purposes of learning and training. The visitors will be provided with opportunities for attending classes, accompanying a support worker on his/her daily work, taking part in consultations and multidisciplinary meetings, assisting with research or a combination of these activities. An example is the study visit of the Rainbow Centre personnel of Singapore to three adult support services in the UK and the Netherlands in 2019 [52] (► www.iassidd.org/international-visits-program). Such programmes are considered to be valuable to many workers and trainees in Asian countries without specialist ID services.

In Australia and New Zealand, the number of psychiatrists who are specialising in IDD is promised to increase in the last decades. Of

course, the underlining need has been to provide adequate training for doctors in both the areas of physical and mental health. In both Australasian countries, this development has been slow. At the University of Sydney, the need for funding of special health clinics in the area of developmental disabilities was identified at the end of the 1990s but it took at least a decade before it was recognised. The inaugural Chair in Intellectual Disability Mental Health at the University of New South Wales was established in 2013. Of course, one of the factors which has inhibited both the training and delivery of mental health services for this population has been the slow recognition that people with ID do experience mental health conditions.

The body responsible for training and education of psychiatrists is the Royal Australia and New Zealand College of Psychiatrists (RANZCP). The highest standards in training pertaining to the psychiatry of IDD are promoted by the RANZCP Section of Psychiatry of Intellectual and Developmental Disabilities (SPIDD). This Section, through its Bi-national Section Committee, is also responsible for promoting and encouraging the study of psychiatry of IDD in all its aspects and the development of knowledge and practice, of the highest standard, for psychiatrists, trainee psychiatrists, medical students, related health professionals, students of related health professions, other medical practitioners and involved legal practitioners.

34.6 Further Considerations

In this chapter, we primarily concentrated on training for psychiatrists and psychologists. Because of lack of space we could not go into the detail of any training for other professionals such as nurses, social workers and speech and language therapists. Although we could not go into the detail of training for support (care) staff, a systematic review by Deb & Roberts [53] found 24 papers on staff training in ID. The authors divided the studies under (a) training in management, policy and intervention implementation; (b) training in behavioural issues and (c) training in the techniques of control and physical restraint. The authors found that most training increased staff knowledge on the subject and confidence to deal with mental health issues and PB in people with ID. However, the impact of training on the actual mental health and PB of people with ID they support is not known. In a more recent systematic review, Rose and colleagues [4] divided the type of staff training under three headings, namely (a) proactive, (b) reactive and (c) responsive training and support (see ◘ Table 34.1). Like Deb & Roberts, Rose and colleagues also found that the training by and large improved the knowledge and confidence among staff.

Further, future reviews on professional education and training should include the profession of *psychotherapist*. Psychotherapy as a profession is currently regulated by law in many European countries, however, there is no heterogenous curriculum in mental health in ID for these professionals. In a growing number of countries, psychotherapy for people with ID is included in the provisions financially supported by the national health insurance systems as there is good evidence on the effectiveness of talk-based therapies for this population [54]. Most recently, Germany has adopted a formal guideline for ID psychotherapy [55]. However, it is not known to what extent these trainings are mandatory in different countries.

Finally, *self-study options* are briefly addressed as these probably continue to constitute most relevant resources for those entering the field of ID mental health or ID in general when not having had the opportunity in their professional training to acquire appropriate knowledge and competencies in the area of ID mental health. In the UK, a family carer organisation, Challenging Behaviour Foundation (CBF), provides online material for family and paid carers on mental health issues in people with ID [56] (► www.challengingbehaviour.org.uk). Also, in the UK, Royal College of Psychiatrists' MindEd programme provides free online material on mental health issues in ID [57] (► www.minded.org.uk). Similarly, in the UK, both the British Psychological Society [58] (► www.

bps.org.uk) and British Institute of Learning Disabilities (► www.bild.org.uk) provide online material for professionals, carers on various aspects of mental health and PB in people with ID including Positive Behaviour Support (PBS). In Toronto Canada, Surrey Place Center provides online material on mental health issues in ID [59] (► www.surreyplace.ca). Autism Europe IPA+ project provides online learning modules on autism [60] (► http://ipa-project.eu/training/).

There are also a number of textbooks and journals that are a great source of knowledge and information in the field. Selective examples of textbooks include (a) *Psychiatric and Behavioural Disorders in Intellectual and Developmental Disabilities* [61], (b) *Diagnostic manual-Intellectual disability: A textbook of diagnosis of mental disorders in persons with intellectual disability, second edition* [62], (c) *The Oxford Textbook of Psychiatry of Intellectual Disabilities* [63], (d) *Advances of clinical psychology in the field of ID mental health* [64], (e) *Understanding and responding to behaviour that challenges in intellectual disabilities: A Handbook for those who provide support* [65], (f) *Handbook of Evidence-Based Practices in Intellectual and Developmental Disabilities, Evidence-Based Practices in Behavioral Health* [66], (g) ► www.intellectualdisability.info [67], (h) *Clinical Topics in the Psychiatry of Disorders of Intellectual Development* [68], (i) *Handbook of Psychopathology in Intellectual Disability: Research, Practice, and Policy, Autism and Child Psychopathology Series* [69], (j) *Intellectual disability and ill health-A review of evidence* [70], (k) *Intellectual Disability Psychiatry: a Practical Handbook* [71], (l) *Practice guidelines for the assessment and diagnosis of mental health problems in adults with intellectual disability (European Association for Mental Health in Intellectual Disability)* [72]; ► www.iassid.org [33], etc.

Some of the journals in the field include (a) *Journal of Intellectual Disability Research* (ed. Richard Hastings), (b) *Journal of Applied Research in Intellectual Disabilities* (eds. Peter Langdon & Jonathan Weiss), (c) *Research in Developmental Disabilities* (ed. Dagmara Dimitriou), (d) *American Journal of Intellectual and Developmental Disabilities* (ed. Frank Symons), (e) *Journal of Mental Health Research in Intellectual Disabilities* (ed. Angela Hassiotis), (f) *Advances in Mental Health and Intellectual Disabilities* (eds. Sarah Blainey & Debbie Spain), (g) *Journal of Policy and Practice in Intellectual Disabilities* (eds. Rhonda Faragher & Laurence Taggart), etc.

34.7 Conclusion and Outlook

Generally, it is considered that higher education training as well as postgraduate education or advanced professional training with respect to mental health are key resources and crucial factors when assessing the quality of a country's mental health provision system [73]. This chapter revealed major disparities in and lack of formal training curricula for mental health professionals in ID mental health, which highlights the need for immediate action in this area. At the European level, there have been suggestions for establishing a European training programme on mental health in ID [35, 74]. However, we have to consider the fact that, within the European Union, health policy is defined at the national level, so a consistent approach to developing a Europe-wide curriculum may have its challenges. However, this also highlights the need for relevant organisations and key players to come together to develop a pan-European curriculum for training in mental health in ID, which could be adapted by individual countries for their own use. This may have a positive impact on the consumers' right for equity in ID mental health services within and between member countries. Good mental health is a significant factor in the successful integration and inclusion of people with ID in the wider society [75].

Key Points

- Most countries in the world do not provide adequate training for professionals who look after the mental health of people with intellectual disabilities.
- Some national and international organisations and associations have set good

examples of providing specific inter-country training in this area.
- There is a need for action to develop a mental health education and training strategy for people with intellectual disabilities that can be adapted to the context of each country.

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Mental Health Services

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Contents

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■ **Overview**

Specialist mental health (MH) services for people with intellectual disability (ID) have been a feature for over 30 years. Early pioneers such as Bouras and colleagues were not only instrumental in developing MH in ID services to complement mental health pathways but also developing Mental Health in Learning Disabilities (MHiLD) as a specialist field of psychiatry. Since the early model, put forward by Bouras, several service models have come into existence; however, robust research into the effectiveness of different models is lacking. This is for a number of reasons such as the methodology used, focussing on different outcomes, changing political picture and subtle changes in function and who is eligible for specialist services. More recently, to achieve equity of provision for people with an intellectual disability, there have been several quality initiatives aimed at improving access to services; however, this has seen varied results across different locations. This chapter will provide the historical context in the development of specialist mental health services for people with ID including a discussion of service models with specific examples including how we measure quality in services. An international perspective will be presented as well as how individual countries may take a national approach to the planning of services.

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Learning Objectives

This chapter will provide a number of learning objectives including the following:

- An awareness of the developments of mental health services for people with intellectual disability and autism over the past decades from an international perspective.
- Increased knowledge on how to improve quality in services and how to measure the utilisations of services.
- An understanding on developing a community-based approach as illustrated by a specialist mental health model developed by Bouras and colleagues in the UK.

35.1 Introduction

The ongoing debate for the past 30 years has been on the model of mental health services for people with intellectual disability (ID), specifically around how these services should be delivered. This debate is focused in the UK on the development of services following the closure of the institutions leading to the development of community-based mental health services [1, 2]. Although our knowledge of mental disorder and how it affects people with ID has increased over the past three decades, this has not resulted in adequate or equity of service provision. As a result, mental and physical health care is more difficult for people with ID to access than for those in the general population [3–6]. This has led to a dialogue on how best to deliver community-based mental health care, whether through specialist teams or by improving access to mainstream mental health services.

In most parts of the developed world, the focus has been also on closing the institutions and at the same time looking at whether disability services will provide for the mental health care of people with ID or whether this will be the remit of mainstream mental health services. One of the perceived benefits to community integration was that there would be a reduction in behavioural problems. However, for many, the transition resulted in an increase in problem behaviours and mental health problems in what was known as 'relocation syndrome' [7]. In countries, such as Australia, the provision of community-based services has led to either disability services looking to provide for the mental health care of people with ID or by improving provision within mainstream health services [8]. This is an illustration of how service delivery and models can differ by country. For example, in Ireland 'parents and friends' groups set up community-based residential services promoting normalisation [9]. In Spain, a specialist service model as a part of the Catalan Mental Health Plan (2006–2010) was developed, inspired by the Mental Health in Learning Disabilities (MHiLD) model in South East London, UK [10]. While in the

Netherlands, the development of a community model comprised of regional centres and the use of outreach models as an alternative to large asylums. This followed a randomised controlled trial in the 1990s that found no differences in outcomes in those with mild or borderline ID receiving hospital or community-based care [11].

Recent national policy in England has more and more emphasised the need to improve awareness and support in access to mainstream services for people with ID and those with/or autism [12]. At the same time, there has been a debate on how we develop specialist teams and how we improve outcomes for people with ID as well as those with/or autism spectrum disorder who have mental health needs. For example, the Royal College of Psychiatrists recently developed the role of a champion for autism within the College, although those with autism have received less attention from the national policy in transforming care [13].

In other countries, such as Canada, there has been an increasing understanding that some people with ID and mental health needs either have access to emergency services or inpatient service, with little in between. The research is mainly focused on the utilisation of these emergency and inpatient services and the benefits to the individuals and families [14].

In England, due to recent national policy on reducing the number of people with ID who are in hospital, there has been a development particularly around services for those with challenging behaviour or forensic history. This national policy has led to the model as described in Building the Right Support [12]. The central principles of this new model of service is that every person with ID should have person-centred care and live in the community and this care should come from mainstream mental health services.

35.2 Quality of Services

Measuring service outcomes has always been difficult given changing functions over time including fundamental changes to the client group served and their level of risk. One attempt was 'the matrix model' [15], which developed an international framework to compare services across five European countries, entitled the BIOMED-MEROPE project [16]. Another initiative aimed at achieving equity of mental health care for people with ID was the Green Light Tool Kit (GLTK), which was introduced in 2003 in England [17]. The GLTK provides a framework and self-audit toolkit aimed at improving mental health services for people with ID, providing a benchmark of what individuals should expect from services by including quality outcomes for services to meet. In 2013, the National Development Team for Inclusion (NDTi) produced a revised GLTK, also aimed at those with autism spectrum disorder (ASD) as well as ID [18]. This followed 'Reasonably Adjusted', a report outlining progress in terms of reasonable adjustments that mental health services were already putting in place for people with ID and/or ASD [19]. The latest GLTK has 27 items to self-report with items 1–9 forming the 'basic' audit, which examines physical health, eligibility and access, secure services, safeguarding, assessment, equalities, personalisation, staff attitudes and values and accessible information; items 10–18, titled the 'better' audit examine research, health and care records and care plans, local plans, how specialist services relate to local provision, skilled workforce, people needing personal care, user involvement in the governance of the service, psychological therapies and working together. The final section, items 19–27, titled the 'best' audit, examines advocacy, commissioning, buildings and environments, leadership, family and friends, employment support, checking services, monitoring and challenging behaviour. In terms of the impact of the Green Light Work, reflections on examples from five NHS Trusts [20] illustrate that the implementation is still a work in progress and in spite of 'positive steps' more time is needed to focus on the challenges and solutions faced. However, the commissioning of ID services in the UK has not always been seen as a priority in spite of a plethora of guidance to the contrary, which has often

seen best practice guidance such as closer to home, least restrictive environments and community placements ignored either due to lack of infrastructure or inability to see past current practices. For change, limitations in current service provision need to be addressed. One method is by quality improvement initiatives. Quality improvement is a systematic approach to improving health services based on iterative changes, continuous testing and measurement and empowerment of front-line teams [21]. Quality improvement is dependent on good leadership with sustained focus over time and a strong emphasis on service user involvement. The definition of quality in the National Health Service in the UK is based on three key areas of:

1. 'Patient safety', which is doing no harm to service users.
2. Experience of care, which is compassionate and delivered with dignity and respect.
3. Effectiveness of care, preventing people from becoming ill, improving quality of life and helping service users to recover.

These principles are applicable to mental health services for people with ID. However, the issue for people with ID and/or ASD is ensuring the effectiveness of care with such a low evidence base. All services can provide care that is safe for this group of service users in terms of their vulnerabilities, such as increased risk of physical health problems, for example, epilepsy, and the risk from others due to exploitation or abuse as happened at Winterbourne Hospital [22]. The issue of service user experience for people with ID and/or ASD also needs to take into account their social, emotional and sensory needs in order to deliver the positive experience for people when in contact with services [23].

A person with ID should have access to specialist health and social care, including access to high-quality inpatient care, when they need it. The debate needs to move on from just looking at models of care to instead asking what high-quality mental health care for people is with ID. The evidence base to date has mainly been on the characteristics of people with ID and mental health needs accessing services, however, in the future, there should be more emphasis on looking more on how to improve long-term outcomes. In countries with highly established mental health services for the wider population, which has an emphasis on prevention such as early intervention and health promotion, these approaches should also include those with ID and/or ASD into these quality initiatives [24].

The question comes what a high-quality service is. Around the world the mental health care for people with ID has very much been dependent on the availability of resources and as well as the availability of experts and specialists. A group in New South Wales in Australia [25] have looked at core models of health service delivery for people with ID. The review identified nine models of health service delivery from the UK including two from Australia. All these models emphasised the need for inter-agency working and highlighted the need for improving integration between mainstream and specialist mental health services. A number of barriers to accessing adequate mental health care for people with ID have been identified previously in Australia but are relevant to other countries [8]. These include the following:

- Clarity concerning roles and responsibilities and service coordination between disability services and mental health service providers.
- Training of mental health providers.
- Coherent service models and funding for ID mental health services.
- Specific inclusion of people with an ID in mental health policy.

The conclusion of this literature review was the need for high-quality research as none of the reviewed studies were meeting the requirements for a systematic review. To deliver services that are effective, safe and give service users a good experience, there is a need to collect further evidence to demonstrate how we can consistently improve the long-term outcomes of people with ID and/or ASD who are accessing mental health care.

The evidence so far for the improvement of mental health care in those with ID does

indicate the importance of inter-agency working [26]. In addition, specialists with expertise in assessment and management to make an informed formulation in the context of the individual's lives and their carers are required. A number of countries are responding to developing close inter-agency relationships and proving specialised psychiatric care in response to the evidence [27]. Difficulties with communication and emotional understanding are important in the assessment of people with ID. Those from a non-specialist background may not have the necessary skills to understand the needs of this population running the risk of over or underdiagnosing of the individuals' mental health needs [28]. Surveys within countries continually highlight the need to ensure adequate training for psychiatrists in the mental health needs of people with ID [29].

The evidence on service user experience is limited but interviews with people with ID accessing health-care services report that half of the participants have been treated unfairly or been discriminated against [30]. These accounts confirmed negative staff attitudes and failure of services to make reasonable adjustments. Some reported evidence on the experience of those with ID from different ethnic backgrounds in accessing mental health services [31]. Although the needs of people with ID from different ethnic communities are being increasingly recognised, they remain unrecognised in most mental health services research. In a comparison of two ethnic communities in South London, the black ethnic minority group were less positive about their overall experience, including the use of medication. There is little evidence as to the experience of individuals with autism of accessing mental health services. However, high-quality services do exist for those with ASD and there is increasing evidence to support the importance of accessible services which include a comprehensive assessment of their mental health needs [32]. The problems and burden of mental illness in people with ASD have been recognised for many years [33] but the delivery of good-quality mental health care in this group is not keeping pace with needs.

There is little research into the effectiveness of specialist models of mental health care for people with intellectual disabilities. However, what is agreed is that high-quality specialist services are characterised by inter-agency working and include professionals with appropriate expertise.

35.3 Service Utilisation

Utilisation of mental and behavioural services is the most commonly used type of health service for those with ASD, with up to 35–55% accessing such services annually in the USA [34]. There is still little evidence around the world on the utilisation of services by people with autism and mental health needs. This study from the USA indicates that those with ASD in contact with mental health services, which is evidence from mainly children and adolescents, found that those receiving individual or family therapy have a lower economic cost than those receiving mixed therapy, which included both family and individual therapy. Those having a co-morbid mental disorder were not associated with any substantial difference in service cost to those without a mental health problem. The average number of sessions per participant was lower than recommended through clinical guidelines. This reduced use of services may be due to the fact that the individuals found other services, such as specialist schools or community-based services, more helpful rather than attending mental health services, which may have presented too greater a burden for the families. Further evidence is needed to consider how to deliver effective therapy in appropriate doses for this group of service users. Social workers provided the lowest cost service, and this may be related to their training. They received training as case managers, and this may lead to reduced costs and benefits. Also, the younger the person, regarding children with autism, higher the dropout rates. The impact of this high

dropout rate may lead to decreased chances of improvement later on, so it is important to address any barriers to ongoing services for young people with autism and mental health needs [35].

Others have attempted to evaluate the mental health needs of people with ID accessing services as a way to understanding utilisation of services which has led to the development of a cluster-based approach. In the UK, this has been based on the Health of the Nation Outcome tool [36], which led to the development of 21 established mental health clusters that are divided into three groupings of psychosis, non-psychosis and organic conditions. This has resulted in a description of a further eight clusters specific to those with ID. The use of specific clusters is seen as an extension of these mental health clusters to ensure the better description of service users using specialist ID health services. These eight clusters including one of a low-level need, such as 'engagement minor support needs associated with mild ID (no autism)' to a cluster with a higher need of 'severe behaviour that challenge associated with autism and mild to moderate ID'. Within these eight clusters, there are three physical health clusters with their own degrees of severity. The challenging behaviour clusters and three physical health clusters together with the mental health clusters may allow greater comparison of services utilisation and better planning and resources within defined geographical areas.

The evidence to date on hospital admissions for people with ID shows those at increased risk for hospitalisation include: younger age, diagnosis of a psychiatric disorder, high levels of irritability, less severe ID, from a black ethnic minority group and not having a stable home [37]. However, the implementation of a community-based crisis and intervention programme can impact on these risk factors and so reduce the need for hospitalisation. The Mansell report [38, 39], first published in 1992, before a second edition in 2007, stated the need for robust supported housing in the community for those with challenging behaviour. Mansell emphasised it is not just the resources, but the quality of the services delivered that makes the difference. However, further evidence is still required on which aspects of community-based models and interventions reduce hospital admissions most effectively in the long term.

35.4 Service Models and Development of Services

In many areas, a lack of local community services has seen an artificial demand for unnecessary inpatient services that have warehoused and restricted people often in out-of-area placements [40–44]. It is only the scandal from Winterbourne view that has seen some attempts to address this problem. This section describes currently recognised models that have been established over time for illustrating an international perspective along with the recent national model for England.

Barriers to service improvement are often underpinned by historical and current philosophies of how society treats people with ID. Although there is outrage to scandals involving abuse, for example, [22, 45, 46], there is also disagreement on basic concepts such as normalisation and how these are interpreted day to day and misunderstanding of mental illness and the abilities of people with ID. This is reflected in the wide variation of approaches and attitudes of providers to certain treatments, for example, access to psychological interventions through Improving Access to Psychological Therapies (IAPT) services. The programme began in 2008 and targeted the treatment of adults with anxiety disorders and depression in England. Although over 900,000 people access IAPT services each year, there are still those who do not offer a service to people with ID. In the UK, there are variations in how and who delivers mental health services for people with ID. The official policy states that people with ID should access mainstream general mental health services [47, 48] with the addition of specialist mental health services for those with complex mental health needs. This group will often present with poor treatment outcomes, multiple admissions and, in some cases, are disadvantaged outside of

specialist services. In the UK, since the advent of community care, the mental health care for people with ID was the remit of Community Intellectual Disabilities Team that are primary care based and a type of one stop shop for looking after the healthcare of people with ID [49]. Arguably, the most celebrated mental health in ID service design is the Bouras and colleagues Mental Health in Learning Disability (MHiLD) model [10, 50, 51]. The MHiLD model predates the original Mansell Report, which put forward (as did the revised version) what is still considered the gold standard of how services should operate; and provided an impetus for the development of person-centred services. The MHiLD service is a dedicated service that enhances and complements existing adult mental health services for people with ID. The approach to early intervention means that those presenting with mental health issues have a chance to avoid potential negative consequences if left untreated or unidentified. This model as well as delivering specialist services has highlighted the need for specialist mental health services and right to mental health care for people with ID [26, 52–54]. Using a tripartite approach, Bouras and colleagues understood the need for service delivery, research and training as essential components of service delivery and set up the Estia Centre to support evidence-based service delivery. It is some years later that Academic Health Science Centres (AHSC) (a similar model) was adopted in the form of partnerships between Universities and large health Trusts in the UK. One of the strengths of the MHiLD model is its range of clinical and educational interventions [10] aimed at mental well-being and recovery, which include the following:

- Specialist assessment (e.g. mental health, neurodevelopmental or risk), all involving a detailed and holistic bio-psycho-social approach.
- Specialist interventions (including the development of an individualised care plan, management strategies, psychoeducation and training).
- Follow-up and review of care plans and mental health needs.
- Follow-up and review of psychotropic medication.
- Therapeutic sessions, support and reassurance.
- Crisis resolution.
- Care programme approach (CPA) coordination for people with ID who have severe mental illness or severe behavioural problems and where there is a high degree of risk.
- Mental health promotion for people with ID.
- Skills training for local ID service providers.
- Carer and family education and support.
- Court and other specialist reports.
- Consultancy and participating in delivering academic courses.
- Use of evidence-based approaches including specialist outcome measures of clinical effectiveness.
- Regular-quality audits and service evaluation.

The concept of models used with service user groups with long-term conditions, such as the chronic care model, which improves outcomes of chronic health conditions in primary care settings, had been looked at to adapt for service models for people with ID. The chronic care model is a framework based on a service user-centred approach with a positive alliance among the family, the carers, the service users and the practitioners. This approach, as described in the USA, ensures that community resources are carefully co-ordinated, follow evidence-based practice, multidisciplinary approach and take a lifelong developmental focussed framework. All members of a multidisciplinary team would receive extensive and ongoing training and supervision to a high level of expertise. In addition, integration into wider, general medical services leads to improved service user satisfaction and better health outcomes. This is exampled in the Centre for Autism and Development Disabilities (CADD) in Pennsylvania, which is an integrated health-care approach for individuals with ASD, or ID, based on the health-care home model, with the emphasis on service user-centred care [55].

However, there are challenges in delivering such specialist teams in terms of training, supervision and availability of specialist to meet the needs of a specific local population.

Others have advocated services to be delivered through a specific therapeutic approach, such as positive behavioural support [56], and that the best way to deliver this type of approach is through practice training in the real environment and reinforcements of staff practice through the local manage, practice leader level [57]. Models that provide easy access to community services and skilled professionals result in better outcomes [58]. Just arguing for more community service does not assure the importance of the quality of the service and does not necessarily address the need of the individual service user. Therefore, future research needs to focus on how the delivery of specialist support and services leads to lifelong improved outcomes for people with ID and/or autism who have mental health needs. Rather than the diluted approach of providing mainstream services to all [59], the evidence is always in the direction of specialist models being most effective [60]. The best example is developing the specialist MHiLD Teams, as already mentioned, in South London [10, 50, 51], where the specialist team support the critical mass of expertise and the development of the speciality, whereas a total generic approach undermines the importance of the need for expertise. This issue is not about mental hospital care or inappropriate care but what makes a difference in terms of outcomes and quality of life.

The introduction of specialist services requires the support of local commissioners along with a strategic vision at a national or state level. A dedicated service model that enhances and complements existing adult mental health services for people with ID seems to be the most effective model.

35.4.1 Service Planning

In the planning of services for people with ID and/or ASD, there is a need to see the wider context of their lives. For example, the level of mental health utilisation may differ according to ethnicity. In a systematic review [68], six studies reported an association between cultural and ethnic factors and mental health utilisation. These included higher proportion of African Caribbean and lower proportions of South Asian referred to MHID services in the UK, while in an Indiana study, African Americans have more doctors' visits. Some of the explanations put forward to try and understand this are utilisation of existing support networks. People with ID and/or autism will commonly be dependent on others for their care and require support from a variety of local agencies. Therefore, it is important that there are clearly defined working relationships between mainstream mental health services and specialist services, such as autism community teams and community ID teams. Whatever the service that is being provided for people with ID, it must have adequate psychiatric input, particularly in the diagnosis of complex presentations with a co-morbid mental disorder [51, 53]. The challenge comes when a person with ID and/or autism requires inpatient hospital care. Few mental health units have the environment to protect an individual with autism from bullying and harassment from other service users. The staff need expertise in the sensory difficulties, communication overload and the need for rituals. In addition, therapeutic approaches are needed to be adapted, including recognised psychoeducational programmes [61]. The physical environment needs to be suitable in terms of layout, noise level, being of low stimulus and suitable social clues. One major issue is a lack of meaningful user involvement in the development and planning of services. There is a need for mutual governance when developing services through service user involvement [17, 20, 47, 48]. Although this is an aspiration of many organisations, often this agenda is left to individuals or teams to ensure involvement [31, 62, 63].

The shifting service module from institutionalised hospital-based care to community-based services with a person-centred approach has been implemented in several countries. In the UK, as already mentioned, there has been an increasing shift to improve access to mainstream mental health services with the specialist services being more for those who have more complex needs. This debate between

specialists versus mainstream service continues but with an increasing acknowledgement that access to both types of services should be available [64]. The response is to some extent dependent on the available resources and expertise in each country; however, in most parts of the world, there has been the development of more specialist health teams. For example, in the Netherlands, the developments of services have been led by physicians. Specialist psychiatric unit for people with ID has been delivering care in Hong Kong since 1995 [65, 66]. In Australia, there are no specialist inpatient facilities for people with ID but there are specialist tertiary services which will meet the mental health needs of people with ID, for example, in Victoria [67].

35.4.2 International Perspective

In line with national and international policies and legislation, services for children and adults with ID are widely provided by both the governmental and non-governmental organisations. World Health Organization reported that only 39% of countries have policies or programmes related to ID [69]. There is a significant variation among the countries regarding the availability of mental health services for this population [70]. Some countries have highly specialised services [71], whereas other countries have limited or no services. This variation can be explained by the lack of clear health policy, lack of comprehensive and up-to-date mental health legislation, limited health-care funds and resources, a paucity of trained psychiatrists and mental health professionals and problems in deinstitutionalisation [72, 73].

There are very few studies that evaluated the effectiveness of mental health services at an international level in addressing this population by randomised controlled trials. In a study conducted across nine Southeast Asian countries, reported that all of those had some mental health services for this population [73]. However, these services varied by their type and range. In three places, Hong Kong, Taiwan and South Korea, there were provision of various types of psychiatric services such as inpatient, outpatient, day hospital and outreach service, for both children and adults with ID, whereas Vietnam provided only psychiatric services to children with ID.

Some reviews have discussed the current status of services in Latin America, South Africa, China, Greece, Asia, India, Taiwan, Serbia and Bulgaria [70, 74–81]. Firstly, these studies reported that knowledge about the effectiveness of these services is limited. Secondly, all countries are alerted about the existence of mental health problems in people with ID and have developed mental health services to tackle that. However, these services fail to cultivate the urgent need for creating a strong network of specialised mental health services for this population. These services are largely concentrated in secondary and tertiary centres of towns and cities, and the rural areas are marginalised [74]. Additionally, these services are provided by inpatient psychiatric units at district general hospitals and specialised psychiatric hospitals, which are mainly located in big University centres only. There is a lack of inpatient specialised mental health services for this population. In addition, in many other places, there are underdeveloped outpatient mental health services that are not enough to provide proper mental health care [81]. Kwok and Cheung [82] commented that decentralisation has added to the burden of general practitioners to manage physical and mental co-morbidities among people with ID. There are major gaps in treatment for people with ID in the community due to a number of factors related to their restricted mobility, behavioural and communication problems, sensory impairments along with the overall lower level of health professional skills and awareness to manage such complex cases.

Further, there are studies that focus primarily on the ethnic variation in utilisation of mental health service among children and young people with ID in high- and low-income countries [83–87]. Findings revealed that, in the UK, South Asian children and adults with ID have lower use of these services than Europeans of Caucasian or African origin [&, 83, 86, 87]. In 2011, Durà-Vilà & Hodes [87] reviewed four studies from three low-income countries – South Africa, Kosovo and

Pakistan –, to see the uptake of mental health services in children and young people with ID and concluded that there may be delays in seeking mental health service that often are related to culturally shaped ideas about ID.

35.4.3 Example of a National Service Model from England

The term learning disability used below is the term still commonly used in the UK to mean ID. In the UK, the principles underlying future service provision is set out in 'Building the right support', which is the national plan for the service model in England for people with ID and/or autism who display challenging behaviour, including those with a mental health need [12]. The principles can be applied in a strategic approach to developing services and these principles are listed below.

1. People should be supported to have a good and meaningful everyday life – through access to activities and services such as early-years services, education, employment, social and sports/leisure; and support to develop and maintain good relationships.
2. Care and support should be person-centred, planned, proactive and coordinated – with early intervention and preventative support based on sophisticated risk stratification of the local population, person-centred care and support plans, and local care and support navigators/keyworkers to coordinate services set out in the care and support plan.
3. People should have choice and control over how their health and care needs are met – with information about care and support in formats people can understand, the expansion of personal budgets, personal health budgets and integrated personal budgets, and strong independent advocacy.
4. People with ID and/or autism should be supported to live in the community with support from and for their families/carers as well as paid support and care staff – with training made available for families/carers, support and respite for families/carers, alternative short-term accommodation for people to use briefly in a time of crisis, and paid care and support staff trained and experienced in supporting people who display behaviour that challenges.
5. People should have a choice about where and with whom they live – with a choice of housing including small-scale supported living, and the offer of settled accommodation.
6. People should get good care and support from mainstream NHS services, using NICE guidelines and quality standards – with annual health checks for all those over the age of 14, Health Action Plans, Hospital Passports where appropriate, liaison workers in universal services to help them meet the needs of service users with ID and/or ASD, and schemes to ensure universal services are meeting the needs of people with a learning disability and/or autism (such as quality checker schemes and use of the Green Light Toolkit).
7. People with ID and/or autism should be able to access specialist health and social care support in the community – via integrated specialist multidisciplinary health and social care teams, with that support available on an intensive 24/7 basis when necessary.
8. When necessary, people should be able to get support to stay out of trouble with reasonable adjustments made to universal services aimed at reducing or preventing antisocial or 'offending' behaviour, liaison and diversion schemes in the criminal justice system, and a community forensic health and care function to support people who may pose a risk to others in the community.
9. When necessary, when their health needs cannot be met in the community, they should be able to access high-quality assessment and treatment in a hospital setting, staying no longer than they need to, with pre-admission checks to ensure hospital care is the right solution and discharge planning starting from the point of admission or before.

This approach allows services to develop to meet the increasing complexity of need with a foundation of services at community level,

through primary care and non-specialist services, which are available to people with ID and/or ASD as they are for the wider population [88]. These services need to have a preventative approach in promoting good mental health and well-being by encouraging healthy lifestyles and by reducing the risk for social isolation that many people with ID and/or ASD experience [24]. There is a need for more specialist well-developed community and inpatient service for those with complex presentations, including those with high-risk behaviours and those resistant to community-based treatment interventions. The literature continues to support that the most effective care for people with ID in the community is provided by services that are person-centred, multi-agency and with evidence-based therapeutic approaches [26]. There is an ongoing need to ensure that we have sufficient numbers of specialist mental health professions including nurses, psychologists and psychiatrists who can provide the expertise to improve outcomes and quality of life [89]. Professionals should act in accordance with legislation covering consent, capacity and decision-making. The service should be able to respond to crisis and there should be provision for out-of-hours care [90].

NICE guidelines on adults with autism (Clinical Guidelines 142) from England [91] describe the concept of local care pathways as the way forward in developing services for this group. These pathways ensure integrated delivery service across all care settings with a range of evidence-based interventions at each stage of the pathway and supporting adults with autism in their choice of interventions. These care pathways are designed to improve the overall quality of health care by promoting organised, efficient service based on the best evidence to optimise service user outcomes.

Tip

- The importance of recognising the need for strategic developments of services with a person-centred approach.
- Services cannot be delivered by one agency but require a multi-agency approach.
- Services must be planned in partnership with service users, their families and carers, respecting the diversity of people with ID requiring mental health care.

In conclusion, despite the existing concern about mental health services for individuals with ID, these services are limited by poor availability, accessibility and evidence of effectiveness [92]. There is a need to know and understand the true prevalence of ID and association of mental health problem [93]. Thus, there are more actions that remain to be done in this area; one is greater investment in mental health services and second is accessible for both urban and rural areas. Given the evidence that people with ID suffer mental health problems at greater rates and often atypically when compared to the general population, there will be always a need for specialist provision for those with most complex needs. The international evolution of comparable models is dependent on the sharing of knowledge, training and technology. In spite of progress and scandals that have made providers and commissioners reflect on what makes up suitable services, the main challenges are long standing [94] and include access to both mainstream and specialist MH services, clear interfaces with other agencies to provide joined up working, proactive commissioning strategies, development of academic centres and further research into service models that deliver improved outcomes.

The need to improve mental health care for people with ID and/or autism spectrum disorder must be based on a strategic approach informed by local and national policy using

the best available evidence. The emphasis needs to shift from what is the best model to which service produces the best outcomes in terms of service user safety, the experience of the person with ID and/or autism and the families that in the long term produce sustainable improved outcomes. This improvement in outcome is not only in the reduction of symptoms and challenging behaviour but also in the well-being and the quality of life of the person. For this to happen, services must be person centred, have staff with sufficient expertise and continue to respect the rights of the person with ID and/or autism spectrum disorder. Each locality within a country is different in terms of the needs of the population and their strengths and weaknesses. Service users come from a diversity of backgrounds but what is important is that the strengths of the local area and services are mobilised through a process of co-production of services developed with the person with ID and/or autism and their families acting as champions.

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Key Points

- There should be care pathways that cover all health, social care, support and education services with each defining their own responsibility clearly for each service.
- A consistency of treatment and designated staff coordinating the care across the different care pathways with a focus on outcome and clear links to other pathways, such as for those with physical health problems.
- Specialist ID and mental health services should have clear aims to ensure that people's needs are met, including those of the families, carers and care workers who should be actively involved in any assessment and treatment of the individual.

Through the leadership of practitioners who have the courage and wisdom to build on the local capacity to overcome weaknesses such as lack of good policy or funding, there will be a constant drive to improve the quality of mental health care for people with ID and/or autism [95–97]. This chapter was written prior to the Covid-19 pandemic and as yet we do not know the full impact of the pandemic on the mental health of people with ID but restrictions on their lives and the increasing use of visual consultations may have long-term consequences that will need to be accounted for in the future planning of mental health services for people with ID and/or autism ([98]; see ► Chap. 43).

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Costing Intellectual Disability Services and Support

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Contents

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Learning Objectives

Costing is a useful approach in its own right but is also a key component in an economic evaluation. This chapter focuses on the estimation of costs incurred by providers as a result of treatment programmes or health service options. Before costs can be estimated, there are three interrelated stages: identification, measurement and valuation. These stages provide a general framework as many of the considerations related to the costing estimation exercise are dependent on the analytical viewpoint or perspective. A distinction is made between intervention and non-intervention to provide a framework for estimating costs in intellectual disabilities. The aim of this chapter is to provide readers with a practical guide to cost estimation in intellectual disability research. We focus on particular categories that have particular relevance to various decision-makers and are important for analysts, such as training, accommodation, health and social care professional input, and caregivers' time inputs.

After working through this chapter, you will be able to:

- Understand why health economics is relevant in intellectual disabilities
- Explain the main steps in cost estimation
- Understand what methods can be used to identify, measure and value resources and support in intellectual disabilities

36.1 Introduction

A comprehensive estimate of the costs of intellectual disability (ID) was reported in a 2016 review by Salvador-Carulla and colleagues [1]. Costs were derived from a model that combined epidemiological and economic data in 30 European countries [2]. The cost (in Euros PPP 2010 prices) of ID (referred to as mental retardation in the article) across all 30 European countries was Euros 43.3 billion, higher than child and adolescent disorders (Euros 21.3 billion), traumatic brain injury (Euros 33.0 billion), personality disorders (Euros 27.3 billion) and multiple sclerosis (Euros 14.6 billion) [3]. These high costs in ID were based on the use of healthcare resources but ID has far-reaching impacts on other sectors, more specifically the social care sector, largely driven by the residential services that respond to the needs of service users [4–9]. In addition, there are also hidden impacts on the families and friends of the person with an ID and the rest of society which are often overlooked [10]. In a study conducted in Australia, Doran et al. [10] found that cost for people with ID was AUS$14, 720 billion (2005–2006) annually of which caregiver time lost contributed 85% of the total costs.

An awareness of the monetary sacrifices involved in providing treatments, services and support to individuals with intellectual disabilities is necessary and important for stakeholders at all levels be they strategic decision-makers such as commissioners of services, charitable organisations or families, as it is often the consequences of these decisions that can be used to influence policy. Consider, for example, government agencies would need information on the types and amount of out-of-pocket expenditure incurred by families to care for people with ID if they are to provide benefit payments that can be used to recompense families and facilitate purchases of non-statutory services and support. But influencing policy responses is not the only reason cost estimates are important. They are also necessary because those who design and implement services recognise that resources will always be scarce relative to demands placed on them, and a central aim is to maximise well-being and quality of life for people with ID, which may require investment in additional resources. A good understanding of value of the sacrifices foregone to various sectors and how these costs are derived can also be useful for researchers and analysts alike as they illustrate the economic impacts of ID.

This chapter aims to provide an understanding of each of the main stages in the calculation of costs of services and support in ID research. Although a distinction can be made between the costs incurred to meet the needs of people with ID and what is provided, the principles used to derive costs in

both contexts are in the main similar. In the following sections, we first give an overview of the importance of economics in the provision of services for people with ID and the principles appropriate for use in economic studies. We then focus on how particular categories of costs are derived, generated from information on the services and support typically used by people with ID.

36.2 Economics of Intellectual Disability Service Provision

Economics is the study of scarcity and explores how choices are structured and prioritised to maximise welfare within budgetary constraints [11]. Economics plays an important role in the provision of care and support in intellectual disabilities given that scarcity has two sides; the high and growing levels of wants for most services and treatments; and a limited budget that can go towards staffing, therapeutic services, treatments and support to meet needs. The simple reason people demand these services and treatments is not only because they want to be healthy but because they want to live the lives that many people without a developmental disability take for granted. It is that desire to fully participate in their communities unconstrained by poor health that has led to an increase in the demand for care. There are a few more specific reasons for the increase in the demand for treatment and services such as population changes, changing expectations and government legislation we will cover in the sections that follow.

36.2.1 Population Changes

Changes in the number of people with an ID [12] have increased the demand for services. The increases are explained in part by an increase in life expectancy, especially among people with Down's syndrome. Other explanations for the increase have been related to natural increases in the population. As the population increases so too will the number of economically dependent individuals with an ID, largely due to the decreased mortality and increased life expectancy. Also, as migrant populations increase, the proportion of young adults from minority ethnic communities with an ID is likely to increase, as greater numbers of young people with severe and complex intellectual disabilities are surviving into adulthood [13]. These changes are also contributing to increases in the number of people with an ID in contact with services.

36.2.2 Changing Expectations

Families of people with intellectual disabilities have expectations of what services their loved ones should receive. More and more the expectation is that children will leave home when they reach adulthood and will live as independently as possible in the community instead of at home with their family.

36.2.3 Legislation

In some countries, government legislation has fuelled the increase in the demand for certain types of services for people with intellectual disabilities. Residential services have been on the rise due to legislation in the United States (dating from the 1960s, CMHA; P.L. 88–164 and 1980s, OBRA; P.L. 100–203) and in England and Wales, the Percy Commission, 1954–1957 recommended that more provision be made for people with intellectual disabilities in residential homes in the community, in part in an attempt to alleviate the strain on hospital provision. A series of inquiries (Committee of Inquiry at Farleigh Hospital 1971; Committee of Inquiry into Whittingham Hospital 1972; Committee of Inquiry into South Ockendon Hospital 1974; Committee of Inquiry on Normansfield Hospital, 1978) [14] were conducted following revelations of ill-treatment and neglect and squalid conditions which led to the Government White Paper [15]. This document reinforced the ambition of providing services in the community for people with mild or moderate intellectual disabilities.

In the United States, the number of people with an intellectual disability receiving residential services increased from 267,682 to 460,597 over 1988 to 2011, while over the same period the number of individuals with ID living in nursing homes and state-provided institutions steadily declined [16]. In England, the number of people living in institutions declined from 51,000 in 1976 to over 3500 in 2002 [17]. Concurrently, there was an increase in the number of people supported in smaller community-based residential provision based on the use of domestic accommodation. The movement to community-based settings, mainly hostels, semi-supported group homes, family placement schemes, bed and breakfast and independent living, primarily focused on people with least severe disabilities. Attention has since moved to the development of community-based housing for people with more severe and profound intellectual disabilities and the provision of health and social care treatment and support for carers.

36.2.4 Resources

The other side of scarcity relates to the finite nature of resources. The term 'resources' covers all inputs used to produce treatment and services. When we consider the provision of ID care at any one time, there is only so much funding available for health- and social-related care for those with typical and complex needs. There are also limited amounts of funding for individuals through benefits to provide mobility and disability allowances. Budgetary constraints impose limits on the provision of care irrespective of the sources of funding (see Table 36.1).

Table 36.1 Funding sources in selected European countries

Country	Funding source
Netherlands and Greece	Central government funded in the main
Spain and Belgium	Social administration on a regional level with some funding from charities, private sources and lottery sources
Sweden	Funded through taxation on a local, regional and national level. Social administration on the local level
Germany	Mixed system. Regional level funding for residential services. Social administration at local level and by care insurance or other social provisions, supported living funded
United Kingdom	Mixed system. Central (regional) level funding from the National Health Service (NHS) for health-related care and for those with more complex needs. There is also funding for individuals through benefit payments such as mobility and disability allowances. Local level, funding is provided through social services with some provision from housing services

Source: European Intellectual Disability Research Network [18]

Fixed budgets, demand and supply pressures impose constraints which limit the provision of care. Costing information is often used to assist decision-makers with evidence on the scale of the problem to better determine how to allocate scarce resources. Although limited because it does not consider outcomes, costing information is used to make a strong case to increase investment in intellectual disability service provision. Given the relevance for decision-making, it is important that researchers and practitioners are familiar with the need for determining the range of costs and components to include in any costing exercise.

36.3 Efficiency Versus Equity

The problem of scarcity presents challenging decisions about how best to combine the limited resources in such a way that can achieve

good outcomes. Health economics provides a theoretical and analytical approach to dealing with this concern within health systems while considering the economic principles of efficiency and equity. There are other principles used in health economic evaluations and we will consider these as we go through this chapter. Moreover, there are differing views on what is equitable and it seems reasonable that an equitable allocation of resources would mean that those who have the greatest needs should receive the most support. Equity therefore relates to the extent to which access to services and the outcomes from these services are distributed in a fair way across socioeconomic groups, elderly and the young and by regions. Economic efficiency, on the other hand, implies that society makes choices based on whether the benefits of service can be maximised for the available resources. The aim of the efficiency criterion is to ensure that the health of all those in society cannot be increased if resources are reallocated but makes no judgement about which members of society benefit from this increase. We include them here as they are important considerations in economics and particularly relevant in value for money discussions, though we do not discuss the implications for costing.

36.4 Financial and Economic Costs

Financial costs are the monetary payments associated with the price of a good or service traded in the marketplace. More specifically, it covers the expenses that a company incurs through operations, from factory costs to surcharges. Examples of financial costs include the cost of raw materials, semi-finished and completely finished products, administrative expenses, such as rent, salaries, insurance and utilities. For an economist, the notion of costs is not restricted to money but reflects the value of the gains given up by alternative options when one particular option is chosen. It is the value of the gains foregone or the losses that are incurred when a service or treatment is introduced that is key to understanding costs from an economic point of view.

36.5 Opportunity Costs

Another useful concept is that of opportunity costs. In economics, the gains foregone or the losses that are incurred when a service or treatment is introduced are referred to as 'opportunity costs'. For example, if Liz decides to stop working and go to school to train as a speech and language therapist, the opportunity cost of this decision is the lost wages Liz would have earned had she continued working. In intellectual disabilities economic research, the opportunity costs principle is often used to attach a monetary value to resources. One type of resource that features prominently in ID research is caregiver inputs. Family members or friends may be full-time carers depending on the severity of the ID. In other cases, caregivers may take the individual to daytime activities. The time a friend or family member spends giving support is often provided without any payment as a consequence and arguably so, unpaid caregivers often therefore forego the alternative use of their time such as taking up employment, education or career opportunities, as a result of spending time in caregiving activities. It is not always an easy task to identify what types of opportunities have been foregone to provide care. We revisit the application of this concept in more detail later in the chapter (see ► Sect. 36.8.4).

36.6 Perspective

The perspective is the point of view you use to examine the range of costs to include in the economic analysis. It is an important consideration as intellectual disabilities and comorbid mental disorders have wide-ranging impacts on the person with an ID, their caregivers, services and society. It is therefore important to be explicit about the perspective as this forms the basis for the types of costs to

be included in the costing. There are a number of possible perspectives to choose from including the following:

- People with intellectual disability
- Caregivers (family or friends)
- Health and social care providers
- Public sector providers (could include one or a combination of providers within health and social care, criminal justice or education sector)
- Third-party payers
- Societal perspective

If a given perspective is adopted, only the costs that are incurred by that perspective are considered. In England, the National Institute for Health and Care Excellence (NICE) guidelines currently advise that only National Health Services/Personal Social Service (NHS/PSS) costs be included for approval of new health technologies [19]. If a public sector perspective is adopted, only costs borne by the public sector agencies such as a health, social care, criminal justice would be included in the analysis. From a third-party payer perspective, only the resources paid for by insurance companies, managed care organisations are included. For example, any reimbursements to patients for income loss are an actual cost to the third-party payer. A societal perspective can include, but is not limited to, costs to public sector agencies, caregiver time costs and out-of-pocket expenses, service user out-of-pocket expenses and caregiver productivity losses.

Losses in the production of goods and services—productivity losses—feature in economic studies, and occur when individuals take time off work (paid and unpaid), become unemployed or take early retirement due to disability, or have reduced productivity at work [20]. Productivity losses have not been explored intensively in ID research, perhaps because a small number of people with ID will be in paid or unpaid employment. It is important to note that losses in productivity can also arise from caregivers taking time off for work to provide care. At the extreme, caregivers may be required to give up work all together to provide support or not take up employment in cases where intensive support for a young person or an adult with an ID may be required.

Reference is often made to a narrow or a broad perspective. A narrow perspective focuses on any one of the possible perspectives mentioned previously, for example, the perspective of health and social care providers or third-party payers. In countries with insurance-based systems, the majority of published economic studies involving people with intellectual disability use health claims/expenditure data, which do not capture the far-reaching impacts of intellectual disabilities which fall on non-healthcare sectors. These studies can be considered to have used a narrow perspective. A societal perspective is the broadest of all perspectives. Choosing any one perspective does not preclude using another. The results can be presented from each perspective and are particularly useful to satisfy the needs of different stakeholders.

Underpinning the costing of care and support in intellectual disabilities research are important concepts such as efficiency, equity, perspective and opportunity cost. Each of these concepts requires consideration, though they are not the only concepts which can be considered. We focus on these as they are the most widely used and applied in costing of care and support in intellectual disability research.

36.7 Steps in Cost Estimation

There are three necessary steps when estimating costs in ID research: identification, measurement and valuation. These steps can be done sequentially and will interact before costs are estimated by taking the quantity of each resource item and multiplying it by monetary *units* such as euros and dollars. The first step is to identify cost categories but these are driven by the perspective of the analysis. It is also useful to be aware of the range of resources and support costs that may be relevant to users of intellectual and developmental disability services (see ► Box 36.1).

Box 36.1 Services and support costs relevant to people with intellectual disabilities

- Accommodation costs (e.g. group homes)
- Training costs (e.g. trainers, advocates, managers)
- Medication costs (e.g. neuroleptics)
- Health and social care service costs (e.g. primary care, inpatient stays, community psychiatrist, social worker services, specialist intellectual disability services, community nurse, support worker)
- Education and training costs (e.g. adult education)
- Education facility costs (e.g. schools for children and young people with intellectual disabilities)
- Respite service costs (e.g. respite care)
- Private-sector services (e.g. home-care worker, alternative therapies)
- Person with ID costs (e.g. out-of-pocket expenses for travel to services)
- Unpaid caregiver expenses and costs (e.g. out-of-pocket expenses for child minding, travel; time spent providing support)
- Unpaid caregiver productivity costs (e.g. time off work)

Considering the range of resources and support, inconsistencies in the interpretation, scope and definition of direct and indirect costs, and how costs have been categorised and applied across a number of economic studies in intellectual disabilities; we categorise costs into intervention and non-intervention costs to describe the cost estimation process.

Intervention costs are associated with treatments, programmes or strategies designed to change behaviour or improve health and well-being and in ID research can be usefully sub-divided into facility-specific and non-facility specific for costing purposes. Facility-specific interventions can include residential services or settings where people with an ID access specialist care and support related to their ID or comorbid conditions, for example, an intensive support service, supported housing. Non-facility-based interventions may include medication or treatment not tied to a facility or place of delivery for the person with an ID. Any out-of-pocket expenses or costs to society can also be included here.

The categorisation of interventions, in this way, can also be used to cover interventions aimed at caregivers of people with an ID. Examples of facility-specific interventions aimed at caregivers might include respite care services for caregivers and non-facility-specific interventions could cover caregiver training interventions.

36.7.1 Identification

The identification process considers resources as cost-generating events in physical units and defines appropriate units for measuring resources and support. The units of measurement can be defined as an activity (e.g. blood test, inpatient days, outpatient appointments), physical resources, such as drugs (e.g. tablets), professional time-based inputs (e.g. social worker visits, psychiatrist contact hour, caregiving hours) and societal impacts, such as time off work (e.g. days per week).

The type of resource and issues surrounding practicalities of measurement are useful guiding principles when deciding on a unit of measure. If the unit of measure requires a complex and resource-intensive recording system, it may not be practical or efficient to measure the resource in those units. It is also necessary to take into account whether unit costs can be readily defined or estimated; otherwise, another unit of measure would need to be identified as informed by available unit cost data.

36.7.2 Measurement

There are two main ways to measure resources used by people with intellectual disabilities and the support they receive from caregiv-

ers; these are through the use of questionnaires and diaries. The Client Service Receipt Inventory (CSRI) [21, 22] is one of the most widely used questionnaires to collect information on service use and support in this population. This questionnaire was developed as part of the community demonstration programme 1984/1988 funded to develop community alternatives to hospital care for people with mental health problems, physical and intellectual disabilities, and was first piloted in people with intellectual disabilities. Since these pilots, the CSRI continues to be modified for use in economic studies. It has gone through a number of iterations having been used in well over 200 economic studies to date across a wide range of diagnostic areas including intellectual disabilities and autism, see, for example, [8, 9, 23–26]. The CSRI's seven main sections can be adapted depending on the scope of the economic study. Sections record client demographic information related to gender, marital status, ethnicity; accommodation—a major component in the provision and costs of care in the community; employment and income; daytime activities; service receipt—a core part of the CSRI which identifies health and social care services and specialist mental health services that are not funded by the accommodation budget; aids and adaptations; and unpaid caregiver support. Other questionnaires have been designed for use in children and young people with autism: Child and Adolescent Service Use Schedule (CASUS) [27] and the Service Assessment for Children and Adolescents (SACA) [28]. Diaries can also be used to collect data on the activity of staff working directly with the care recipient, but there is a balance to be stuck between precision and cost to implement. Professionals are asked to fill out how they use their time over a defined period, for example, a week, and then state for each period what they were doing, anything else they were doing at the same time, where they were or how they were travelling. Although not used in intellectual disabilities research, this approach can be adapted and used in recording unpaid caregiver inputs, where the time spent in different activities can be variable (see ▶ Sect. 36.8.4.1).

36.7.3 Sources of Resource Use Data

Questionnaires and diaries are used to collect data on resource use and support when economic analyses are conducted alongside intervention studies (e.g. randomised trials, non-randomised trials, matched *controlled trials)*, but there are a number of other sources from which data can be obtained. Sources such as administrative databases, clinical databases, medical records, focus groups and the literature can be particularly useful when populating economic parameters in economic decision analytical models (e.g. decision analysis and Markov chain analysis). However, there is a misleading distinction between the methods of data collection and the study design in which economic modelling can generally use data from trials (obtained from the literature) and economic analyses within a trial can involve economic modelling, particularly where there is the need to explore the impact of the intervention beyond the trial period. In many disorders, it is not possible to fund a study enrolling enough patients for a long enough period to collect the necessary data to establish sustainability of the intervention, which means that the efficacy of the results is limited to the duration of the trial.

Decision analytical modelling is used to estimate costs, outcomes and cost-effectiveness of alternative interventions and programmes. Data from several data sources are synthesised, and statistical techniques are applied with the aim of providing decision-makers with information on which of the alternatives should be adopted. Decision analytical modelling is growing in popularity in mental health and is already widely used in physical health research. Despite the flexibility it provides analysts and decision-makers, we have only been able to identify one published economic study to date that has employed this approach in the field of intellectual disabilities [29].

36.7.4 Valuation

In the valuation step, the aim is to determine the value for each resource which reflects the opportunity cost of an appropriate unit. For example, during the valuation stage, the analyst seeks to value a hospital inpatient stay, time GP spends during a visit or home care hours. The unit cost for a hospital stay, GP visit or time spent in home care by a home care assistant, for example, would be needed.

There are two main costing methods that can be used to derive unit costs: bottom-up or top-down costing methods. Bottom-up costing methods typically sum information on capital and overheads, salary, and salary on costs divided by the total number of units for the year. When this approach is used to derive, a cost per hour for a professional, salaries and the cost of shared resources such as overheads, buildings, utilities and equipment for the year are added together and the total number of hours is adjusted for travel to provide care or where pay affects by absences allowances would need to be made for annual and statutory leave days (see ► Sect. 36.8.3.1).

Unit costs also may be obtained from publications and national unit cost databases. In the United Kingdom, each year since 2001, the Personal Social Services Research Unit (PSSRU) has published annual estimates of the unit costs of health and personal social services in the community or other settings for people with an intellectual disability, autism and mental illnesses. This widely used national compendium derives costs using the principles outlined in this chapter and is recognised internationally as an authoritative source for unit costs in economic studies.

The values of services based on the prices offered in markets free of government involvement are good measures of the unit cost of a service as they represent the true opportunity cost of resources. Moreover, in countries where the government provides health care such as the United Kingdom or Sweden, the use of market prices will not represent opportunity costs. The same is true for tariffs. Tariffs are prices set by the government or an insurer for payment to healthcare providers. These prices may be set to include incentives for a level of provision. Prices may be set high to encourage provision or low tariffs to discourage it [30]. In some countries such as the United States, the charges from providers to insurers are used to value service use and, in many cases, these charges are set higher than the opportunity costs to subsidise the cost of other activities within the hospital.

A top-down approach takes the total budget and divides it by the unit of measure. For example, to estimate the cost per hour of therapy using a top-down method, the total therapy budget would be divided by the number of hours of therapy delivered. This approach is particularly useful when for some services it is difficult to determine a cost per unit of measure due to complexities around payment arrangements [21].

There are three basic steps conducted in any costing exercise in ID research: identification, measurement and valuation. The Client Service Receipt Inventory (CSRI) was one of the first service use questionnaires to be used to measure services and support in service users with an intellectual disability. The CSRI was developed as part of the community demonstration programme 1984/1988 funded to develop community alternatives to hospital care.

36.8 Costing Intellectual Disability Services and Support

We can apply the stages seen in (► Sect. 36.7) when resources are used to provide training programmes, accommodation, professional services to meet the needs of people with intellectual disabilities and when costing caregiver time inputs.

36.8.1 Training

Training programmes have been used as a way to build capacity in staff but also in families, friends, paid support and front-line staff to put individuals with an intellectual disability

at the centre of matters related to their personal, social and economic well-being. The approach to costing training programmes applies the three stages but includes a preliminary stage—description of the programme. The training programme would need to be described with a view to identifying the resources consumed regardless of whether they can be measured or not; then quantified into physical units such as per person, per hour or per session; and valued by applying a cost to each different type of resource identified. The resource costs across the training sessions would then be derived by summing the costs across all sites (in a multisite study) and weighting it by the number of trainees to derive a cost per trainee.

Usually, when training costs are calculated, they are allocated according to either the number of people assigned to the group or the number of people attending a group. The former approach is more commonly taken because group sessions will usually proceed unless no one attends. It has also been argued [31] that this is the most appropriate approach for estimating participant-level costs, with the exception of programmes with limited places where dropouts are likely to be replaced from a waiting list.

When conducting training programmes not only for the benefit of the participants but also for the benefit of a third party, there are additional concerns that may arise. In this case, using data on the number of participants trained to allocate the cost of the training programme would not provide a link between the cost of training and the individuals the programme is directed towards—people with intellectual disabilities. To explore this relationship, the training costs could be allocated to the number of participants in the training programme or the number of participants who achieved the outcome for which the training was conducted instead of the actual number of people trained. If the number of participants in the training programme is greater than the number of participants who achieved the desired outcome from the training, then the cost per person is likely to be on the conservative side.

► Study example 1—Training

Robertson et al. [32] and Romeo et al. [33] illustrate how a cost per participant can be estimated in a training programme used to support people with intellectual disabilities. Training across the sites was undertaken in a variety of settings; in some cases, a nominal rent was paid for the facility and in other areas use of the facility was provided 'free' of charge. To ensure that capital was consistently employed across all sites, standard capital cost estimates were used. Capital costs were estimated using new build and land requirements for a local authority training room, annuitised at a rate of 3.5 percent over 60 years. To calculate the cost per trainee of attending training, the average costs of each of the elements were summed to give an average cost for the training programme.

36.8.2 Accommodation

Care for people with ID includes some combination of housing provision and services and support that are coordinated by providers external to the residential setting. The type of residential provision is very important for a person with ID as they may need varying levels of support given challenges remembering information, grasping the concept of time, managing self-care and related activities, and carrying out daily life skills. Many individuals live in a house or flat with relatives, or in supported housing or residential care homes. People who can live more independently and do not require any support may choose to live in their own home, rent from private landlords, council or housing associations. When attaching a cost to rented accommodation convention indicates that the rent (fee charged to the service user) can be used as a proxy, this fee is set at an amount that covers both capital and revenue covering the cost of the original capital investment [21]. Similarly, when costing private sector residential or nursing home provision, the fee or charge for boarding and care can be a practical option.

There are various forms of supported housing arrangements: adult placement, sheltered housing, group homes and hostels. There are

instances where people with an ID in supported settings may be charged rent and required to cover additional costs, for example, service charge for communal area maintenance and a charge for any support. Perhaps, the most flexible housing arrangement provided by individuals unrelated to the service user is adult placement schemes. Adult placement schemes involve the individual with an ID living in the home of their landlord or landlady, who also provides them with care and support. There are examples of placement services in the United Kingdom that charge a placement fee that covers rent paid directly to the carer, usually out of the benefits the client receives. There can be an additional fixed payment for food and utilities, agreed upon by the individual with ID and the carer [34].

Residential care can be provided in a registered care home managed and run by a service provider who is responsible for all aspects of the client's daily needs and well-being. The main groups of service elements that make up residential care and the information required to derive costs are detailed in Beecham [21] (adapted in ◘ Table 36.2). The cost can be categorised into establishment costs and costs of services provided by external services. Establishment costs are built up from capital costs, revenue costs and other personal living expenses for items such as food, utilities, personal care and leisure.

When calculating the capital cost of public sector services, the annuity generated by the valuation of the facility over the useful life of the building represents the annual opportunity cost of the capital investment in that facility. In order to derive the annual cost of the capital information on the rate of return or rate of interest on public sector investments, the building valuation and the useful life of the facility commonly estimated at 60 years is required. Revenue costs can be obtained from income and expenditure account held by the finance department and produced annually. There are challenges when using this approach to calculate establishment costs for private sector services as buildings' valuations in the private sector may not be readily available. In this case, the fee or charge to the resident can be used as a proxy for capital and revenue elements.

◘ **Table 36.2** Costing residential care

Elements	Sources
Capital:	
Building, location and size	Capital costs for building and land can be calculated using market valuations of property. Capital costs would be annuitised over 60 years
Revenue:	
Staffing (direct and non-direct staffing)	Staff-related costs and expenditure accounts
On-site administration	Expenditure accounts
Agency overheads	
Other costs:	
Personal living expenses for items such as food, utilities, personal care and leisure	Expenditure accounts if provided by the service. Personal allowances/benefits can be used as proxies for personal consumption if these expenses are not covered by the service

▶ Study example 2—Accommodation costs

Felce et al. [35] estimated and compared both the costs and quality of life outcomes of adults with intellectual disabilities living in semi-independent accommodation to similar individuals living in fully staffed group homes. Costs of accommodation were estimated from agency financial accounts that managed and provided room, board and support for each individual participant. Accommodation costs included the costs of direct and non-direct staffing, administration (on-site) and central agency overheads. Costs of non-accommodation included the costs of daytime services, hospital and community-based services and were estimated from a service use questionnaire. To calculate the total weekly cost of the accommodation

service packages, the average costs of accommodation and non-accommodation costs were summed to provide an estimate for the average total weekly costs of semi-independent living and fully staffed group homes.

36.8.3 Non-accommodation Service Costs

Services and support provided external to the setting where people with intellectual disabilities live can be delivered in a healthcare system with many agencies providing a variety of services. Often there is no basic package of care offered to people with intellectual disabilities and services such as health, social care and specialist mental health services which may not be funded within the budget of the housing setting. Typical services provided external to the living setting may include but are not limited to hospital-based inpatient and outpatient services or speech and language therapy and GP services. Support services provided by a home care assistant to help with personal care, washing up, dressing, housekeeping and other domestic work or services, such as meals-on-wheels, may also be available. In some countries such as Denmark, the government provides home help which may also include some physical exercise and mental stimulation of the individual to help maintain physical and mental capabilities. Currently, in England, local authorities have the powers to make direct cash payments to individuals with intellectual disabilities, who can then arrange and pay for their own care. Families and friends can be paid out of these payments for the care they provide, once they are not living at the same address.

36.8.3.1 Costing Non-accommodation Services

There are different methods that can be used when costing the time spent by professionals who visit individuals where they live or are visited by people with an ID in a clinical setting. The bottom-up costing methods can be used to calculate a cost per hour of contact with a professional. The professional's annual costs are aggregated using elements related to the profession, grade, working hours; capital based on location size of the office or clinic; overhead costs to the provider for administration and management as well as office, training and utilities and general management and support services such as finance and human resources. For each of these elements, information on salary-related costs, salary scales, national insurance, superannuation rates; capital, overhead costs to the provider for administration and management, office, training and utilities; overheads such as general management; and support services such as finance and human resource departments are summed and then divided by the total number of hours worked for the year based on the professional's conditions of service. If the total number of hours includes annual and statutory leave, it is assumed that pay is not affected by absences. When pay is affected by absences, the total number of hours would need to be adjusted accordingly. For professionals who make home visits, the denominator—the total number of hours—will be substituted by the average number of domiciliary visits per week. ◘ Table 36.3 illustrates the valuation of an hour of time for a community occupation therapist.

36.8.4 Caregiver Inputs

Often tasks provided by family members for which no payment is received (referred to as unpaid caregivers) can also be performed by paid caregivers. For simplicity, most economic analyses, make no statements about the equivalence of the quality and efficiency of the care provided by paid and unpaid carers. In these analyses, the implicit assumption is that the care provided by both paid and unpaid caregivers are equivalent in both respects, although there may be marked differences [36].

Despite the importance of unpaid caregiver support to the individual and impacts on the care recipient's outcome, there remains a paucity of studies that have included caregiving support in economic analyses in intellectual disabilities. Our own review of published articles covering or including economic analy-

Table 36.3 Illustration of community occupational therapist

Element	
A. Salary/wages	£32,419
B. Salary on costs	£8873
C. Overheads	£18, 582
Working time	40.9 weeks per year; 37 hours per week
Travel time	£2.80 per visit
Ratio of direct to indirect time:	
Face to face	1:0.67
Clinic contacts	1:0.33
Home visits	1:0.73
Length of contact	60 minutes per home visit
Unit cost:	
£40[a]	Per hour
£53[b]	Per hour in clinic
£71[c]	Per home visit

[a](32,419 + 8,873 + 18,582)/(40.9 × 37)
[b][(32,419 + 8,873 + 18,582)/(40.9 × 37)] × 1.33
[c][[(32,419 + 8,873 + 18,582)/(40.9 × 37)] × 1.73] + 2.80

sis of caregiver inputs in intellectual disability up to February 2018 identified no studies that focused solely on the costs of informal care; however, five studies incorporated the cost of care provided by paid or unpaid caregivers into their economic analyses [25, 26, 37–39].

Although public decision-making bodies in some countries may not explicitly recommend the inclusion of unpaid care costs in cost-effective analysis of health and public health interventions, it is important to note that a failure to include unpaid caregiver impacts from costs-effective analyses of health and social care interventions, which could lead to different conclusions about value for money as seen in a study of people with dementia [40]. Careful consideration of the likely impacts of a failure to include unpaid caregiving impacts is required [41]. Tranmer and colleagues [41] take the view that as a greater proportion of care is being provided and received within the home and community settings, the exclusion of these costs could provide a misleading estimate of the true costs of healthcare programmes. It is therefore advisable that any decision to exclude unpaid caregiver costs is justified.

36.8.4.1 Measurement of Caregiver Time

Measurement in this context involves quantifying how much time is used to provide care to the person with an ID by the family member, friends and other individuals such as volunteers. There are three main methods that can be used to collect time use data: interviews, diaries and direct observation. Interviews involve asking respondents to recall retrospectively the time devoted to caregiving activities over a defined time period, for example, average time spent preparing meals in a typical week. Care should be taken when looking at the period of recall. Shorter periods of time are more easily remembered than longer periods. It may be possible to collect more accurate data for the past week than the past year. Interview questions would need to be carefully worded as caregivers may perform more than one activity at the same time (also referred to as joint production) such as preparing meals and socialising with the care recipient. Shearer et al. [42] suggest that concerns with allocating time to each activity can be overcome by wording one of the two activities as the main activity and the others as secondary or combining both activities into one. Another method that is a more accurate way to record time inputs than an interview but can be time-consuming is the use of dairies. Diaries involve the caregiver recording the time spent supporting the care recipient with caring activities. Information can either be recorded over the course of the day or retrospectively. It is less time-consuming than diaries but potentially a greater imposition is the use of direct observation. During direct observation, the observer records as accurately as possible the amount of time the caregiver is spending on various activities. One of the

advantages of this approach is the richness of the data. An observer who has no personal involvement with a caregiver may also be able to gain valuable insights, which a person more familiar with a situation may miss. It also requires less active involvement and cooperation by the caregiver. Despite these advantages, this method is resource intensive requiring a trained observer to perform that can be susceptible to observer bias.

36.8.4.2 Valuation of Caregiving Inputs

Different methods have been used when valuing caregiver time inputs; many have been described as controversial [40]. Three methods considered by Brouwer et al. [20] include market price, replacement costs and opportunity cost.

Caregivers often provide support free of charge, but although there is no money received for the support they provide, caregiving inputs are not free in an economic sense. The time the unpaid caregiver is involved in caring activities is time which could be spent in any of a number of different activities, for example, paid work, unpaid work or leisure. Consequently, then, there is a cost in terms of the opportunities they have foregone. Using this approach, the alternative activities given up as a result of the time devoted to caring can be identified and valued. If the time spent caring is at the expense of paid work, then time inputs would be valued by wages. Similarly, for unpaid work, time could be valued by estimating the wage paid to someone in the paid care sector who would be employed to provide the same activities. For example, wage rates for home care assistants can be used to value time spent providing support with managing money, dressing, mealtimes and shopping. Leisure time valuation is particularly tricky as there are no widely used methods or conventions when valuing leisure time inputs for unpaid caregivers. Hourly wages as shadow prices have been suggested for valuing leisure time by caregivers [43].

The replacement valuation method is based on the cost of buying the same number of hours of unpaid care from the formal care sector. In the replacement cost method, the wages for paid caregiving support would be used to value time spent providing unpaid support. However, replacement cost valuation methods may not be different from the opportunity cost approach, in which the unpaid caregivers could theoretically take on paid caring roles if they were not caring for their family member or friend. The value of the unpaid caregiver's time could be reckoned in terms of the wages of a paid carer [42]. In research conducted by Barron and colleagues in an inner London Borough with adolescents with intellectual disabilities and challenging behaviour between the ages of 16 and 18 years, the authors found that there was a potential saving to local authorities in England of £1554 per week where unpaid caregivers were providing unpaid care and support [37]. In a study of a health check intervention designed specifically for adults over the age of 16 years implemented in the Greater Glasgow Health Board area, Scotland, Romeo and colleagues (2009) applied an hourly rate of a home care work to full time unpaid caregiver inputs to provide a monetary measure of the cost of these time inputs [26]. In the same study, opportunity cost principles were applied where the average gross hourly earning of caregivers in full time or part time paid employment was used to value the unpaid care giving inputs by those caregivers to service users. In research where unpaid care was provided by students, caregiving and support was valued at the minimum hourly wage and an average hourly pre-retirement wage for retired caregivers [25, 38, 39].

Other methods of valuing caregiver time can be used which reflect the preferences or the loss in well-being experienced. Preference-based methods such as contingent valuation and conjoint measurement can also be applied to value unpaid care. Both of these valuation methods measure from the caregivers themselves how much they feel they should be paid. In the case of contingent valuation techniques, the caregiver is asked how much they are willing to accept (WTA) to provide care for an additional hour [44] and in the case of conjoint analysis, trade-offs are made between

the caregivers current caregiving situation and hypothetical caregiving situations [45]. The well-being method considers the psychological impact on the caregiver and is based on the loss in well-being suffered as a result of caregiving activities. Using this method, the value of unpaid time inputs is estimated as the amount of income required by the unpaid caregiver to maintain the same level of well-being after an additional hour of care is provided [46, 47].

Tip

The three main steps in costing estimation exercises have tended to be used when estimating costs for training programmes, accommodation and professional services offered to people with intellectual disabilities. It can also be used to cost caregiver support provided to service users.

Key Points

- Estimating costs is necessary and important as it is often the economic consequences that influence policy responses even though the health and quality of life impacts of intellectual disabilities are usually the focus of public concerns.
- The main steps in the estimation of costs are identification, measurement and valuation of resources and support to people with intellectual disabilities. The estimation process mainly applies to studies where data can be collected as part of a trial or observational study.
- The majority of studies in intellectual disabilities to date are trials or observational research which do not explore the economic impacts beyond the study period. These limitations can be overcome through the use of decision analytical modelling where data can be obtained from the literature, large administrative datasets and focus groups.

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Residential Care and Community Living

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Contents

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Learning Objectives

The aim of this chapter is to give the reader:

- An awareness of how residential and community-based services for people with intellectual disability have changed over time.
- An awareness of the current living situation of people with intellectual disability.
- An understanding of the importance of outcomes in thinking about the quality of services.
- An understanding that although an ordinary home dispersed in the community is a necessary condition for better quality of life outcomes, it is not sufficient on its own.
- An understanding of the importance of paying attention to structures and processes, in particular, the nature and quality of the direct support provided.

37.1 Introduction

Over the past decade and a half, there has been a growing recognition internationally that being an active participant in society is about fundamental rights and that governments should provide the necessary support to enable all people to enjoy these rights, including persons with disabilities [1, 2]. The move from institutional to community-based support for all persons with disabilities is an important part of this process [2, 3].

In 2006, the United Nations adopted the Convention on the Rights of Persons with Disabilities [4] that since then has been ratified by the majority of governments across the world. The Convention marks a global paradigm shift in the recognition of disability as a predominantly medical/clinical to a human rights and social issue [5, 6]. The treaty calls on all states to take "effective and appropriate measures" to facilitate the full inclusion and participation of people with disabilities in the society, including respect for a person's autonomy and independence (Article 3). It also requires that States ensure "the equal right of all persons with disabilities to live in the community, with choices equal to others" and "have access to a range of in home, residential and other community support services, including personal assistance to support living and inclusion in the community" (Article 19).

This chapter will provide and discuss the definitions of community living and residential care as concepts as well as different models of each. It will then explore some of the historical contexts: "deinstitutionalisation", the drivers and lessons from the process so far in the United Kingdom and internationally. Third, it will focus on the evidence base for community living and residential care in terms of outcomes for people with intellectual disabilities and what is needed for good quality of life outcomes. This chapter will primarily draw on the already published literature, but it will also draw on experience as a practitioner and consultant, working with colleagues and organisations to bring about improved outcomes for people they support (see Acknowledgements).

37.2 What Is "Residential Care"?

Residential care is an arrangement where the person is provided with both accommodation and support by the same provider, where the two are linked, whereas community living is associated with the idea of "independent living", which does not mean unsupported functional ability but choice and control over daily living arrangements and support. People can live independently in the community with formal (paid) support, with informal or no support, or a combination of the two. In the past residential care was synonymous with large size and institutions but as a result of deinstitutionalisation, models of residential care have become more diverse. Meanwhile, the term institution/institutional is increasingly used to describe the structural characteristics of services, marked by four key features: "depersonalisation" (the removal of personal

possessions, signs and symbols of individuality and humanity), "rigidity of routine" (fixed timetables irrespective of personal preferences or needs), "block treatment" (processing people in groups without privacy or individuality) and "social distance" (symbolising the different statuses of staff and residents) [7, 8]. Nonetheless, there was, and still remains, a clear relationship between such institutional care practices and the size of settings, which Mansell and Beadle-Brown [9] argue is not coincidental.

There has been no research that has provided evidence that institutions can achieve the same outcomes for people as good community-based services [10–12]. It has also been recognised that the same institutional practices and ways of thinking can be—commonly—found in services in the community, even in very small settings [13, 14].

This has led to definitions of institutions such as that used by People First of Canada: "An institution is any place in which people who have been labelled as having an intellectual disability are isolated, segregated and/or congregated. An institution is any place where people do not have or are not allowed to exercise, control over their lives and their day-to-day decisions. An institution is not defined merely by its size."

1. People with intellectual disability have a right to live in the community, with choice over their living situation, on an equal basis with others and with support to be active participants in society.
2. Residential care is an arrangement whereby people receive accommodation and support together.

Tip

We need a nuanced view of accommodation and support options—it is not as simple as residential care equals bad and supported living equals good.

37.3 Deinstitutionalisation

37.3.1 What Is It?

Deinstitutionalisation is the replacement of institutional services with community-based support, and it has been described as one of the most important changes social care in the last decades [15]. It is important to note that deinstitutionalisation is not just about the closure of hospitals or large residential care services but about the development of services in the community that both prevent people from being admitted to institutional services in the first place and provide a place for people to move to from institutions. For a successful transition, the services that need to be developed are not just accommodation and support services, but transport, health care, social care, education and other public facilities all need to be made accessible and skilled enough to provide services for people with disabilities, if people are to be truly integrated into the community.

37.3.2 What Drives the Process?

Mansell and Ericsson [16] noted four key drivers of deinstitutionalisation in countries which started the process early. Although the exact process has differed in different countries, these four factors have consistently remained important. These drivers included (1) changes in *ideology* (e.g. the rise of normalisation, social role valorisation and an Ordinary Life [17–19]; (2) the availability of *alternative models* often led by professionals, especially for those with more complex needs [20–22]; (3) revelations in the media of *scandals* in institutions and public opinion (this has continued to be important in many countries, with recent scandals such as the Winterbourne View in the United Kingdom); and (4) the balance of *costs*, that is, whether community care was seen as less expensive, as in the United States, better value for money, as in the United Kingdom, or the right thing to do and therefore cost was not seen an issue, as in Sweden [16, 23, 24].

37.3.3 Deinstitutionalisation of Services for People with Intellectual Disability: The Example of the United Kingdom

Trajectories of institutionalisation and deinstitutionalisation differed across the four nations of the United Kingdom with many similarities and some notable differences. Devolution has further promoted diversity in the provision of social services, including for adults with intellectual disabilities. We attempt to highlight some of these below.

By the Second World War, large residential institutions, often built in the nineteenth century, were the typical form of provision for people with intellectual disabilities, people with mental health problems, orphans and the homeless in the United Kingdom [3].

Institutions for people with intellectual disabilities and those with mental health conditions were transferred as hospitals to the newly created National Health Service after 1948. However, it was in the late 1960s when a series of scandals involving ill-treatment, overcrowding, understaffing, poor environments and various indications of management failure in hospitals came to media attention that things began to change.

In 1971, the white paper "Better services for people with mental handicap" [25] proposed a reduction in institutional places through the provision of more places in "small" residential homes provided outside long-stay hospitals.

During the 1980s and 1990s, substantial progress was made towards the development of community-based supports and the closure of the long-stay hospitals for people with disabilities in England and Wales [20]. This happened without the government clearly articulating the goal of closing institutions and a "firm legal or constitutional basis for non-institutional services", which led to "trans-institutionalisation" (the move from one type of institution to another) for many people [3]. In more recent years, this same lack of clear guidance, policy and legal basis has resulted in new institutions being built, in particular, for people with specific needs such as challenging behaviour or autism. The size of community-based services has never been specified in terms of policy or legislation in the United Kingdom (unlike in some other countries such as Ireland or Norway), although NICE guidance on the treatment and management of adults with autism has recommended no more than six people on one site [26]. Some local authorities are beginning to stipulate the size of services they will commission but in general there remains no legal or financial barrier to setting up large, congregate and clustered services.

In Scotland and Northern Ireland, progress with deinstitutionalisation was initially slower, however, gathered pace in more recent years [27, 28].

Alongside the largely professionally led replacement of institutions with community-based services, the disability movement began to develop the idea of independent living. This was an alternative model of support based not on buildings and services but on the provision of personal assistance, through the provision of direct payments [29], and later personal social care, health and education budgets can be made in lieu of services, so that people could make and control their own arrangements. The emphasis on individualised funding mechanisms has not been without criticism, in particular, in terms of implementation and how much choice people really had with a personal budget [30–32], but the government has continued to promote individualised funding and services for everyone with disabilities [33–35].

1. Deinstitutionalisation is the closure of institutions and the development of housing and community-based support.
2. Key drivers of change have included ideology, cost incentives and disincentives, scandals, and the available of alternatives.
3. The power of each driver has varied by country.

37.4 The Paradigm of Community Living

One of the first definitions of "community living" came from the Ordinary Life Initiative in the United Kingdom [17] which referred to community living as

- Local housing with support for people of all levels of disability.
- Work and work experience as an alternative to day care.
- Widening participation of people with all levels of disability in community life.
- Supporting people with challenging behaviour in the community.

More recently, Mansell [36] and Mansell and Beadle-Brown [9] pulled together these and other pre-existing definitions from a range of sources to offer the following definition of community living:

> The aim of community living is to enable people with disabilities to use the same range of accommodation, living arrangements and patterns of living that are available to the rest of the population, and to have a good quality of life, participating as full citizens in social, cultural and economic activities to the extent and in the ways the individual chooses. ([9], page p. 106).

This requires that those providing services and support are

- Using accommodation located among the rest of the population, which is adequate, appropriate and accessible to the individual.
- Using the range of accommodation options ordinarily available to the wider population.
- Enabling people, to the greatest extent possible, to choose where, with whom and how they live.
- Providing whatever help is required to enable people to participate successfully in the community.

This latter point means that in order for community living to be successfully implemented there has to be attention paid not just to housing and support for daily living but to the willingness and capacity of communities (including generic services) to welcome and support people with intellectual disabilities. Attention must be given to accessibility of leisure facilities, work environments, health and social care environments in the community, opportunities for social connectedness, etc.

37.5 Types of Living Arrangements Used by People with Intellectual Disabilities

In many countries, the majority of adults with intellectual disability still live with their families, although in some countries such as Norway, more people live in their own home with support or in group homes than with their family [37]. In England, in 2014/15, the most common living situation for adults with intellectual disabilities getting long-term social care support was with family/friends [38].

There are also very little data available about the nature of accommodation options for people with intellectual disabilities across Europe [2, 3, 39]. In particular, there is little information about the size and quality of these services. The information that does exist from different sources uses different definitions and often conflates accommodation and support. Ireland is the only country that has a register of people with intellectual disability recording the numbers who access different types of services [3, 27], although Norway has also been developing such a register [40]. Although limited by the lack and completeness of data, Šiška and Beadle-Brown [39] reported that there were at least 1,438,696 children and adults with disabilities and mental health still living in institutions. The number of people in institutions was almost the same as the figure reported by Mansell et al. [3]. At least some residential care was still provided for all groups, even for children without disabilities. Only in Sweden was all residential provision for adults with disabilities, small scale and community-based. In most countries, such small residential services

were a minority form of services. This study also identified that those who had seen the least change were those with intellectual disability and those with mental health problems. Looking in more detail at England, the most recent source of data on accommodation status for people with intellectual disability is from 2015 to 2016 (Community Care Statistics, Social Services Activity—England, LTS004.b).[1] According to this, in 2015–2016, 129,055 people with an intellectual disability aged 18–64 were receiving social services support from local authorities in England. More than 75% (96,285) were reported to be in "settled accommodation" in the community: with tenancy rights either as an individual or as part of a household. The remainder were classed as being in "unsettled accommodation" without tenancy rights, such as registered care homes (22,435), nursing homes (1090) or acute/long-term healthcare residential facility or hospital (470). In addition, the most recent figures, at the time of writing, from the Assuring Transformation data set[2] indicate that at the end of January 2019, there were 2325 people with learning disabilities, autism (or both) placed in inpatient settings by NHS England and Clinical Commissioning Groups. Just over half of people (52%) in hospital were in general (non-secure) settings, 28% were in low-secure settings, and 20% were in medium- or high-secure settings. More than half of people (57%) had been in hospital for more than 2 years and 15% more than 5 years. Although the majority of the population in these units are adults, there are also some children aged under 18 years (11%).

Outside of Europe, there is also generally a lack of data available on living situation of people with intellectual disability in Asia, where most of the research has focused on children and the impact of the transition from institutional care. In Australia, as in Europe, the majority of people with disabilities live in their own home or with their family [41], while those with intellectual disabilities, in particular, those with more severe disabilities, primarily live in residential care settings. Most of these tend to be relatively small (6 or fewer people) although some are larger in size, with 11,000 people (under 65 years of age) living in "cared accommodation" (e.g. hospitals, nursing homes, hostels and other homes with six or more people) and with almost 8000 people with disabilities under 65 identified as living in aged care services [42][3].

In the United States, the situation varies from State to State—with some states only providing community-based settings (e.g. Vermont) but with others still providing residential institutions for a substantial number of people with intellectual disabilities (e.g. Texas). Lulinski, Tanis & Nelis [43] reported that in 2015 more than 21,000 people with intellectual disabilities (18% of all people living outside the family home) across 37 states still lived in congregate accommodation including state institutions. The remaining 82% lived with six or fewer people and in 2015, 13 States had no state-run institutions [43].

In the remainder of this section, I will outline in more detail the different models of residential and community living accommodation and support options in use in the United Kingdom. The boundaries between some of these models have become increasingly blurred over the past 10–15 years, while others have remained relatively unchanged. As such, this section will attempt to describe both the "pure" model where one exists and outline where some of the newer models sit. Where available some indication of the size and numbers of people accessing each different type of service will also be included. Most of these models—sometimes under different names—can be found in other countries in Europe, Australia and North America.

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1 From 2016 to 2017, only expenditure is reported in the Adult Social Care Activity and Finance Report with limited breakdown available.

2 ► http://digital.nhs.uk/catalogue/PUB30227

3 ► https://www.aph.gov.au/Parliamentary_Business/Committees/Senate/Community_Affairs/Young_people_in_aged_care/Report

1. The majority of adults with intellectual disability live with their families.
2. When not living with their family, people with intellectual disabilities are much more likely to be living in large congregate settings (institutions) than people with other types of disabilities.

37.5.1 Congregate Settings

Congregate settings are usually settings where people are accommodated together in larger groups under one site and under one roof. In the United Kingdom, the old learning disability (mental handicap) hospitals fell within this category. These types of provisions have different names in different countries (e.g. institutions, residential hospitals, social care homes). These tend to range from around 30 places (e.g. in France and Ireland) to around 1000 people in some parts of Eastern Europe. The main types of congregate settings still used in the United Kingdom are hospitals, secure/forensic units, and assessment and treatment centres.

■ Hospitals, Secure/Locked Units and Assessment and Treatment Services

Although all the old long-stay hospitals for people with intellectual disability were closed by the mid-2000s in England and Wales, newer hospitals and secure residential settings have grown up over time, primarily to serve the population of people with learning disability who have complex health needs, show challenging behaviour or have forensic support needs. These tend to be run by independent (usually for-profit) organisations or the National Health Service.

Some of these services were designed as long-term secure accommodation options but most were set up as short-term "assessment and treatment" services, many with a focus on challenging behaviour. However, as noted above, almost 60% of people were in these settings for more than 2 years. Although originally people could be detained in such units without a legal basis, recent figures indicate that 90% of people admitted to these inpatient services were subject to the Mental Health Act (MHA), with 51% subject to MHA Part II. Of the 10% who were admitted informally, 92% were in general (non-secure) settings.

■ Transitional Services/Step-Up–Step-Down/Rehabilitation Services

In the early stages of deinstitutionalisation, the concept of transitional services for people to learn the skills needed to live in the community emerged. These were sometimes referred to as "half-way houses" and were often on the site or just outside the gates of the initial institution. For some people, these became permanent homes but for the most part people moved on to smaller group homes or more independent living arrangements.

This option reflected what was known as the "readiness model"—the idea that people had to be "ready" to move into the community. As time went on, this was replaced by what became known as the "support model", in which it was considered possible for anyone and everyone to live in the community with the right support [44].

Recently, we have seen a resurgence in transitional or step-down services (sometimes referred to as rehabilitation units) in the context of people moving from inpatient units as part of the Transforming Care programme in England. The idea that people will need to get to the point where they are "ready" to live in the community has re-emerged. While there may be a small number of people who have to get re-accustomed to certain aspects of living in the community, it is more likely that it is professionals and service providers who need the "safe space" in which to find out how best to support the person in the community. Whatever the situation, such an arrangement should always be short-term, with a clearly defined purpose and services should still conform to best practice. Very little is known about how many such services exist or how many people they provide services for in the United Kingdom. Of the 2465 people recorded in the Assuring Transformation data in January 2018, 18% were living in what was known as forensic rehabilitation or complex care/rehabilitation.

37.5.2 Clustered Settings

Clustered settings are usually settings where people live in smaller self-contained houses or flats but with more than one home grouped on the same site. There are three main types of clustered settings: village communities, campuses and cluster housing.

Intentional and Village Communities

These communities are typically founded on spiritual principles and distinguished by having support workers, who are often unsalaried, and sometimes their families living communally with people with intellectual disability to facilitate close personal relationships, and provide them with a social and cultural framework for work, community service, worship, and education. In some communities, there is a core set of paid staff which is supplemented by volunteers often from overseas. The size of these village communities varies but some have over a hundred residents.

Examples of village/intentional communities include Camphill, Ravenswood or L'Arche. Some of these communities have changed over time and now also providing dispersed, community-based support. Usually, village communities for adults provide accommodation, support and meaningful occupation for between 11 and 30 people.

In the United Kingdom, village communities are registered as residential care and there is no breakdown as to the numbers of people specifically in this type of setting. However, Mansell and Beadle-Brown [12] note that village communities are typically a relatively small part of the total amount of service provision, partly because they rely heavily on volunteers wishing to share their lives with people with disabilities. There are little data available on village or intentional communities in other countries but we know that at the time of writing L'Arche had "more than 154 communities and 19 projects in 38 countries"[4]; there are more than 100 Camphill Communities in 23 countries.[5]

Campuses

Residential campuses were often developed as part of the programme of closure of institutions and were modelled partly on village communities and partly on residential services in the Netherlands and Belgium. Like village communities, they are often self-contained with day provision and other services on-site and in some cases also had schools and colleges on-site. These were often set up by the National Health Service (NHS) for people with severe and profound disabilities, for whom dispersed housing was not considered a viable option at the time. In 2007 the Department of Health responded to evidence of poor quality care and announced the complete closure of all NHS residential campuses [45]. At that time, there were just over 1600 people in NHS campuses in England, living in units of 8–10 people [46]. Just over 10 years later, most of these campuses have now closed.

However, some campuses run by independent organisations still exist in England, registered as residential services, usually alongside more community-based options run by the same organisations. Some of these settings were originally set up by families, usually around a particular activity such as farming, and some developed into campuses from one larger home as the land around the larger home was available for building on.

Recently, a range of services have emerged in the United Kingdom, which call themselves "villages" but they do not have the social, spiritual or cultural framework of the village communities outlined above, and they can be considered campuses.

Cluster Housing

Cluster housing typically consists of a relatively small number of houses on the same site; for example, forming a cul-de-sac in housing for the wider community, or several houses in the same road all accommodating people with disabilities. Cluster housing can

4 ► https://www.larche.org/en/what-we-do

5 ► https://camphillfoundation.org/about-camphill/

operate as a registered residential care service where housing and support are provided as part of a package or can offer supported living arrangements for people (see below). The most recent developments in this area are clustered flats where six or more people have individual or shared flats within a larger development of flats all for people with intellectual disabilities or autism. Some of these developments can be quite big—we know of some as big as 30 people living in flats, sometimes all receiving support from the same provider.

■ Residential Schools and Colleges

As already noted above, some residential schools and colleges are part of a village community or campus-type arrangement. There are others that are standalone schools and colleges run by private or voluntary organisations.

Children who attend residential schools tend to be those with complex health or behavioural issues. A recent study by Tomlinson et al. [47] identified 342 residential educational settings in England with a total of at least 8740 placements available for young people aged 16+ with intellectual disabilities or who are autistic, with the majority of these being full-time and often out of area.

37.5.3 Community-Based Residential Care Homes, Group Homes and Individualised Services

In the first wave of deinstitutionalisation, services conceptualised as community-based were smaller than the old hospitals but still quite large—for example, it was not uncommon for early services to be residential care homes/hostels for between 12 and 20 people. Even now there still remains a substantial number of places in the United Kingdom that support more than 10 people. Later, a more common form of provision was group homes that were initially for around eight people living together and overtime these have reduced in size.

The number of people with learning disabilities in England supported in residential care in 2017–2018 was just over 25,000.[6] The most common type of residential care setting in the United Kingdom has for some time been what is usually called a "group home". In the United Kingdom, most group homes accommodate between three and eight people. The Care Quality Commission (2016) found that services that were smaller than 10 places performed better in terms of inspection standards. Staffing arrangements in group homes vary from a few hours per week of outreach support for specific tasks to 24-hour support. Currently, in the United Kingdom, it is unusual to find people in group homes only receiving drop-in or outreach support—for the most part these people would now be in supported living settings where they rent (or occasionally, own) their own home and support comes to them.

Internationally, there are differences in what is defined as a group home [3, 37]. For example, in Germany, group homes more commonly provide accommodation and support for around 24 people with a mild to moderate intellectual disability. In Sweden, a group home usually consists of six or eight individual flats clustered together with staff facilities and a communal area. In Ireland and Norway, a group home can be no more than four places. In Australia, this type of service might be referenced to as shared living (but this term would also be used if people were renting the property). In Australia, people with much less severe disabilities have lived in group homes until much more recently than in the United Kingdom [11], and people of similar levels of ability (at the upper end) can be found in both group homes and in supported living arrangements [48].

■ Adult Placement/Shared Lives Schemes

Adult placement, or Shared Lives schemes as commonly referred to in the United Kingdom, is used as an alternative to residential care for

6 ► https://digital.nhs.uk/data-and-information/publications/statistical/adult-social-care-activity-and-finance-report/2017-18

adults with a variety of support needs. The disabled person is matched with an approved carer who provides them with accommodation and support in their home, sharing their family and community with the individual.

Shared Lives is a relatively small form of provision in England with around 2–3% of people supported by local authorities using such service (Community Care Statistics, Social Services Activity, 2015–16).

Supported Living

The main tenet of supported living is that housing and support are separate. People live in an ordinary house that they own or rent (the same type of house in which people without disabilities live in that country), giving them security and the rights that come with a tenancy or ownership. Support then comes into their home and depending on their needs it can vary from a few hours a week to 24-hour support.

The main principle behind supported living is the one of choice: people have choice and control over where they live, with whom they live, who supports them and how, and what lifestyle they live [49]. The National Disability Team conceptualised supported living as follows:

- Separating housing and support.
- Focusing on one person at a time.
- Full user choice and control.
- Rejecting no one.
- Focusing on relationships; making use of informal supports and community resources.

In the United Kingdom, there are a series of standards called the REACH Standards for Supported Living (Paradigm, 2002; revised 2013) which identify the following as essential for any arrangement to be supported living[7]:

- I choose who I live with.
- I choose where I live.
- I have my own home.
- I choose how I am supported.
- I choose who supports me.
- I get good support.
- I choose my friends and relationships.
- I choose how to be healthy and safe.
- I choose how I am part of the community.
- I have the same rights and responsibilities as other citizens.
- I get help to make changes in my life.

The litmus test for whether an arrangement is truly supported living is to ask whether the person could decide to move house but keep their current support provider or whether they could stay in that house but choose to be supported by a different support provider. If both of those situations do not apply, then the arrangement is not truly supported living.

Of the 18,845[8] people with learning disabilities in England reported in 2015 by local authorities as having tenancies or ownership, there is no information about how many of those are meeting the REACH standards. While there are many good providers of supported living arrangements, it is generally accepted that there are many arrangements in the United Kingdom that are called "supported living" but which really are not supported living. There are a number of reasons for this.

At the point Supporting People funding[9] became available, many providers and local commissioners saw the possibilities for this

7 Accessed 21/03/18 ► http://qualitycheckers.org.uk/about-aqc/quality-checking-tools/the-reach-standards

8 ► https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/613183/PWLDIE_2015_data_tables_NB090517.pdf, page 14.

9 Supporting People was a government initiative launched in 2003 with the initial aim of supporting people to live independently and help them to remain in their home by providing them funding to maintain their social housing tenancies. However, over time it was broadened to provide housing related support for anyone regardless of housing status. This included private and social tenants, home owners, homeless people and those in temporary accommodation as well as people in sheltered accommodation or other specialist housing. Eventually though the funding stream was stopped as it had cost the government substantially more than ever anticipated.

pot of money to be used for funding people with intellectual disabilities. As a result many providers de-registered existing care homes and, in one way or another, created a tenancy for people and then referred to the service as supported living in order to access the funding. In general, nothing changed for the people who lived there in terms of their support, their staff or their level of choice. Staff also did not see any change—on a project trying to implement person-centred support into services recently de-registered and turned into supported living, staff where asked what is "supported living", "what does it mean to you?", the response was invariably "it is just the new name for residential care".

The second factor that influenced the expansion of supported living arrangements in the United Kingdom was the introduction of the Ordinary Residence principle (published in 1993). Although, a complex principle, in essence, the implication was that when people who had been placed out-of-area moved into supported living arrangements, responsibility for funding that individual shifted from the placing to the receiving authority. This created some perverse incentives for placing authorities to "move people on" to supported living arrangements in the area in which they had lived [36], sometimes without even moving house.

Although not documented particularly well, there has been a recent surge in the development of what I will call *quasi*-supported living. Examples include where people are paying rent to a housing agency that is a branch of the same organisation providing the support; where ownership or management of the property has been transferred to the local authority but with the agreement that the original organisation would provide support to the people who lived there; where the setting is a clustered flat arrangement where people rent from a letting agency, property developer or from the local authority but where the local authority has made an arrangement with one provider to support everyone in the flats. The latter is a relatively common response to the Transforming Care agenda—local authorities and private providers are building blocks of flats as provision for people moving out of inpatient services but this is the only option available immediately limiting the choice people are experiencing.

One of the major contributing factors to this situation is a lack of social or affordable housing in almost every area of the country, compounded by a lack of creativity and collaborative working between different social service departments (e.g. adult services, housing and town planning) and the lack of a long-term view for service provision and creating inclusive communities. Austerity has made this situation worse as there is now greater need for social housing as poverty affects more and more people, including those with disabilities (Robson[10]). Lack of any housing, never mind adequate and appropriate housing, was a key theme across Europe in Šiška and Beadle-Brown's [39] recent review.

1. In most countries with an existing service system, there remains a mixture of service and support types, from larger hospital style settings to supported and independent living.
2. Only in a very small number of countries there are only community-based options, although these still include residential care for those with more severe disabilities.

37.6 Quality and Outcomes of Residential Care and Community Living

37.6.1 What Do We Know from Official Statistics?

As reported in Mansell et al. [3] and Šiška et al. [2] official statistics rarely report on the quality of services and measures of quality of life outcomes rarely feature in official reporting by inspection and registration bodies. In some countries such as the United Kingdom,

10 ► https://www.jrf.org.uk/report/using-social-housing-green-paper-boost-supply-low-cost-rented-homes, accessed 21/03/18

reports from inspection bodies do look at the experience of people in terms of whether they have their basic needs met, have choice, are treated with respect and dignity and can complain if things are not going well for them. In most countries, official statistics, where they exist, are more likely to monitor where people live (although even here they are limited, often to just the numbers in institutions with little information on where people live in the community) and how they are funded. There are few countries that collect data that allow us to monitor whether people with disabilities are really experiencing community living in the sense of being a full and active member of their community [2, 3]. Although there are a number of state and federal level outcome monitoring schemes in the United States, these all come with some challenges in terms of giving a clear picture of how well community living is being achieved in the United States [50].

37.6.2 What Do We Know from Research?

Over the years, there have been many different models for looking at the quality of services, but it is generally agreed that ultimately the quality of any service should be defined by the outcomes it achieves for the people supported [51, 52]. However, in order to achieve good outcomes for people, certain "inputs" or "structures" and "processes" are also important to recognise and understand, especially if outcomes are not as good as they could be [53, 54].

Defining Outcomes

The most common way that outcomes have been defined in this field has been through use of the concept of "quality of life". There are many other publications summarising the concept of quality of life and how it can be measured (e.g. [55–59]). The following brief summary is provided to contextualise the relationships between quality and the type and design of services.

Schalock et al. [57] suggested that quality of life is experienced when a person's needs are met and when they have the opportunity to access life enrichers. The international collaboration outlined eight domains and indicators of quality of life, with both objective and subjective indicators being seen as important (see ▶ Chap. 15 of the present textbook). Often we hear people in services saying that certain quality of life domains might not be "appropriate" or "achievable" for those with severe and profound intellectual disabilities and those with multiple disabilities or we experience people having lower expectations of what each domain might look like for those with the most severe disabilities. Bigby et al. [59] describe potential indicators for each Quality of life domain for people with profound and multiple disabilities, illustrating that the domains apply even to those with the most severe disabilities.

Engagement in meaningful activity and relationships has been considered a particularly important outcome, in which it is the vehicle by which almost every aspect of quality of life are realised [60–66]. In addition, Mansell and Beadle-Brown [64] argue that engagement is also an indicator of good quality of life—if people choose to be engaged then it is a sign of emotional well-being and a sign that the support for that person is working well.

Quality of Service and Quality of Life Outcomes in Residential Care and Community-Based Settings

Research has consistently shown that community-based services and support produce better quality of life outcomes than large congregate settings [67–70] and that dispersed settings produce better outcomes than clustered settings [12]. Supported living is considered to be particularly important for allowing people the security of tenancy rights or ownership and potentially more choice and control over where and with whom they live and who should support them in what activities. Research into the outcomes of supported living is limited but what there is suggests that choice and, in some studies, participation in recreational or community-based activities were better in supported living. However, outcomes in terms of safety (vulnerability to

exploitation), the frequency of scheduled activities, health and managing money were found to be poorer (See Bigby et al. [48] for a review).

One very consistent finding across the research looking at outcomes in community-based services is the variability in outcomes within different types of services. For example, Emerson and Hatton [67] found substantial variation between engagement levels in small group homes in the community across different studies. Although levels of engagement were on average higher in community-based housing than in small or large institutions, there were people in small group homes in the community, which were experiencing similar or poorer outcomes to those in institutions. However, no study has ever shown that congregate or clustered settings can achieve as good outcomes as have been achieved in community-based settings. And some research has shown that size is particularly important in terms of choice and control [71]. These findings led to the conclusion that small, ordinary and dispersed housing in the community is a *necessary* but *not sufficient* condition for a better quality of life [9, 72].

What has emerged very clearly from the research over the past 50 years is that, apart from the severity of disability, only one factor has been found to consistently and directly relate to quality of life outcomes and that is the way that people are supported minute-by-minute [11, 72]. Outcomes have been found to be significantly better when staff used an enabling and empowering style of interaction that focused on doing things *with,* not *for* or *to*, people. This style of support is usually referred to as "Active Support" (See Mansell and Beadle-Brown [64] for a review). Mansell and Beadle-Brown [64] describe active support as an enabling relationship by which staff and other carers provide graded assistance to ensure success—assistance that is tailored to the needs, pace and preferences of the individual delivered in a person-centred, warm and respectful way and making the most of all the opportunities available at home, in school, in the community, at work.

Of course, active support is not the only form of person-centred support but has been argued to be an important foundation for other approaches such as positive behaviour support, alternative and augmentative communication, autism-friendly practices such as the SPELL (Structure, Positive approaches and expectations, Empathy, Low arousal, Links) framework [54, 64, 73–78]. It is also argued to be a good indicator of support that produces better outcomes for people with Intellectual and Developmental Disabilities [73]. Active support is founded on evidence-based practice—combining elements of applied behaviour analysis, learning theory, occupational therapy and physiotherapy but in a holistic person-centred package, which can be practiced every moment of the day in any environment [64]. Research has found that, in addition to increasing levels of engagement in meaningful activity and interactions in the home, implementing active support results in more involvement in a bigger range of activities at home and in the community, more opportunities for community inclusion and for self-determination, increases in skills, reductions in challenging behaviour, and improvement in mental health [53, 73, 79–86]. It has also been found to improve job satisfaction and retention of staff [79].

In terms of what determines whether staff work in this way, the evidence base is still developing. To date research has indicated that practice leadership by front-line leaders plays a key role, and that training (including both classroom-based and hands-on elements) is also a contributing factor of higher levels of active support [73, 87–89] . In addition to the impact of practice leadership and training in active support, recent research from Australia [90, 91] also indicated that inputs and structures such as size of setting are important in determining how well staff provide active support and how well it is maintained over time—a setting for more than six people was associated with lower levels of active support and poorer maintenance of quality of support over time.

Experience of supporting organisations to implementing change has shown that management awareness of and commitment to new practices to be introduced is key [64], and this has been supported in recent research by

Bigby et al. [92]. Bigby et al. found that better implementation of active support was associated with a number of organisational and senior manager factors:

- Fewer settings provided by the organisation.
- Greater time implementing active support.
- Senior leaders shared prioritisation of practice and active support.
- Senior leaders strongly supporting practice leadership.
- Practice leadership located close to front-line service delivery.
- Concentration of practice leadership tasks with front-line management.

Although research to explore this would require a multi-national study, having a supportive wider motivational context is likely to be important in helping organisations to work in ways that make a difference in terms of outcomes for people in any type of setting. Policy, funding systems, inspection and registration systems, auditing systems and the focus of staff education and workforce development will all influence to what managers and staff pay attention [64, 80].

1. Living in an ordinary house dispersed in the community is a necessary but not sufficient condition for a better quality of life.
2. The factor that makes the most difference is whether people are supported in a way the enables and empowers them.
3. For staff to work in this way, they need the right skills through training and leadership and they need to be motivated.

37.7 Benefits, Risks and Solutions

This final section summarises how well some of the current models of community-based housing and support can promote a good quality of life and how well they reflect Article 19 of the UN Convention on the Rights of Persons with a Disability (UNCRPD).

Owning or renting your own home, a home that is part of the ordinary community and does not set you apart from others in the community is unequivocally the best option for people's quality of life. This is what is intended by Article 19 of the UNCRPD. This model can provide a higher level of emotional well-being (security), material well-being (owning or renting own's home), social inclusion (physically part of a local community) and rights (e.g. tenancy agreement) than any other form of accommodation or support arrangement.

Of course, as mentioned above, just owning or renting your own home does not guarantee a good quality of life in any other domain. Without the right support to make choices about where and with whom you live, to take advantage of all the opportunities available in the home and community, and to experience full and active citizenship, people can perceive as much disengagement, disempowerment and isolation as in a large institution [73].

Even when coupled with a personal budget to pay for personal assistance, many people with intellectual disability still need support to make choices about who they want to support them or how to spend their time. They will only have real choice if there is more than one good service to choose from, or a bank of staff with the right knowledge and skills to employ. Employers (whether an agency or an individual themselves) have to be able to provide training, support and leadership to staff if they are going to be able to work in a way that improves outcomes for the people they support. While people with physical disabilities and those with less severe communication difficulties can often guide and even train staff themselves, this is not so true for those with more severe disabilities. Families can often be a source of useful knowledge but they themselves may not be aware or have had little training in how to support higher levels of quality of life. Of course, it is perfectly possible to create a system whereby anyone who wants to work with people with disabilities in any setting can gain the right skills (which then would be adapted to the individual(s) they are supporting) and where those

managing staff can learn about and receive support to manage a staff team. However, currently this does not yet exist in any country.

While supported living, personal budgets and personal assistance offer the greatest possibilities for choice and control and potentially for being a full part of society, these are not yet options in many countries where the money available for people to live in the community is very limited. People do not have benefits such as housing benefits that can help them pay rent. Rightly or wrongly, mental capacity and guardianship laws while under revision in many countries, often mean that people cannot sign contracts. Housing in the main community can be so poor that it can be unsafe for those with disabilities and people do not have the money to fix or maintain such environments.

Even in countries where supported living and personal budgets are available, issues such as lack of adequate and appropriate housing to rent can create barriers, as discussed above. For example, this is one of the reasons often given for why those in assessment and treatment units and private hospitals in England face delays in being discharged and why there has been an increase in the development of clustered flats for those with intellectual disability and/or those on the autism spectrum.

As such, in order to ensure that people are not living in institutions or other larger congregate settings, we may need to compromise slightly in order to make change in the right direction happen. If we wait to make until personal assistance and/or supported living is available everywhere, then there are several risks and it can be counter-productive in the progressive realisation of Article 19 of the UN Convention. First, it will mean that those in institutions will stay there for longer, potentially forever in some countries. Second, it will mean that organisations will develop services that are called "supported living" but which are not in the true spirit of supported living, that is, where people really do not have choice over their living and support arrangements (or in many cases over their day-to-day lives) and will live in larger groups that sets them apart from the rest of their community.

Jim Mansell once wrote about compromise for a workshop focused on developing community-based services for people with disabilities in Eastern Europe:

» I would not agree that group homes were *needed* for people with more severe disabilities, since we know that everyone can have a good life in their own home. I am more comfortable with the justification that we are providing some group homes because that is all we can pay for at the moment (i.e. the justification is nothing to do with the individuals we serve and all to do with the context in which we are working at the moment)… I would not agree that grouping more than a handful of people together was sensible. So for me, it would be no to institutions, villages, campuses, hostels, units or clusters. But what about a group home for three or four people? Well the research on group homes shows that run well they can achieve many good things for the people living in them, and that the difference between a well-supported group home and supporting people in their own homes is not necessarily very great. I would still prefer to support people in their own homes (because I would value the greater housing rights this would give people as well as the symbolism) but if a small group home was all that could be achieved [right now] I would be inclined to agree, while all the time pushing for the features that would make it more like supported living (so e.g. choice of co-residents, choice of staff, legal tenancy)…

» My picture of the service development landscape is that way ahead in the distance are the *present boundaries of the possible*. These are the things that we know are possible and desirable but they are the leading edge. This boundary is a horizon, so that as we approach it the definition of what is now possible moves ahead (because of changes in technology but also changes in attitudes and expectations). Behind this boundary there is an area in which we are working with models that, though we know they are not ideal, seem to us to be steps on the road to the horizon, not least because they are *capable of transformation*. At the back of

> this zone is another boundary, beyond which models of service seem now to be unacceptable. We look at them and think "how could we ever have thought that was acceptable"? Our task in supporting principled service development is to move this *frame of consensus* forward, connecting people to the vision on the horizon and helping them see that they are on a journey. Not everyone is going to be able to be a pioneer out there near the horizon but those who come behind need to know they are heading in the same direction.

In conclusion, the model most in line with the UNCRPD is that of supported living/personal assistance—where people receive the support they need and choose, in their home that they rent or own (individually or with others, according to their choice). This home needs to be dispersed in the community, chosen by the person from the same range of homes in which other people in the community live and which is adapted to the needs of the individual. Where this is not possible right now for resource reasons, then the model that offers the next best outcomes for people is that of small group homes (residential services where the organisation provides both the accommodation and the support). Ideally, these should be for 3–4 people and certainly for not more than six people. Where people want or need to live on their own but appropriate housing is not currently available for them to rent or own in their own right, then living in an individualised residential service, where an organisation already owns or rents the property, should be considered over and above a clustered setting where everyone has an intellectual disability. In such small individualised or group home settings, it is possible for people to have a key to their front door and be supported to use it, for staff to work on the basis that they are supporting people to live their lives in their own home and own community, for people to be supported to make choices about where and with whom to live, whom they wish to support them, how they wish to spend their money. The main quality of life domain limited in this model is that of rights—in particular, tenancy rights and security of living situation. Owning or renting your own home is also an indicator of material well-being and an indicator of status in most societies. Even if people have to live in small good quality residential care settings initially, looking for possibilities to expand the range of future possibilities for living in their own home with support should be the next stage of progressive realisation of article 19 of the UNCRPD.

1. Supported living ideally through the use of personal budgets and personal assistance is the model of housing and support that is most in line with the UNCRPD Article 19.
2. However, this option is not yet available in all countries for all people with intellectual disabilities.
3. If we wait until the system is fully developed before we move people from institutional settings, then we risk people never moving and attitudes of society will never change.
4. If compromise is needed in the form of small residential care settings, then we have to pay attention to ensuring that these promote the inclusion, self-determination and rights of the people who live there.

Key Points

- People with intellectual disability have a right to live and participate in the community with choice over where and with whom they live and support to achieve this right. Many people with intellectual disabilities still live in congregate settings and experience poor quality of life outcomes.
- People with intellectual disabilities are more likely than other people with disabilities to live in large congregate settings, with little choice and with limited participation in the community as active citizens.
- At home people with intellectual disabilities spend most of their time disengaged and with little contact from

others, including those they live with. This is likely to result in challenging behaviour.

- Quality of life outcomes is determined not only by where people live but by the support they receive—how people provide support is critical. Outcomes are better when support is enabling and empowering, doing "with" the person not "for" or "to" the person. This type of support has become known as person-centred active support. When staff have the skills and motivation to work in this way, outcomes for people are substantially better.

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Work and Occupation

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Contents

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Learning Objective

- The main purpose of our contribution is to expand some aspects of the research and practice about employment and employment services for people with intellectual and developmental disabilities (IDD) for which authoritative summaries and presentations are already available [1–5]. We will explore the role played by the co-occurrence of intellectual and psychiatric disorders on employment outcomes, and we will extend, whenever possible, the analysis to different national contexts, emphasizing the role of contextual factors in getting and maintaining a job.
- The reader will learn that with the approval of the United Nations Convention on the Rights of Persons with Disabilities (UNCRPD), a complete and binding declaration of rights of persons with disabilities has been established [6–8]. Despite these international commitments, the majority of persons with IDD continue to work in segregated settings and with substantial pay gaps compared to non-disabled population.
- One of the fundamental learning objectives of this chapter is to offer an international overview of the employment rate and presence in the labor force of people with IDD. The chapter then focuses on the main empirical evidence on factors explaining access to work of people with IDD [9].
- The complexities of employment projects for people with IDD are elucidated as well as the need for an employability conceptual framework to guide employment projects.
- The panorama of the strategies, services, and techniques of Vocational Rehabilitation (VR) is presented, and the reader can have an updated reference framework on sheltered workshops, supported employment, and social firms' programs.
- The scientific evidence supporting the importance of work on personal identity development, social inclusion, and quality of life of persons with IDD is discussed.

38.1 Introduction

The issue of access to work for people with disabilities has long been central in debates on national and international welfare policies and programs of care and rehabilitation.

In the last decades of the last century, the scientific community agreed on considering employment as one of the fundamental aspects for social inclusion, independence, and quality of life of persons with disabilities and therefore one of the ultimate goals of rehabilitation processes.

However, to date this position has not been sufficiently implemented in many systems of policies and services across the world, and people with disabilities, especially those with intellectual disabilities (ID) and/or autism spectrum disorder (ASD), continue to have much lower opportunities and employment rate than the rest of the population.

In recent years, scientific research in the field gained momentum, and several studies have addressed barriers to employment participation and effectiveness of the interventions adopted.

Policies and support programs are very heterogeneous across different countries, both in terms of nomenclature and methodology, and thus difficult to compare. Nevertheless, available studies are providing important indications on the most effective actions that should be put in place by health and social services to increase occupational opportunities.

The present chapter provides a comprehensive overview of the current knowledge on work and occupation of people with ID and/or ASD, with a main focus on factors that can hinder or facilitate work access, the role played by the co-occurrence of psychiatric disorders, and employment options and support programs that can improve outcomes, especially person-centered outcomes.

38.2 Employment Rate and Labor Force Participation in the Population with Intellectual and Developmental Disabilities

Studies on employment rate and labor force participation of persons with IDD do not share common definitions and metrics [11–13]. The identification criteria of population are quite different, and often the characterization of participants is poor or absent with regard to comorbidities and ability levels. Very often disability is self-reported and not clinically confirmed. It is worth noting that similar problems affect the majority of the scientific literature on this topic [14]. Common definitions include terms such as "mental retardation," "learning disability," "cognitive performance restrictions," "cognitive disability," "developmental disability," "developmental delay," "intellectual and developmental disabilities," and "autism." Sometimes, it is not clear whether the definition refers to clinical diagnostic criteria or functioning profiles. Also, the way occupational outcomes are conceptualized makes international comparisons difficult. Furthermore, in many countries, current legal framework allows persons with disabilities to work in segregated firms, commonly indicated as sheltered workshop or paid facility-based work, and with subminimal wages [15–17]. In many cases, statistics on employment rates do not distinguish between jobs in open labor market and in segregated settings.

To give an updated picture of the employment rate and work conditions of persons with IDD, we chose population-based studies and tried to select data and information useful to enhance between-country comparisons. Cameron [18] has reported data from the 2006 Canadian Participant and Activity Limitation Survey (PALS). Persons with "intellectual disabilities" were identified by asking the participants if a doctor, psychologist, or other professionals ever said them that they had a developmental disability or disorder. Twenty-five percent of the identified persons with ID were working compared with about 50% of people with other disabilities and about 75% of people without disabilities. Persons with ID were about six times more likely than others to have never worked. The Canadian survey did not report data about the proportion of people working in sheltered workshops. For working-age people with ID and any employment income in year 2005, their average earnings were $18,172 compared with $29,669 among people with other disabilities and $37,944 among people without disabilities. The proportion of people with ID below poverty threshold was 19.7% and among persons without disabilities, 10.2%. A secondary analysis of the 2012 Canadian Disability Survey confirmed that 24.1% of people with "developmental disabilities," self-identified as in PALS survey, were employed, while the estimate of the specific employment rate of people with autism was 14.3% [19]. The proportion of labor force participation was 34.1%, approximately half of the proportion of the pooled group of persons with disabilities.

Yamaki and Fujiura [20], analyzing data derived from the Survey of Income and Program Participation, a national representative longitudinal household survey of noninstitutionalized adults in the United States, have found that only 27.8% of adults with IDD had a job in any given month compared with 75.1% of the general population. Monthly earned income for workers with IDD was approximately half of that of population without disabilities. Average earned income was estimated $1298 for persons with IDD versus $2206 for general population. In this case, adults with IDD were identified using the following criteria: they were between the ages of 22 and 65 years, had a childhood originated condition that resulted in work limitation, and had either any of the categorical labels associated with the old definition of IDD or functional limitations in two or more of the six life activity domains, as described in the Developmental Disabilities Assistance and Bill of Rights Act (2000). The 2010 survey edition [21] has confirmed an estimate of 26.1% of employed persons with ID and 30.8% of persons with other developmental disabilities. Siperstein et al. [22] estimated that 34% of adults with IDD aged 21–64 years

were in the labor force compared with 83% of adults without disabilities. The study sample was selected from approximately 341,000 households. Respondents were parents/guardians of adult children aged 21 or older who declared that their adult children had been diagnosed with an ID. Of the adults in the sample with mental retardation or intellectual disability, 18% were in competitive employment and 13% were employed in a sheltered setting. The remaining 3% were either self-employed, or their employment setting was not categorized. The unemployment rate in the sample was 9% versus 21% in adults with and without disabilities, respectively.

National Core Indicators project, based on the 2012–2013 Adult Consumer Survey collected a wide range of information on recipients of state developmental disability service in the United States. The employment rates of non-institutionalized working-age persons (ages 21–64 years) with IDD based on the 2012–13 survey were 14.7%. Slightly less than half of the service recipients participated in an unpaid facility-based activity during the day. Over one-quarter (27%) were in paid facility-based jobs, and 22.6% took part in unpaid community-based activities during the day [23]. Since 2008, the American Community Survey (ACS) allowed people to self-report "cognitive difficulty" on six questions regarding physical, mental, emotional problem, having difficulty remembering, concentrating, or making decisions. Data for 2014 show that persons with any disability or a "cognitive difficulty" were employed at much lower rates (33.7% and 23.5%, respectively) than those without disabilities (72.9%) [24].

The National Longitudinal Transition Study-2 (NLTS2) focuses on the experiences of the US secondary school students with disabilities, as they go through their early adult years. A subset of young adults with disabilities, who were out of secondary school, and in the ages of 21–25 years were selected, telephone interviews were conducted with their parents and, whenever possible, with the young adults themselves; 38.8% of persons with "mental retardation" and 37.2% with "autism" were involved in any type of paid employment outside the home at the time of the interview [25]. Average hourly wage was $7.90 for "mental retardation" and $9.20 for "autism." The hourly wage for all categories of disabled persons ranged from $7.90 to $11.10.

A recent study on wages of persons with disabilities in the United States has provided important insights into the theme. Based on a comparison of several national databases and an accurate evaluation of the work position and qualification of workers, the study has confirmed the wage gap between disabled and non-disabled population but the estimated gap was smaller than that showed by a crude comparison. Persons with "cognitive disability" appear among the most disadvantaged groups, but the main source of inequality is related to the hiring process and the kind of job acquired [26].

International data show lower rates of employment in Europe than in Canada and the United States. This trend has been clearly depicted by Parmenter [1]. Although the issue should be better investigated, we will try only to highlight some partial evidence. Through the Intellectual Disability Supplement of the Irish Longitudinal Study on Aging (IDS-TILDA) [27], a representative sample of 753 individuals with ID (45% male and 55% female) from across Ireland were drawn from the National Intellectual Disability Database [28]. The nature of service provision in the intellectual disability area in Ireland ensures an almost complete capture of data on all individuals with a moderate, severe, or profound ID. Overall, 6.6% of the study participants were in real paid employment (supported or competitive) and a further 12% working in a sheltered workshop. A total of 73.5% of respondents were not in labor force.

In 2013–14, the proportion of adults with a "learning disability" in paid employment in England had decreased to 6.7% from 7.0% in 2012–2013 and 7.1% in 2011–2012 [29]. Similar estimates were published in the "People with learning disabilities in England 2015: Main report" [30]. Overall paid/self-employment rate for working-age adults with

learning disabilities reached a peak at 7.1% in 2011–2012 and was at 6.7% in 2013–2014. In 2014–2015, the employment rate was 6.0%. Employment rates are broken down by gender, with men with learning disabilities consistently reporting employment rates 1.1–1.8% higher than women with learning disabilities.

In Norway in 2010, 22% of adults with ID participated in state-organized job creation or the regular workforce. Sixty percent had their day activities in municipal day centers, and 17% of these were mainly involved in production activities. Three percent participated in adult education, while 16% had no day activities of any kind, a rise from 3% in 1989 [31]. Similar results were found in Sweden where a significant and increasing majority of people participated in day activity centers [31].

Official estimates of the population with disabilities in Italy are based on a National Health Interview Survey (NHIS) [32], conducted every 5 years. The survey is carried out on a sample of about 60,000 households and 162,000 respondents distributed in 1456 municipalities. 8.7% of individuals between 15 and 64 years of age, self-reporting mental retardation in NHIS 2013, were employed, and the labor force participation estimate was 17.7%. There are no national estimates of the proportion of people with IDD involved in segregated work settings. A Finnish study [33] based on a cohort of people with ID born in 1966 showed an employment rate in the period 1993–2000 of 24% with a mean duration of the working periods of 6.8 months. The same group experienced an unemployment rate of 56% with a median duration of unemployment of 138 days per year.

Studies on employment rate and labor force participation of persons with IDD do not share common definitions and metrics; therefore, only conditional indications can be drawn. Population-based studies confirmed the presence of a gap between disabled and non-disabled population in employment rate and in average earned income. International data show lower rates of employment in Europe than in Canada and the United States.

38.3 Employment Outcomes in Selected Groups of People with Co-occurring Intellectual and Developmental Disabilities and Mental Health Problems

The clinical and epidemiological literature have described the importance of co-occurrence of IDD and mental illness. Despite the difficulty of the prevalence estimates and the limitations in the available studies, it seems established that co-occurring mental disorders in children, adolescents, and adults with IDD are more predictive of major restrictions in educational and vocational participation and social inclusion than the severity of the IDD [34–36].

Unfortunately, the specific effects on employment of co-occurring ID and mental illness or autism and mental illness have not been investigated at population level. In order to assess the effect of these co-occurrences, we have to consider clinical studies on selected populations. To the best of our knowledge, the original studies are very limited in number and with many methodological pitfalls.

Considering that the international literature has developed a considerable evidence about the low rates of labor force participation, below-average earnings, and poor working quality for people with mental illness, it seems reasonable to hypothesize that individuals with co-occurring IDD and mental illness encounter additional barriers and problems in getting and maintaining a job, and that cognitive impairment worsens work problems [37]. Cook et al. [38] designed a randomized implementation effectiveness trial by comparing different models of supported employment with a variety of different control conditions in a population of 1273 persons with severe mental illness.

Co-occurring ID was specifically considered an independent variable both in experimental and control groups. Diagnosis was coded using a uniform protocol from personal case file by a panel of three disability experts. Employment outcomes evaluated during the 24-month follow-up period included total

earnings, total hours worked, and access to competitive employment. The study supported the evidence of an influence of comorbidities on some but not all work outcomes and has also shown an effect of intellectual disability on competitive employment: individuals with ID were less than two-thirds as likely to work competitively. The supported employment group has shown better results than the control group for the main employment outcomes.

The already cited cross-sectional study based on Intellectual Disability Supplement of the Irish Longitudinal Study on Aging (IDS-TILDA) [27] has shown that the proportion of people in real paid employment is strongly associated with the severity of ID. People with a diagnosis of depression in real paid employment were half the number of those in sheltered workshops, and most elderly people with ID are unemployed, as McCausland and colleagues said [39].

Martorell et al. [40] studied the personal factors that determined work outcome in a group of 179 adults with ID, in Spain. The predicted outcome was the achievement of remunerated employment that means, with reference to the Spanish context, assignment to "sheltered workshop" versus "sheltered employment program." Presence of psychiatric disorders was evaluated using the Spanish version of Psychiatric Assessment Schedule for Adults with Developmental Disability (PAS-ADD) [41]. The presence of psychiatric disorders is significantly related to a lower likelihood of paid jobs.

Holwerda et al. [42] analyzed data from a Dutch cohort study, "Young Disabled at Work" whose aim was to find predictors of work participation in young adults aged 15–27 years. The study enrolled only persons with mild ID certified by a physician of the Dutch Social Security Institute. Interestingly, people with moderate and severe ID were excluded because very often certified, a priori, as unable to work. Occurrence of psychiatric and other comorbidities was coded from the same certification process records. The authors selected a long list of personal and social influencing factors or work-related participation ranging from sociodemographic variables to psychological characteristics (self-esteem, motivation, expectations, etc.), to social and family support.

The two final Cox regression models estimated for explaining job finding and job maintaining failed to show any direct effect or comorbidities and specifically of psychiatric comorbidities on occupational outcomes, while individuals living with parents or family or living independently appeared to be three times more likely to find work than individuals living in residential placements or sheltered accommodation. Highly motivated individuals were three times more likely to find work than less motivated individuals. The authors suggested that living situation and motivation may be interpreted as a proxy for disability severity that was not directly included in the study. In particular, individuals who were in residential placements probably had more severe disabilities and in general showed very low levels of participation in any kind of work.

Siperstein et al. [43] evaluated the effect of a large set of predictors on the pathways to employment of people with ID. The study group was composed of 1055 parents or guardians of adult children aged 21 years or older contacted by telephone. The final estimated multinomial logistic regression models showed that the presence of a diagnosis of an "emotional/behavioral disorder" halved the likelihood of employment in both competitive and sheltered settings.

Sung and colleagues [44] studied the predictors of competitive employment in a sample of people with Autism Spectrum Disorders (ASD) receiving Vocational Rehabilitation (VR) services in the United States. Data were extracted from the Rehabilitation Service Administration Case Service Report. The final sample comprised 857 males and 839 females aged 16–25 years. Co-occurring anxiety and depression caused a 43% reduction in the odds of finding employment in males but not in females.

Bush and Tassè [45] provided a comparative cross-sectional analysis of employment outcomes of three groups of persons with dis-

abilities: ID, ASD, and Down Syndrome (DS). Data were based on National Core Indicators' Consumer Survey that randomly selected approximately 20,000 individuals receiving public services from their state developmental disabilities system. The authors found that 16% of individuals with DS, 14% of individuals with idiopathic ID, and 10% of individuals with ASD were employed in the community. For all three disability groups, lower severity levels of ID were associated with increased probability of paid community employment. Reporting fewer reasons for "taking psychotropic medications" was related to an increased probability of paid community employment in all the three groups considered.

Despite scarcity and methodological shortcomings of available studies, it seems that in children, adolescents, and adults with IDD, co-occurrence of mental disorders increases the risk of restrictions in educational and vocational participation and social inclusion more than severity of IDD.

38.4 Framing Services and Practices for Employment of People with Intellectual and Developmental Disabilities and Related Research

38.4.1 Target Population

The access to employment or VR services is regulated by country-specific eligibility processes and criteria. In many countries, these processes include a medical evaluation of the work capacity. In a series of interconnected studies, the Organization for Economic Co-operation and Development (OECD) [46–49] has documented the progressive growth, in all OECD countries, of the percentage of people recognized as unable to work. The low participation in the labor force of persons with disabilities and in particular of people with IDD and mental health problems may be due, at least partially, to evaluative practices, dominant in legal medicine, which emphasize the limitations in activities due to impairments. The role of contextual factors is ignored as well as the impact of physical and relational barriers or the enabling potential of environmental facilitators [50].

Bickenbach [51], in discussing the opportunity to overcome the concept of work capacity and arguing on the opportunity to introduce an evaluation model based on the International Classification of Functioning Disability and Health (ICF) [52], recalls the endorsement of ICF conceptual framework made by the US Institute of Medicine. In particular, the Institute of Medicine studies of the Social Security Administration's (SSA) disability evaluation [53, 54] has concluded that the ICF interactive model of work disability was both conceptually and empirically more realistic than the impairment-based model implicit in the SSA disability evaluation of the claimant's ability "to engage in any substantial gainful activity." Nonetheless, "SSA does not have the resources to perform such an extensive assessment for each of the approximately 2.6 million disability applicants who come through its doors each year" [54].

Bickenbach argues that evaluative practice based on a strict consideration of impairment evaluations not only increases social spending, but it would be a violation of basic principles of human rights, and the underlying values of equal dignity and respect. Similar risks have been noted also when the access procedure is directly managed by employment and vocational rehabilitation services [55]. The use of the ICF in the evaluation of workability of people with disabilities has been the subject of intense research work in the countries of northern Europe in the last 10 years but, still, with limited practical implementation [56].

Although country-specific differences, evaluation of work capacity is generally limited by an excessive consideration of impairment-derived limitations in activities and an underestimation of the impact of physical and relational barriers, environmental facilitators, and contextual factors.

38.4.2 Labor Market

It has been argued in several studies and literature reviews that the employment of persons with disabilities has traditionally been studied from the point of view of the "supply side," examining the medical, educational, psychological, and vocational inputs that affect a person's functioning and job skills. This approach, however, ignores the fact that the employment requires an alignment between the supply of individuals and the demand for labor on the part of the employer [9]. Labor market can be defined as the market in which the employees look and compete for employment and the employers look and compete for workers. The labor market affects the decisions of the participants who in turn, by their own decisions, affect the labor market itself. The major idea of labor market economics is the analysis of the determinants of various dimensions of labor supply and demand. Moreover, the dimensions of labor supply and demands interact to determine wages, employment, and unemployment.

Labor market is influenced by global competition, free trade, change, and technological innovations. Especially in recent years, the rapidity of technological, communicative, social, and political changes is making the labor market and the supply and demand dynamics that rule it ever more unpredictable than before [57]. Services for the employment of people with IDD are confronted with the dynamics of the labor market. Far from being a mechanical process of "matching" between the person and a specific job, as it was sometimes conceived in the past, the main goal of services is to increase the chances of their recipients getting to work. We can generally describe their effort as means to increase the "employability" of persons with disabilities.

Traditional principles of labor market economics, based on the analysis of the determinants of various dimensions of labor supply and demand, ignore the fact that employment requires an alignment between the supply of individuals and the demand for labor on the part of the employer and need to be adapted to specific characteristics of persons with ID/ASD.

38.4.3 Employability

Promoting employability of disadvantaged people is one of the main goals of labor market policies promoted at national, regional, and local levels [58, 59]. Employability has been defined as a "cornerstone of the new labor approach to economic and social policy" [60]. The United Nations (UN) has considered employment as one of its four policy priorities. In 2001, the UN Youth Employment Network has suggested that "…all countries need to review and re-orient their educational and vocational training and labor market policies to facilitate the school to work transition and to give young people a head start in working life" [61]. However, employability is often used with reference to a wide range of meanings. Over the years, different definitions of the concept have been used. Some of these mainly emphasize the role of individual skills and competencies [62, 63].

McQuaid and Lindsday [64] have conducted a current and historical analysis of the different applications of the concept in labor market and suggested a broad conceptual framework with three main interrelated components such as (1) factors related to individuals (including employability skills and attributes, demographic characteristics, health and well-being, job seeking, adaptability, and mobility), (2) personal circumstances (including household circumstances, work culture, access to resources), and (3) external factors (including demand factors, enabling support factors). They have provided a detailed list of domains and variables describing these three main components.

This employability framework is consistent with the theory of evolutionary systems and the bio-psycho-social and contextual models of functioning and disability [64–66], which consider the individual evolution as the result of the interaction between the person

and the context [67–69]. Research literature on the employment of people with disabilities can be easily classified within this conceptual framework, considering domains and variables included in the analysis and the causal links investigated. Rarely, all the domains of the depicted framework are included in a single study and, in general, an excessive simplification of the employability framework is the main methodological limitation of many studies. Some areas, for example, personal circumstances and the relationship between individual and external factors, remain underrepresented in international literature.

Employability research must be guided by the consideration of a broad list of personal and contextual factors organized in an appropriate conceptual framework. Many empirical studies traceable in literature are based on poor representation of the factors influencing employability.

38.4.4 Employment Services and Practices for People with ID and ASD

The World Health Organization has defined rehabilitation as "a set of measures that assist individuals, who experience or are likely to experience disability, to achieve and maintain optimum functioning in interaction with their environments." It is a process aimed at enabling disabled persons to reach and maintain their highest level of adaptation, to remain in or return to their home or community, live independently, and participate in education, employment, and civic life [70]. It is common in international literature to identify VR as an umbrella term to identify a large area of practices and services that consider employment and full inclusion of adults with disabilities in society the main goal [44, 71]. However, there is no consensus on a common definition of VR at international level, and also the implementation of VR service at national level is extremely different. An adequate understanding of the VR organizations would require a country-specific description that is outside the scope of this text. We will highlight some common traits, and we will suggest some useful literature references.

For the Vocational Rehabilitation Association VR is "… a process, which enables persons with functional, psychological, developmental, cognitive and emotional impairments or health conditions to overcome barriers to accessing, maintaining or returning to employment or other useful occupations" [72]. Marnetoft enriches the definition, including all actors involved in VR, and emphasizes the search for evidence-based practices. In its perspective, VR is "a multiprofessional, evidence-based approach that is provided in different settings, services, and activities to working-age individuals with health-related impairments, limitations or restrictions with work functioning, and whose primary aim is to optimize work participation" [73].

The absence of a general consensus on the definitions but, above all, the great diversity in the practices at the national level have not helped the construction of solid scientific outcomes. Several authors have claimed that evidence on employment outcomes for people with IDD and mental disorders remains fragmented and insufficient to develop guidelines for best practice [74]. However, the literature reviews and systematic reviews [75–78] converge in identifying main macro-approaches to VR in respect to which a vast amount of research has been carried out over the past 50 years. We briefly describe these main approaches distinguishing them into three groups: *Sheltered Employment programs*, *Supported Employment programs*, and *Social firms*.

The term *Sheltered employment programs* is used to address a wide range of segregated vocational and non-vocational programs for individuals with disabilities [79]. These programs present wide heterogeneity in terms of mission, services offered, and sources of funding. They are designed for individuals considered temporarily or permanently unemployable in competitive settings. The majority of employees are individuals with disabilities, who perform typically easy, repetitive tasks such as prod-

uct assembly or packaging, often below minimum wage. Support and skill training are provided on-site [80]. Employees may receive a training allowance in addition to other social benefits [1].

The large majority of literature on sheltered employment programs comes from the US research and debate. Sheltered employment programs are usually classified into two groups [81]:

- Extended employment programs: They refer to a long-term or permanent placement.
- Transitional employment programs: They provide specific training required for a successful employment in the open market.

Sheltered employment programs and services remain the most common destination of adults with IDD in the United States despite a large political and legal effort to their re-sizing [82]. In the last 20 years, approximately 80% of public resources have been directed to facility-based work or non-work settings programs versus 20% directed to supported employment programs [83]. This unchanging situation generated a wide and lively debate and a rich research work aimed at highlighting "pros and cons" of this type of program. There is a substantial evidence showing that sheltered employment programs have a low rate of successful transition of individuals to competitive employment [80, 84]. So the long-term impact of sheltered employment on community integration of individuals with disabilities is very limited [85].

The sheltered employment wages are low or insufficient to allow economic independence of employed people [83]. A well-known study by Cimera [86] has compared employment outcomes for two cohorts of people with ID: one in supported employment programs and the other that participated in sheltered workshops. The study has revealed no statistically significant difference in the percentage of people employed in the two groups; the weekly earning was significantly higher for supported employees than for people in sheltered workshop and the employed sheltered workshop cohort was more expensive to serve than the supported employment group. Similar results were found in studies comparing selected cohorts of people with ASD and ID [87, 88].

In a research aiming to investigate "Why do adults with intellectual disabilities work in sheltered workshops?" [16], it was found that employment outside sheltered workshop was often considered too complex or frustrating for many persons with IDD [89]. In other cases, the decision to stay in a sheltered workshop was related to lack of transportations accessing community employment settings, and fear of losing disability benefits. However, Migliore [16] discovered that 40% of the families, 46% of the adults with ID, and 60% of staff members of sheltered workshops reported that nobody had encouraged the adults with ID to seek employment outside of sheltered workshops.

The supporters of sheltered employment emphasize the avoidance of physical and psychological risks of competitive work and the greater opportunities that protected work offer to establish and maintain friendly relationships [90]. Despite these uncertainties, most authors, research work, and the political commitment support the idea of re-sizing sheltered workshops in favor of supported employment strategies. In this regard, the National "Employment First" initiative in the United States promotes employment in integrated community-based business as the first preferred option of youth and adults with significant disabilities [82].

There is no common definition of sheltered workshops at the European level; however, like in the United States, an important research identified, also in European countries, two main types of sheltered workshops. Traditional sheltered workshops are long-term permanent places of employment for people whose disabilities preclude them from entering the open labor market, while transitional sheltered workshops aim to provide people with disabilities with the support and the skills needed to access non-sheltered employment. For some European countries, it has been documented that people with IDD

are the prevailing group in sheltered workshops (i.e., 77% in Germany) [91].

The employment status and remuneration of sheltered employees vary across the European Union (EU). Some member states grant employee status to disabled people in sheltered workshops, which entitle them to a minimum wage. However, the majority of sheltered workers do not have employee status and receive varying forms of remuneration but not a salary. It is not possible to collect comparable data on the nature and scale of sheltered workshops for the EU. This is primarily due to the existing differences in the names, structures, and legal frameworks, which implement sheltered workshops in the member states. The research documented a shift toward the transitional model of sheltered workshops in Europe. However, the estimate of people in transitional sheltered workshops that move on to the open labor market is no more than 3% [91].

Supported employment (SE) is a VR alternative approach to sheltered employment clearly oriented to place people with disabilities in competitive job positions without any extended preparation and training [1, 77, 92]. Since its introduction in the early 1970s, it has become a worldwide practice to help individuals with disabilities who are unable to successfully gain or retain employment by themselves and to enter the open labor market with the same dignity as people without disabilities.

People with disabilities are supported in finding as quickly as possible an employment in competitive settings. Furthermore, they receive intensive on-job support and training through job coaches. All supported employment projects, especially those for people with IDD, share the belief that long training programs drive away from the job placement, risk to promote dependency, discourage people from seeking a competitive employment, and are not effective in helping people with disabilities to acquire the skills required by the future job [93, 94]. The SE programs have gradually spread to become the internationally preferred VR practice, in particular, because they respect the core principles of the UNCRPD and support the right to inclusive work for all persons with disabilities.

According to a widely shared vision in literature [95–97] and public documents [98], SE can be defined as "a competitive paid work in an integrated setting with ongoing support services for individuals with severe disabilities for whom competitive employment has not traditionally occurred or for whom competitive employment has been interrupted as a result of a severe disability" [96]. The European Union of Supported Employment defines supported employment as a practice that provides "support to people with disabilities or to other disadvantaged groups to secure and maintain paid employment in the open labor market" [99].

Both definitions incorporate the main values and criteria that have guided the development of SE programs in the past decades. They are as follows: (a) every individual, regardless of the severity or the type of disability, may aspire to a job in the community; (b) services have to respect self-determination and choices of person with disabilities; (c) wages and benefits have to be equal to that of co-workers performing the same or similar jobs; (d) job coaches have to focus their intervention on capacity and capabilities of the person with disability; (e) within the workplace mutual respect and acceptance have to be established between disabled and non-disabled workers; and (f) supports have to be provided before, during, and after the job placement [4, 94, 96].

The effective opening of the SE programs to all persons with disabilities has often been challenged. McInnes et al. [100] explored panel data of all clients served by South Carolina Department of Disabilities and Special Needs from 1999 to 2005 with a sophisticated statistical modeling in order to investigate the hypothesis that IQ and presence of emotional–behavioral problems may negatively affect the likelihood of entering SE programs. In other words, the authors supposed that the better outcomes of SE programs observed in many studies may be due to a selection bias. The authors concluded that even after correcting for these biases better statistical treatment effects of SE remain.

Depending on the mix of the support intensity, duration, timing, and characteris-

tics, scientific and professional literature has defined several SE practices. Therefore, for example, in customized employment programs the support is specifically finalized to the personalization of the relationship between employees and employers in ways that meet the needs of both. Customized employment assumes the provision of reasonable accommodations necessary for an individual to perform the functions of a job. In the United States, the Office of Disability Employment Policy provided a formal definition of customized employment, its characteristics, and a practical manual [101].

Individual Placement and Support (IPS) is an evidence-based variant of SE originally developed to help job search for people with severe mental illness. In IPS, the client's preferences are central. The client decides whether or not employers and potential employers know about her/his mental illness and whether or not the employment specialist will talk to employer/s on his or her behalf. The client also decides which jobs to apply for and how much he or she wants to work. Recently, IPS has been extended to people with autism [102].

SE programs have also assumed the form of targeted projects. The Treatment and Education of Autistic and Related Communication Handicapped Children (TEACCH) system, since the '90s, developed a specific version of SE for people with autism [103]. "Project SEARCH," also involving people with autism, is a recent and complex version of SE aiming to realize a seamless transition from school to work [78]. "Project SEARCH" can be described as a business-led transition model where schools create collaborative partnerships with local large businesses such as hospitals, banks, or government centers. Such businesses afford students in transition to adult life the opportunity to work in internships across their senior year of high school. The importance of the program consists of an integrated real-life work experience, training in employability, and independent living skills [104].

While in the United States, Australia, and Canada, SE programs have a clear legal foundation and an established organization, a recent study on SE promoted by the European Commission has revealed that in many of the investigated countries, funding of SE appears to be unstable and unpredictable. In particular, the risk is that the service model is not institutionalized and that it continues to have a pilot project character with a more limited impact [105, 106].

The cited European report has underlined that, for the majority of European states, unemployed people in general receive support through the public employment services while disabled jobseekers tend to be referred to external agencies [105]. Paradoxically, one of the most studied service models and one of the better documented in terms of outcomes is not yet completely and coherently recognized in European countries, as a first choice in accompanying people with IDD to employment. Also in most of the studied countries, reliable statistical data on SE programs and clients are not readily available [105].

Social Firm/Enterprise (SF): Like other textbooks and authors, we included SF within the framework of VR practice even if their main aim is the creation of new employment opportunities for disabled people. The Confederation of European Social Firms, Employment Initiatives and Social Cooperatives (CEFEC) underlines these issues defining the profile of a Social Firm [107]:

- It is a business which uses its market-oriented production of goods and services to pursue its social mission (more than 50% of its income should be derived from trade);
- A significant number of its employees will be people with a disability or other disadvantage in the labor market;
- Every worker is paid a market-rate wage or salary appropriate to the work, whatever their productive capacity;
- Work opportunities should be equal between disadvantaged and non-disadvantaged employees. All employees have the same employment rights and obligations.

Worldwide SF includes different business models under different labels. In Western Europe, most social enterprises operate under

the legal form of either a nonprofit association or a cooperative.

The general idea that guided the development of SF was to create completely new ways of meaningful production and working to find an answer to the problem of unemployment and discrimination in the labor market by increasing the available work opportunities for the most disadvantaged groups. The first social cooperatives were established in the late 1970s by mental health professionals and former psychiatric patients and they gave a decisive impulse to the psychiatric service reform promoted in Italy by Franco Basaglia [108]. The involvement of persons with disabilities in the foundation and management of the cooperative was a "provocative" choice and remains a model of participation with unchanged values in terms of empowerment and respect of disabled people's capacity and role.

The idea of creating employment as an alternative to traditional sheltered workshops contrasting the low employment opportunities in the private sector proved to be successful. As a result, the number of SF in Italy and, later, across other countries in Europe increased constantly. Currently, there is a wide variety of SF models across Europe with a different business focus, models, and approaches to support disabled and disadvantaged people. Many SF started as standalone business start-ups, mainly from the social sector, as vocational training projects, established by larger nonprofit organizations that manage a variety of supporting programs for disadvantaged and disabled people [109]. SF represent an important chance for employment, even if the scientific research on individual outcomes is quite limited [108].

Vocational Rehabilitation is an umbrella term to identify a large area of practices and services which enables persons to overcome barriers to employment or other useful occupations. In this frame, Supported, Sheltered Employment, and Social Firm are the main Vocational Rehabilitation modalities reported in the literature.

Tip

There is substantial (however, not definitive) evidence that sheltered employment programs have a low rate of successful transition to competitive employment and limited long-term impact on community integration.

38.4.5 Development of Work Skills

A broad research work has tried to give scientific evidence to practices and interventions whose aim was to teach relevant instrumental and relational work skills.

Dutta et al. [110] describe a rich and varied list of interventions commonly proposed by the US VR services, ranging from assessment practices to vocational rehabilitation counseling and guidance, from occupational/vocational training and basic academic remedial literacy training to disability-related, augmentative skills training, job placement, job search assistance, and on-the-job support. The authors show that job placement, on-the-job support, counseling and guidance, and miscellaneous training practice were significant predictors of employment for people with mental disability in a large population of people with disabilities coming from the US VR services [110].

Several studies investigated the role of in-school service variables in predicting employment of individuals with IDD [111–116]. Carter et al. [112] investigated factors associated with the early work experiences of adolescents with severe disability using data from the National Longitudinal Transition Study-2. Severity of disability had a stronger influence in reducing access to paid work. A strong role was also played by family expectations. Further, youths who were given more household

responsibilities had more than twice the odds of holding a paid job. Three prevocational experiences seemed to have had a positive impact on acquiring a paid work: career skill assessment, job skill training, and internship.

Mazzotti et al.'s systematic review [115], based on the study of Rowe et al. [116] in which 16 in-school service predictors of employment are identified, shows positive evidence for the majority of interventions investigated in the literature. Fifty-five significant positive effects across the 11 studies reviewed were detected ranging from $r = 0.19$ to $r = 0.934$ with a median effect of $r = 0.335$. Twenty-one relationship results in large effect sizes such as those for self-care/independent living skills, social skills, goal setting, and parent expectations.

The study of Holwerda et al. [114] confirm the strong influence of expectations on employment for people with IDD and give evidence that school teacher's expectation plays the main role. Park and Bouck [117] using a sample of people with ID from the National Longitudinal Transition Study-2 show that those who received vocational instruction, job readiness vocational services, instruction in looking for jobs, and placement support had a better chance of employment after high school. However, different effects were observed for the group with mild ID versus the group with moderate/severe ID. It is worth noting that when individual transition plans included competitive employment, the students were three times more likely to be in paid employment than other students, while the students who had sheltered employment plans were 0.17 times less likely to be in paid employment than other students.

Moving from macro-categories of interventions to specific interventions, the research landscape appears fragmented and the evaluation of effectiveness appears difficult to summarize. We present here only some interesting references. A large number of studies have focused on social skills training for people with ASD, and they have reported positive results on employment outcomes [118]. The TEACCH-supported employment program was developed using a structured ASD-specific visual supporting approach. A report about 96 clients who received TEACCH-supported employment services show good hourly wages and job retention rate (89%) which was not related to clients' status at referral [103, 119]. Training oriented to enhance conversational skills [120], emotion understanding [121], perspective-taking [122], utilization of sight words in self-instruction [123], and acquisition of self-help skills [124] seem to have some effect in increasing the likelihood of obtaining an occupation.

During the last 10 years, we have witnessed the development and application in the VR field of technologies and techniques used for different training purposes, such as tactile prompting, picture guiding, audio cuing, video modeling, computer-aided instruction, virtual reality, and robotics, implemented mainly among individuals with ASD [125]. The use of personal digital assistant reduced the number of hours of job coaching and increased the number of hours worked in a supported environment [126]. A multimedia program was found to improve interviewing skills and rates of attaining competitive employment [127, 128]; touch screen tablet computers increased independence, confidence, time management, and organizational skills at work [129]. These studies have provided support for the use of behavioral techniques to teach workplace skills and modify behavior in individuals with ID and ASD [130]. Other studies have described interventions conducted by video modeling or audio coaching [131–134] and behavior training on different skills and tasks using smartphone applications [131].

Lattimore et al. [135] evaluated a multiple-stimulus assessment for identifying work preferences among adults with autism prior to beginning supported jobs. Opportunities to work on preferred tasks can enhance job performance as well as overall quality of work life and is a specific requirement of the US-supported employment legislation.

Lattimore et al. [136] showed a substantial increase in independent job performance with newly assigned tasks when simulation training was provided prior to on-the-job training. Wehman et al. [137] investigated the effect of an enhanced version of Project

SEARCH in which the structure and intensity of the learning experiences were increased by the use of applied behavioral analysis (ABA) techniques. A treatment (n = 38) and control group (n = 18) of youth aged 18–21 years with ASD were compared on employment outcomes with 49 non-ASD youths (18–21 years). The results of the study demonstrated the effectiveness of the supported employment practice in producing positive employment outcomes and which reduced significantly the support intensity needs (as measured through the Supports Intensity Scale; SIS) [138] in employment activities. At 3 months post-graduation, 90% of the treatment group acquired competitive part-time employment earning, compared with 6% of not-in treatment conditions; 87% of those individuals maintained employment at 12 months post-graduation compared with 12% of the control group. The study provided also a detailed description of project activities connected with Project SEARCH guidelines, method used, and team leader identification.

In a recent systematic review of interventions on adults with ASD, Headley et al. [118] postulated that despite the availability of many studies there are serious problems in generalizing results due to the large range of skills and interventions investigated and the small number of individuals involved in these studies.

The literature on Vocational Rehabilitation describes a rich and varied list of interventions commonly proposed by services. It also underlines the importance of prevocational experiences such as evaluation of professional skills, professional training, internships, and in particular competitive work experiences. Research findings also describe the important role that self-care and independent living skills, social skills, self-determination, and parents' "and teachers'" expectations have on the likelihood of getting a job.

Tip

Future researches should ascertain the promising impact of TEACCH and ABA-based techniques on supported employment programs as well as the usefulness of new technologies for training purpose in the Vocational Rehabilitation field.

38.5 Employment-Related Outcomes

As we stated previously in this chapter, the majority of authors consider employment as a fundamental social goal with great value for every human being. Competitive employment promotes the sense of belonging to society and positively affects the quality of life of people with IDD [139]. For this reason, placement rates, the duration of employment, earnings, and job tenure are often considered main outcomes in themselves [140].

However, researchers consistently believe that the effect of employment on quality of life (QoL) and other non-working outcomes have the same importance and require to be carefully, empirically investigated. It is worth remembering that QoL's objective indicators correspond to condition of individual's life as it appears to an external observer, considering that there are aspects of life that have a qualitative value for everyone. The subjective dimension corresponds instead to the individual perception of satisfaction for existence or for a specific life domain and can be assessed only through the opinion of the person himself. Both dimensions can be considered fundamental components of any QoL concept (see ▶ Chap. 15).

Relatively few studies have explored the impact of employment on QoL and subjective well-being of people with ID and ASD [141, 142]. In the study of Kober and Eggleton [143], 117 people with ID were interviewed using the QoL questionnaire [144]. The results of this study indicated that people with ID in open employment report a better QoL than those in sheltered employment. In particular, open employment correlated with a greater sense of social belonging and empowerment.

Community-integrated employees also reported a greater level of QoL in the domains of health, productivity, and emotional well-being than those involved in less inclusive activities [92]. Jahoda and colleagues [145] reported a systematic review of 15 years of studies on QoL, social life, and autonomy of people with ID related to the employment experience. Fifteen studies were selected. Assessment instruments documented in the review appeared heterogeneous as well as research designs (in many cases no control groups were selected) and sample sizes. Entering employment seems to produce positive outcomes for all the main dimensions investigated. However, the authors observed that the opportunity that work experience offers to increment social relationship and interactions with peers and co-workers did not seem to translate into a sense of belonging or reciprocal relationship.

A recent systematic review supports the idea that the positive effect of paid employment on physical or mental health is similar for adults with and without IDD [146].

Lack of social acceptance remains a prevailing concern for people with ID. Service providers should place more focus on assisting their clients and find placements that would provide them with adequate social and emotional support. In a recent study by Blink et al. [147], the QoL of 447 adults with ID was evaluated with Essential Data Elements, an 85-item comprehensive, structured interview and compared among three groups involved in three different settings: open employment, sheltered workshops, and adult day care centers. Participants in community-integrated employment indicated a higher sense of community inclusion and reported more opportunities of choice and control on several dimensions of their life and financial autonomy, compared with participants of sheltered workshops and adult day care centers [148].

A dimension closely linked to the concept of subjective well-being is "job satisfaction." It can be defined as "a pleasure or positive emotional state resulting from the appraisal of one's job" [149]. It is basically defined by the same constructs as subjective well-being (see ▶ Chap. 14), whereas the affective part addresses either negative or positive affect and the cognitive part refers to an individual's global evaluation of their satisfaction with their job. It is the result of both situational work environments and personal characteristics and of the interaction between them [150, 151]. We do not have a satisfactory review of the studies in this area of research. However, the existing literature indicates crucial aspects of job satisfaction of people with ID such as the nature of their work, working conditions, financial benefits, growth opportunities, and self-determination [92, 152–155]. Examining the subjective experiences of a group of people with ID, who were starting employment, Jahoda et al. [156] found that employment helped people with ID to "get out of their house" and to lead a more active life. At the same time, work may cause anxiety, especially at the beginning of the job experience.

A recent study analyzed, adopting "Photovoice" method [157], the job satisfaction of nine employees with ID in integrated and sheltered employment [158]. The authors considered nine domains of job satisfaction: (a) the nature of work itself, (b) working conditions, (c) experienced job demands, (d) social relations at work, (e) received support, (f) perceived autonomy, (g) opportunities for using competencies, (h) opportunities for growth, and (i) development and meaningfulness. The results indicated that people with ID appeared to consider most of the same topics relevant to job satisfaction as do people without ID. However, some domains may be more distinctive for workers with ID such as received support, opportunities for using competencies, perceived autonomy, and opportunities for growth and development. These data showed that people with ID seemed to be aware of their abilities and limits as a natural aspect of their own working life. On the other hand, the sense of autonomy and growth showed a desire of these individuals to become more capable and independent.

A systematic review of employment programs and intervention outcomes targeting

adults with ASD dedicated a section to non-employment-related outcomes [14]. Only eight studies corresponded to the selection criteria. The examined outcomes were heterogeneous, ranging from examining the effect of supported versus sheltered employment on core ASD symptoms, QoL, and cognitive skills to evaluating the impact of specific interventions in improving social behavior and social communication, literacy, or reduction of challenging behavior in the workplace. The number of subjects involved was quite small, and the heterogeneity of the evaluated outcomes made generalization difficult. Furthermore, the authors concluded that the few studies investigating outcomes unrelated to employment were generally of poor quality so that, to date, evidence is lacking, in terms of outcomes beyond obtaining employment, such as cognitive, mental health and well-being, symptom severity, adaptive functioning, and quality-of-life factors.

Social inclusion as an outcome of work experience has been relatively poorly investigated in the literature. As mentioned in the introduction, the employment itself is considered an adequate indicator of social inclusion. In their scoping review on inclusion as a focus of employment-related outcomes, Lysaght and colleagues [159] found that no more than five articles focused on central aspects of inclusion, such as sense of belonging, reciprocity, and fulfillment needs. This area of research should receive more attention.

A useful descriptive framework oriented to a joint analysis of the effects of employment on QoL [160], social and economic engagement [161], and social inclusion [162] has been proposed in a recent comparative study on the perceptions of a sample of 51 people with ID involved in three different types of employment, namely, open employment, SE (the Australian term corresponding to sheltered workshops in the United States and Europe), and social enterprises. The authors found that across all employment types, relationships and getting paid or achieving material well-being were the most important employment outcomes to people with ID. Skill development and participation were the next most important outcomes. People working in open employment appeared to have the most knowledge about rights, respect, and equity.

Emerson and colleagues [163] investigated the influence of employment on health status of persons with ID analyzing a population-based cohort from which a proxy measure of childhood IQ was derived. Despite people with ID and borderline intellectual functioning had lower employment rates and poorer health than other participants at all waves of data collection, those in full-time employment showed a better health status than those in part-time employment or inactive, and those in any kind of employment showed a better health status than those unemployed.

- Most common employment-related outcomes are represented by placement rates, length of employment, earning, and job tenure, while little consideration has been given to person-centered outcomes.

- Available literature reports that persons working in the open labor market show higher quality of life, job satisfaction, and community inclusion than those in sheltered employment.

Tip

Employment has been commonly regarded as an indicator of social inclusion; hence, research on the impact of employment on community inclusion is limited, especially in reference to persons with ID/ASD. It is to be hoped that this gap can be filled in future studies.

38.6 Employability: Workplace Characteristics and Employers' Attitude

As already noted employability is modulated by a complex set of "external" factors, that is, not directly related to the person, among which McQuaid and Lindsday [63] consider

labor market factors, macroeconomic factors, recruitment factors (including employer and colleagues' attitude, required skills, and workplace characteristics), employment policy factors (including availability of vocational services, employment agencies, and policies), and other enabling policies such as accessibility and affordability of public transport, child care, and other support services. We will briefly address some of these issues.

38.6.1 Access to Work for Persons with Disabilities: Legal Protection and Reasonable Accommodation

A long tradition of research and practice, with its roots in the 1970s, has underlined the crucial role of accessibility and workplace adaptation in facilitating employment of disabled people. The general goal was to overcome, as much as possible, the disability effects and to enable the disabled person to participate in the labor market. Some of these interventions regarding a barrier-free design and adaptation of workplace, barrier-free access to communication, the provision of facilities, and means of transportation (bus, train, aircraft) may be crucial for people with ID.

Hendricks [164], reviewing a rich literature on the topic, has provided a list of workplace modifications commonly needed by persons with ASD, such as noise reduction, avoiding crowding, adjusting lighting, and facilitating space navigation. "The workers with autism benefit from clearly defined work tasks and, also by providing a consistent schedule for completion of work tasks, job predictability can be increased. Idle time in the workplace may prove to be problematic and also requires restructuring" [165].

However, to facilitate employment of people with IDD, a wider set of other opportunities ranging from education and professional training to hiring practices, customizations, or restructuring of job tasks are required [165]. Special employment protection rights like disability quotas [166, 167] or additional allowances and benefits, such as tax relief [168], are common in many countries. Over the years, cumulative practices and research are offered to professionals and people with disabilities' guidelines to access services and benefits and/or tools for the analysis and evaluation of the needed workplace adaptations [169, 170].

Article 27 of the UNCRPD summarizes 4 macro-principles, widely recognized and shared in international literature, in national legal frameworks and embedded in service practice:

(a) Non-discrimination, as the right to work on an equal basis with others;
(b) Accessibility, as the right of living in a work environment that is accessible, identifying, and removing barriers that hinder persons with disabilities from carrying out their work on an equal basis with others;
(c) Reasonable accommodation, as a necessary and appropriate modification and adjustments not imposing a disproportionate or undue burden were needed in a particular case, to ensure to persons with disabilities the enjoyment or exercise on an equal basis with others of all human rights and fundamental freedoms;
(d) Adoption of positive measures to promote employment besides a duty to impose obligations on private- and public-sector employers [8].

In the last 50 years, laws and regulations at national level have granted rights and benefits to disabled people. One of the first and most cited is the American with Disability Act (ADA) [171].

A recent report analyzing legal developments at UN and EU level (the 28 EU Member States, Iceland, Liechtenstein, and Norway) has shown that "all the countries, with the exception of Liechtenstein and Iceland, have implemented non-discrimination and reasonable accommodation duties, although a range of different approaches are taken to the types of legislation in which this duty is located. Barriers to enforcement are often associated with the costs of the proceedings, access to legal assistance, and advice and procedural issues such as time limits for bringing actions. In addition, disabled claimants sometimes

encounter barriers in the form of inaccessible court (or other) buildings and inaccessible communication or information" [172]. The report has also revealed a lack of data about the number and nature of cases on reasonable accommodation, in most countries.

In this regard, an interesting source of information is the US-Integrated Mission System of the Equal Employment Opportunity Commission for allegations of discrimination involving Title I of the ADA. Van Wiren et al. [171] analyzed all US-resolved allegations from 1992 and 2008 and found that only 1626 involved people with ID (1459) and ASD. By comparison, the same database contained 45,560 allegations for back impairments, 14,515 for diabetes, and 11,421 for hearing impairment. The small number of allegations involving people with ID and ASD may be related to obstacles to both recognizing and reporting instances as well as to the low presence of ID and ASD in the labor market. As far as we know, there are no comparable data available for other countries.

The UNCRPD includes four principles that have been widely recognized in the international literature as external factors that can considerably facilitate employment of people with ID/ASD: non-discrimination, accessibility, reasonable accommodation, and adoption of positive measures to promote employment.

38.6.2 Employer Attitude and Practice

The definition of a legal framework has to be supported by an effective capacity to promote the rights of persons with disabilities by all the involved subjects and first of all by employers, managers, and work colleagues. It seems strategic that workers with ID and ASD were recognized as a valuable addition to the workforce and not included on the basis of solidarity principles or corporate social responsibility only [173, 174]. A recent scoping review of the research literature around employer responses to disability in the workplace found that the number of studies on the topic has increased significantly since 1991 [175]. However, Ellenkamp et al. [10] in their systematic review on work-related factors of employment found no more than 26 articles in the last 20 years specifically related to ID. Most of the selected studies in this review were conducted in the United States. While employers' surveys have generally found that employers express positive attitude toward the idea of hiring people with disabilities, these findings do not seem to translate to real-world behaviors, perhaps because respondents to such surveys tend to give socially desirable answers [176].

There is strong evidence that in the workplace employees with ID were confronted with misconceptions about their capacities. Underestimation and negative attitude of colleagues seemed a relevant issue [177]. A study of workers with ID in competitive employment has revealed that "awareness" of stigma themes was found in almost half of the interviewees [10]. Comparing people with different types of disabilities, several studies have shown that employers' attitude is the least favorable for people with ID and mental disorders [178, 179].

However, employers who had experience with people with IDD had a more positive attitude and lesser stereotyping and disengagement attitude toward these employees compared with employers without such experience [10]. The importance of engaging employers throughout the process of creation of inclusive practices and policies with work environments is critical to the successful employment, career navigation and economic parity, workplace accommodation, job restructuring, and job customization [180].

Expanding and improving relationship between employers and VR services may create a great focus on work participation of employees with ID, but as noted by Luecking [181], employment service providers do not speak the same language as employers, and also disability employment service providers and employers use different success metrics. The author strongly supported the idea of a dual customer approach, that is, the employer is simultaneously targeted as an end user of

employment service program while the program supports the job-seeking individual.

A well-known initiative trying to develop a sharing language and vision between vocational service and employers is the already cited Project SEARCH [104, 182]. However, Luecking [181] supported the idea that a close relationship between employers and employment services could be established in an ordinary way without the "proliferation of special programs."

It is worth noting that in the majority of the 26 studies reviewed by Ellenkamp and colleagues [10], the employees' perspective was not included. The authors suggested that "self-advocacy of employees with ID supported by job coaches or families in promoting dedication to their work in combination with disability educations for employers and colleagues could be helpful in generating ID awareness by employers and reducing underestimation of capacities of workers with ID" [10].

There is strong evidence that employees with ID/ASD, especially those with co-occurrence of mental disorders, are exposed to prejudice about their capacities, negative attitude, and discrimination by colleagues and employers.

Key Points

- We have tried to synthesize a huge and complex literature, and this complexity does derive from the intrinsic nature of the matter. Accompanying people with IDD to employment is an interdisciplinary work, which requires specialized skills on the medical, rehabilitation, educational, social, and legal levels. Job placement takes a long time and involves a number of institutions: schools, health and social services, vocational services, and companies. Generating employment opportunity for people with IDD would require overcoming stereotypes and stigmatization, compliance with anti-discrimination rules, and a strong alliance with employers. Kirsh et al. [176] in an interesting summary article "of the lessons learned" in decades of research and fieldwork have identified the key prerequisites for advancing research and social changes in the field of work integration: cultural, legal, technical aspects, professional, and organizations should find full synergy and an operational synthesis. However, this scenario is far from being accomplished even in countries with a more consolidated and rich welfare system.
- Despite the different research approaches and the difficulties in interpreting and comparing data from different social and cultural contexts, there are some convergent empirical evidence in employment trends of people with IDD. Labor force participation and employment rates remain at the lowest levels among the different groups of people with disabilities in all countries for which data are available. There is some evidence that among the group of developmental disabilities people with autism show lowest levels of labor force participation and employment rates. Hourly wage and total earnings of people with developmental disabilities are approximately half of those of persons without disabilities and lower than other people with disabilities.
- Despite the different definitions (diagnosis, medications, emotional problems, depression, etc.), the large and mixed set of psychological variables considered, the difference in statistical measurement of adaptive behavior and behavioral problems, mental health problems seem to reduce employment rates and increase probability of unemployment and participation into sheltered workshops. However, the effect of comorbidities and severity of disability on employment rates and labor force participation at population level is only partially documented. In particular, the representation of the psy-

chopathological conditions appears inadequate; often, diagnostic categories come from administrative documentation and are not clinically confirmed; in other cases, we can observe only proxy variables. To our knowledge, no study demonstrated to respond to stringent quality criteria for diagnosis [34].

- The number and variety of available studies testify to a notable interest and commitment of the international scientific community on the issues related to the employment of people with IDD. Nevertheless, the heterogeneity in the assessment of clinical conditions and severity of disability, associated with small samples investigated and methodological limitations in research design, make it difficult to generalize and compare outcomes of different employment programs. To our knowledge, the literature reports only one experimental study and no meta-analysis.
- However available evidence suggests that supported employment is the most effective practice in improving employment rate and significant employment-related outcomes relevant for people with IDD. The positive impact of employment on quality of life and subjective well-being of persons with IDD is relatively well documented in literature and particularly in the case of supported employment programs. Many specific aspects of employment-related outcome need to be better investigated, for example, the impact on functioning profiles. The heterogeneity of the evaluation tools used and limited data comparability remain two of the main problems in this crucial field of research.
- There is worldwide evidence of no significant improvement in the IDD employment rates at population level, at least in the last 10 years. Labor force participation and employment rates of people with developmental disabilities seem higher in North America and Australia than in Europe, but this may be partly due to the lack of reliable data on sheltered work in European countries and differences in definition of target population. Access to supported employment services is not guaranteed to all persons with IDD, and the level of funding has been substantially stagnant in the last 10 years, both in Europe and in the United States. However, the absence of significant increases in access to work for people with IDD should not be interpreted only as a failure.
- The labor markets are undergoing extraordinary changes that seem to move toward reducing occupations suitable for people with IDD but also for non-disabled workers. Even without following the catastrophic prophecies of Rifkin about the "end of work" [183], it is impossible not to be worried about the future. The "aseptic" OECD employment outlooks underline the role that technological progress and globalization are playing in polarization and de-industrialization "both associated with disruption in workers' lives and rising inequality" [184]. Reflecting the changes in organizational structure of companies, Chan [173] has observed that: "the organizational chart is now flatter and team-based and the emphasis is on flexibility, productivity, and workplace socialization skills." It seems clear that people with ID are facing tremendous and growing challenges for access to work. These challenges extend to employment services. Chan argues that "… therefore rehabilitation professionals must have a thorough understanding of the real concerns of employers about the hiring and retention of persons with disabilities and needs. Helping people with disabilities develop flexible, versatile, and adaptable work skills to meet employer expectations and requirements for jobs in the

new economy will improve their odds of obtaining employment" [173].

- The effort focused on enabling personal skills and abilities is not enough. It has to be accompanied by a parallel effort in creating new employment opportunities and perhaps the culturally dominant idea of work has to be reinvented as well [185]. International data and research suggest some possible tracks. Public campaigns, legal frameworks, employment benefits, and services seem to be targeted to big companies. The potential of small firms for the inclusion of people with IDD has to be largely explored [186]. Additional effort should be dedicated to initiative for promoting self-employment and microenterprises led by disabled people [187–189]. A policy of support of SF may also open new employment perspective [1, 190].
- Dealing with work for people with IDD has also a political value. If people with IDD can find work in the future, opportunities and hopes will also open for many non-disabled people. The complex sector of institutions and interventions, we have described so far, can also be seen as a great cultural, social, and political "laboratory" at the service not only of people with disabilities but of great utility for the whole society.

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Cultural Issues

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Contents

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Learning Objectives

- Cultural issues related to the lives of people with intellectual disability (ID) and Autism Spectrum Disorder (ASD), at the policy, service, and clinical levels.
- Impact of these cultural issues on services and programs as well as implications for care and interaction with individuals and families.
- Organizational culture and its impact on programs and services.
- Current evidence-based screening and diagnostic procedures and tools in different countries.
- Availability of services that facilitate independent living and community integration for people with ID and ASD in many cultural contexts.
- Variations between how informal caregiving are provided.
- Indications for better policies, practices, and future research.

39.1 Introduction

Culture permeates the context in which people live and many contextual factors are closely related to culture (in fact, culture itself is a contextual factor). Both constructs—culture and context—are important frameworks for understanding personal and environmental factors that influence human functioning and quality of life, for supports planning and for public policy development and improvement. Therefore, in this chapter we will discuss both cultural and contextual issues that have a major impact on lives of persons with intellectual disability (ID) and Autism Spectrum Disorder (ASD) and explore how these elements constitute frameworks for providing high-quality services and supports as well as developing public policies. In addition, we will review how certain cultural practices and issues can lead to stigma and abuse, or low-quality services.

Although the concept of culture has been frequently mentioned as an important issue in terms of research about services for people with ID and ASD—in terms of measurement of prevalence, detection and diagnosis to some extent, and support services including especially residential or day programs—a variety of definitions of culture have been used and in many instances culture has been mentioned but not actually taken into account in analysis, evaluation, or planning. Bigby et al. [1] developed a definition of culture that is useful for evaluating and improving support services (including residential and day programs) for people with ID and ASD. In addition to a formal culture as endorsed by an overarching institution that provides administrative indications and proposes the model of care to be implemented, there is also an informal culture that is expressed through what care professionals and teams do in their day-to-day activities in order to implement this model of care (or often, not implement or at least not implement entirely) [1]. These authors adapt a definition proposed by Schein (1992) which states that culture is "A pattern of shared basic assumptions that the group learned as it solved its problems of external adaption and internal integration, that has worked well enough to be considered valid and, therefore, to be taught to new members as the correct way to perceive, think, and feel in relation to those problems" (Schein 1992, p. 12, cited in Bigby et al. [1]). Adapting Schein's ideas, Bigby et al. [1] posit three levels of culture: artifacts (visible organizational structures and processes); espoused values (strategies, goals, philosophies or espoused justifications); and basic underlying beliefs, perceptions, and feelings, which are unconscious or taken-for-granted, for example, "local" common sense among the staff.

As with the construct of culture, the term context is often referred to in relation to ID and ASD but a consensus on the definition is lacking. According to Shogren et al. [2], "Context is a concept that integrates the totality of circumstances that comprise the milieu of human life and human functioning" and incorporates "factors that affect both positively and negatively human functioning." Contextual factors include culture and related aspects such as ethnicity and place of residence (including institutional or family, rural or urban, or specific country), among others.

In order to understand why within Shogren, et al.'s [2] definition of context, human functioning is so central, one needs to consider how definition and measures of disability, and the organized social response to them have changed in recent decades [3, 4]. An important engine behind this transition has been the political organization of people with disabilities themselves [5, 6]. While previously disability was defined as an individual characteristic or condition that was usually perceived to be a deficit, this construct has been developed into "an interactive human phenomenon with its genesis in organic and/or social factors" [2]. It is both the organic and the social factors that lead to functional limitations, always depending on the context (physical and social). Thus, functioning limitations of persons with ID or ASD cannot be attributed only to an individual impairment but to the complex relationship between this impairment, the impaired person, and the whole context (especially supports provided) in which the person lives. This makes it easier to visualize the context as "an intervening variable that can influence human functioning through organization and systems-level policies and practices, and public policies" [2].

Just as the definition of disability has changed at the theoretical level, in terms of policy this has been intertwined with the tendency in some countries to move away from institutionalization of people with disabilities and toward greater inclusion, in schools, living accommodations and communities, and employment [5, 6]. Thus, policies in some places have transitioned from an individual, medically focused perspective to a social approach that recognizes the contextual factors that can disable people (for example, make them unable to use transportation, move through the community, or receive information) [5, 6].

In summary, these definitions of disability, culture, and context are useful as frameworks for thinking about and understanding ID and ASD in order to take into account the impact of culture [1] and contextual factors [2] on human functioning. They can also act as tools to evaluate and improve the outcomes achieved by support systems and other services and care for people with ID and ASD.

Culture and contextual factors are interrelated fundamental frameworks for a correct conceptualization of ID and ASD as well as for adequate provision and planning of mental health services. Implications on grading of individual functioning impairment, support needs, and quality of life are also relevant.

39.2 Diagnosis and Services for People with Intellectual Disability and Autism Spectrum Disorder in Different Cultures and Contexts

In many middle- and lower-income countries, effective models of primary care with broad coverage are often lacking for health issues in general and especially for mental health [7]. Professional competencies in mental health are rarely strengthened. These issues, as well as cultural and other variables, lead to a lack of screening and early identification of neurodevelopmental disorders [7]. A clinical perspective focusing on the individual tends to predominate, while a focus on the family as well as cultural and social dimensions and the need for broad screening is lacking. In general, a public health approach to mental health for ID/ASD and an appropriate consideration of cultural and social issues (at the conceptual or practical levels) are absent. This includes educational, economic, and employment issues. This public health and sociocultural focus, and specifically providing broad screening, is necessary in order to maximize equity and social justice.

Implementation of population-level or large-scale mental health interventions is highly complex. In addition to this, there is an extremely low level of spending on mental health in almost all middle- and lower-income countries. This reflects both the existence of many other priorities in health care and a lack of recognition of the importance of mental health issues. The low level of spending on mental health is often compounded by the tendency to invest all funds in the operation

of psychiatric hospitals and treatment for addictions. This reflects a generalized model of care that focuses on curative medicine instead of prevention and what in the field of ID and ASD has often been called rehabilitation as well as support systems. Also, the number of psychiatrists in relation to the population is often extremely low in middle- and lower-income countries. For example, while Switzerland has close to 44 psychiatrists per 100,000 inhabitants, Mexico has only 3.68, and the proportion of child psychiatrists is often lower, such as in Mexico where there are only 0.69 per 100,000 inhabitants under the age of 15 years. Similarly in Pakistan, there are less than ten child psychiatrists for about 100 million children and adolescents.

Likewise, in many lower-income countries, overwhelmingly in public health care and to some extent in private care, current, evidence-based tests for diagnosing ID or ASD are not available. This is in great part because many entail a high cost. At the clinical level, many diagnostic tests for ID and ASD have been adapted to different languages and cultures. This has often occurred through formal procedures, although other times (perhaps especially initially, before formal adaptation has taken place) there has been informal adaptation and use in clinical practice. However, as mentioned, in a variety of contexts the cost of using current, high-quality diagnostic tests or scales are prohibitive and so older versions or in many cases non-evidence-based diagnostic tools are used.

In many middle- and lower-income countries, a public health approach to mental health for ID/ASD is often lacking, especially in reference to preventive interventions. The number of psychiatrists in relation to the population is often insufficient.

These and other sociocultural and contextual factors lead to difficulties and delays in the identification of persons with ID and ASD and in the implementation of adequate management. Up-to-date tests for diagnosing ID or ASD are not available, because their cost is prohibitive. Even guidelines for diagnosis and treatment are often inappropriate and partial.

Studies in Africa suggest that ASD is rarely diagnosed except in the presence of ID, indicating the likelihood that only the most severe spectrum of ASD presents to orthodox practice in Africa [8]. It has also been suggested that high comorbidity of ASD and ID among sub-Saharan African children may be due to the fact that onset of ASD coincides with a period of vulnerability of children in African contexts to neurological infections and complications like cerebral malaria, meningitis, and kernicterus, among others [9]. So, comorbidity of ASD and ID is very common among sub-Saharan African children [9], and ID co-occurs with other neurodevelopmental disorders [10], likely due mostly to sociocultural and contextual reasons.

Assessment of ID in Africa, as regards to Intelligence Quotient (IQ) score, presents a peculiar problem because of the influence of wide sociocultural variations across cultural and ethnic groups in the region, with a tremendous impact on child-rearing practices in different African communities, which may affect early child development. Given this, standard Western intelligence scales or assessment instruments such as the Wechsler Intelligence Scale for Children (WISC) have shown limitations for use in most African settings. This is because of variations in expectations for and levels of early childhood development in many African communities, which are influenced by a variety of sociocultural practices. Therefore, often an adaptive or modified method of IQ score assessment is employed in sub-Saharan Africa [11].

A substantial proportion of the population residing in African countries (close to 50% and with even higher levels in rural communities) attribute the etiology or origin of ASD and ID to spiritual causes, such as witchcraft, possession by evil spirits, a sinful act, and ancestral curses, among others [12, 13]. These aetiological explanations, which may also be held by caregivers or parents of children with ASD and ID, tend to influence perceived or lived stigma as well as help-seeking behavior [13]. A majority of caregivers or parents of children with ASD and ID in African societies experience frequent stigma, ranging from derogatory comments from

neighbors to outright social exclusion. Many of these parents are ashamed to let members of their communities know that they have children with ASD or ID [9, 13]. Consequently, many of these children are repeatedly locked into their homes and thus deprived of necessary early interventions or eventually any services or care (as well as, obviously, social inclusion, and deprivation of basic rights), given the generalized and severe stigma associated with their condition and the lack of policies to combat it [9, 13, 14].

Because aetiological explanations and other cultural issues greatly influence help-seeking behavior, many children with ASD and ID in Africa have local traditional and religious healers as their first points of contact in the help-seeking process. Local traditional healing settings may offer some form of psychological support to parents and their children with these conditions. However, practices in these settings often lead to late presentation at formal healthcare support, depriving such children of necessary early interventions. Religious healers consist mostly of Alfas and Pastors in Pentecostal Churches, who promise spiritual healing and very often employ exorcism. Religious healers may provide some psychological support, but at the same time they give false hopes of a cure, which often leads to delay in presentation at formal healthcare settings where evidence-based care might be provided [8, 9]. Collaborative work between clinical healthcare providers, public health institutions, government entities, and traditional and religious healers is needed to diminish stigma and promote help seeking by families both from traditional or religious healers and formal (clinical) health care. Healthcare providers need to receive training on acceptance of cultural beliefs held by parents and other caregivers, in order to provide empathic care, which is more acceptable to the parents. Also, these traditional beliefs do not necessarily conflict with clinical etiology [13].

In addition to cultural practices which can have a negative impact on help seeking, a number of other factors, mentioned below, have been identified as being responsible for the late presentation and diagnosis of African and Asian children with ASD and ID in formal healthcare settings [12]:

- Inadequate healthcare facilities and trained personnel for interventions.
- Social, cultural, and economic obstacles in pathways to care/help-seeking behavior.
- Poor knowledge and awareness about ASD and ID.
- Stigma manifested in negative cultural attitudes, beliefs, and practices toward ASD and ID.

There is also a lack of implementable social policy in many African countries. Laws and social policies that are enabling and needed to enhance service provision for persons with ASD and ID are often lacking in African countries. The absence of such laws and social policies makes person with disabilities even more vulnerable to human rights abuse and leads to deprivation of services like medical care and education [15]. The possible areas of human rights abuse common among individuals with ID and ASD in Africa include physical, psychological, and sexual abuse, the last of which tends to be more common in individuals of female gender [15]. Many parents in Africa are often subjected to exorbitant, out-of-pocket payments for social and rehabilitation services (both medical and educational) for children with ASD and ID. Thus, access to education, especially in independent living and other opportunities to learn skills for children, adolescents, and adults with ID and/or ASD is limited in most African societies.

In Africa, the absence of protective laws and social policies makes persons with ID/ASD highly vulnerable to human rights abuse and leads to deprivation of services like medical care and education. A substantial proportion of the population residing in African countries attribute the etiology or origin of ASD and ID to spiritual causes, and this tends to influence perceived or lived stigma as well as help-seeking behavior. Other factors, as inadequate healthcare facilities and trained personnel for interventions, economic

obstacles, and stigma, are responsible for the late presentation and diagnosis of African and Asian children with ID/ASD.

As with Africa in general, in Latin America there is a lack of early detection and referral of children with ASD and ID. A Latin American Guide for Psychiatric Diagnosis exists which is approved by the Latin American Psychiatric Association. However, this guide tends to lead to diagnosis of severe cases only and, therefore, to under-registration and misclassification of people with mild to moderate ID. Nor does the guide offer general norms for diagnosis or criteria appropriate for different age groups. In general, diagnostic guides produced in Latin America are lacking in criteria for diagnosing different levels of ID severity or ASD support needs. Some lack clear references to ASD, while others include tools or standards that are very out of date. A Latin American guide that is up-to-date based on local and international expertise and clinical research is needed for the region [16].

Although in general Latin American countries do not have policies that specifically seek to segregate and institutionalize people with ID or ASD, the lack of a culture (and accompanying policies) of respect for human rights, inclusion, and non-discrimination often leads to social exclusion. The social exclusion that people with ID or ASD experience in Latin America has three components: economic deprivation, sociocultural aspects, and political issues. In terms of economic deprivation, in Latin America, people with ID or ASD often have no employment or irregular, very low paid employment with no job security, and so have insufficient or no income. Social deprivation can imply isolation, segregation, and weakened or ruptured family and social ties due to institutionalization or because their families hide them due to discrimination, stigma, and the complete lack of opportunities for cultural, social, and community participation. Most people with ID or ASD in Latin America have little or no control over their daily lives nor do they have political representation, even through proxies (such as their families or guardians). Their social exclusion and lack of opportunities for employment lead to greater poverty for them and their families. This exclusion and segregation is the result of the interaction between their condition, local cultures of discrimination, and the incapacity of social institutions to provide access to education, employment, health care, and public services in general. Cultural, social, and political processes must be instigated and supported in Latin America that will lead to universal access to health care and other services. Also needed is professionalization of healthcare personnel who work with people with ID or ASD in Latin America, including clear definitions of the competencies they must achieve and the diverse training curricula as well as promotion of psychiatry and associated specializations as careers [16].

High-quality measurement of the prevalence of ID in children, adolescents, and adults in Mexico is lacking, and only very recently have improved tools been applied for a better population-level measurement in children and adolescents. Therefore, the magnitude of this public health problem is unknown. At the clinical level, the presence of ID and ASD at all ages is underestimated, due to the use of inadequate diagnostic tools and lack of clinical competencies. Specialized (tertiary) public hospitals provide diagnostic services to a limited proportion of the population with treatment, therapies, and rehabilitation focused almost exclusively on children. In terms of health policies related to ID and/or ASD in Mexico, there is a scarcity of adequately trained human resources. Guidelines for prevention, early diagnosis, and management are lacking as well. Once people with ID or ASD become adults, if they are living in a public residential institution they are expelled when they reach age 18 and there is a complete lack of governmental health infrastructure or social services available to them. In summary, the Mexican State is basically absent in terms of public policies for people with ID or ASD. The most important element of this context is perhaps the lack of an institutional model focusing on independent living, social inclusion, and employment. In private care, there are a few exceptions, but in public services this type of approach is absent.

Nongovernmental organizations fill in some of the gaps, but most of these organiza-

tions only serve those under 18 years of age and their work is often not based on scientific evidence. Also, given lack of resources, the interventions offered by nongovernmental organizations are local and have very limited coverage. Most programs developed by such organizations in Mexico are the result of anecdotal, nonscientific evidence and fail to achieve the desired outcomes in the disabled population. They have also been unsuccessful in altering public policies and opinions, which would allow for greater reach of programs and services (this would only be appropriate when services are evidence-based and undergo rigorous evaluation). Above all, the topic of autonomy or independent living and promotion of social inclusion and employment for people with ID or ASD tends to be absent in these programs.

Education policies for children with ID implemented in Mexico have focused on the creation of schools for special education as well as on the integration of children and adolescents with ID in general primary education. Although these strategies are useful for promoting diversity in the classroom, the skills taught are academic and not useful for facilitating independent living. In addition, specialized teacher training is either nonexistent or inefficient in providing teachers with skills for meeting the needs of students with ID or ASD, particularly when attempting to use them in a classroom integration model.

The situation in Argentina is somewhat different from other countries in Latin America since in 1966, legislation was passed to provide resources to support people with ID. This legislation stipulates that disabled people must receive basic care and services from the health system. In addition, the judicial system has supported this legislation by guaranteeing funding when services have not been provided. The obligatory services are broad and include early interventions, support for school integration in regular or special schools, therapeutic centers, centers for promoting preparation for employment as well as day and residential centers. Nevertheless, as in other countries in the region, service providers are insufficient and not always adequately trained, since training opportunities are often limited and very expensive [16].

In Chile, there is a national policy for inclusion of people with ID, although the education sector is the only area which has achieved high levels of integration. A systematic program for integration of children with any type of disability, including ID, was implemented. Also, more recently, financial resources for the purchase of educational materials and for teacher training have increased. However, as in other countries, once children with ID or ASD complete secondary education, there are limited opportunities for obtaining paid employment and they are exposed to discrimination and social exclusion since, as with the rest of Latin America, services focusing on rehabilitation, training, or social inclusion for adolescents or adults with ID or ASD are extremely limited and are provided exclusively by nongovernmental organizations, many of which lack appropriate funding [16].

- In Latin American countries, causal factors of social exclusion of persons with ID or ASD can be classified into the following three groups: economic deprivation, sociocultural aspects, and political issues.

- In Mexico, public policies are lacking as well as an institutional model focusing on independent living, social inclusion, and employment. In Argentina, most health service providers are still insufficient and not adequately trained, although the right of persons with ID to specific health care has been adopted into domestic law for over 50 years. In Chile, there is a national policy for inclusion of people with ID, but the education sector is the only area which has achieved high levels of integration.

In Asia, the prevalence of ID and ASD appears to be similar to that in other countries. As in other cultural, social, and economic contexts, discrimination and stigma can cause social exclusion of people with ID or ASD as well their families and especially their caregivers. In Asian countries where research has been done, such as Vietnam, cultural issues which lead to stigma and discrimination have been confirmed. One such issue is the perception that community harmony is

upset when members with ID or ASD are unemployed and unmarried (and with no evident prospects or opportunities for becoming employed or having social relationships). This is seen as a threat to the person's and the family's status. These cultural perceptions lead to lived stigma and discrimination that in turn can determine social exclusion of both caregivers and their family members with ID or ASD [17].

Many Asian countries have not had a history of institutionalization of people with ID and ASD; instead, they have resided with their families, although this has often not implied social inclusion. As in other regions of the world, in Asia, access to and use of the limited services that do exist for people with ID and ASD can be impeded by stigmatization. The limited access to such services can be caused by, as well as lead to, social exclusion. In Taiwan, a greater amount of research and publications about people with ID and ASD exists as compared to many Asian countries. Disability-rights advocacy has also existed in Taiwan, which has not been the case for some other Asian countries. Public policies contribute to reduced costs for some healthcare services for people with ID and ASD in Taiwan, although inequalities in health care are still present (for people in rural areas, for example) and the quality of care needs to be improved. Most caregiving is informal and is provided by female family members. In Taiwan, people with ID and ASD may live longer than in other contexts, which leads to needs for greater care and their caregivers tend to be elderly as well; however, there are limited or no public services to help families resolve this difficult situation [18].

Pakistan is the fifth most populous country in the world with a population of about 220 million. Half of this population is below age 18. There are very few child and adolescent psychiatrists and limited services for children and adolescents including a complete lack of structure and services for individuals with ID and ASD and their families [19]. High rates of depression and anxiety have been reported among caregivers who take care of these individuals in Pakistan [20]. In addition, there is lack of knowledge among professionals regarding ASD and ID leading to delay in diagnosis and timely interventions [21].

Self-determination within a context of living and working in the community is the primary indicator of quality of life. Community centers can be an effective alternative in achieving this goal, as has been widely documented for people with ID or ASD living independently in residential placement, where they can acquire much needed skills and adaptive behaviors. However, such centers must implement programs based on scientific evidence and undergo external evaluations to guarantee high-quality care and achieve the proposed outcomes for people with ID or ASD. This type of program is generally absent in lower- and even middle-income countries.

In order to achieve successful community inclusion (successful in that it improves their quality of life) of people with ID or ASD, there is a need for broad coverage of programs that incorporate certain primary components. These include the following:

- Self-determination developed through previous training.
- Providing sufficient skills for community and home living.
- Guaranteeing good access to services for physical and mental health and related behavioral problems.
- Periodic planning for recreational activities.
- Good distribution of free time.
- Ongoing support for maintaining contact with families as well as personal relationships.

The State must provide an organized social response to the needs of people with ID or ASD and their families, not only with respect to health needs but also in terms of social and labor inclusion. More than a medical issue, this situation reflects the enormous social and economic disadvantages faced by people with ID or ASD and their families in many parts of the world.

Future research, activism, and policy-making as well as service provision in general should focus on the following:

- Defining the magnitude of the problem of ASD and ID and needs assessment for individuals with disabilities in lower- and middle-income countries through well-implemented epidemiological studies and the use of other methodologies (qualitative research especially) when useful.
- Genetic and environmental studies as related to ASD and ID in lower- and middle-income countries, which are of importance in order to continue furthering our understanding of risk factors.
- Capacity building in terms of training for healthcare personnel and promoting human resource development in the area of interventions for ASD and ID in lower- and middle-income countries.
- Full implementation of existing legislation when useful and passage of pending or new legislation when needed that promote education (including independent living skills specifically) and opportunities for employment and community inclusion for people with ID and ASD in lower- and middle-income countries.
- Social policy formulation that addresses the financial or economic aspects of healthcare and special education provision for individuals with ASD and ID since this is not presently covered by any social policy in the majority of lower- and middle-income countries.
- Lastly, research and social policy development should focus on massive public health education to positively influence help-seeking behavior for children, adolescents, and adults with ASD or ID, in order to avoid late diagnosis and provide interventions and services aligned with the UN (United Nation) convention on the rights of persons with disabilities (CRPD) [22].

In Asia, discrimination and stigma can cause social exclusion of people with ID or ASD as well their families and caregivers [14]. Although in general Asian countries do not have policies that specifically seek to segregate and institutionalize people with ID or ASD, the lack of a culture (and accompanying policies) of respect for human rights, inclusion, and non-discrimination often leads to social exclusion. In Taiwan, disability-rights advocacy, academic research interest, and public policies are higher than in other Asian countries as well as life expectancy of persons with ID/ASD, although inequalities in health care are still present, especially in rural areas.

Tip

- In lower- and middle-income countries, there is a general lack of a public health approach and population-level screening for developmental delays and other indications of the possibility of an ID or ASD.
- An important pitfall in diagnosis is not using evidence-based tools; lower- and middle-income countries need greater dissemination of current, evidence-based tools, and most importantly training in their application.
- Psychiatrists and other healthcare providers need to take into account the cultural beliefs of parents of children (or adolescents or adults) with ASD or ID when providing diagnosis, treatment, or services; working with traditional or faith healers can also be important in some cultural contexts.
- Governmental and nongovernmental organizations must provide an organized social response to the needs of people with ID or ASD and their families, not only with respect to health needs but also in terms of social and labor inclusion. The services provided should receive input from both healthcare professionals and family members of people with ID or ASD and where possible, people with ID or ASD themselves.

39.3 Informal Caregiving

The type and level of informal caregiving provided to people with ID and/or ASD, especially adults, varies from culture to culture, from context to context. The burden of informal caregiving is also highly dependent on sociocultural issues as well as on existing policies and services. In many low- and middle-income countries, most adults with ID and/or ASD are cared for at home by family members. In higher-income countries, there is a greater variety of services that provide support both to people with disabilities and their families and caregivers [24]. In England, for example, about 35% of adults with ID live in an informal setting (with family or friends). The economic value of caregivers' work is enormous but infrequently estimated or considered; in the United Kingdom, it is reported to exceed the annual budget of the whole health system [23].

In low- and middle-income countries, the proportion of adults with ID and/or ASD seems to be relatively high, although data on prevalence are lacking. In fact, prevention of developmental disabilities is generally much lower (sometimes nonexistent) than in high-income countries, and survival into adulthood is increasing worldwide in this disabled population. Thus, the need for services and support both for the persons with disabilities themselves (such as educational, employment, and community participation, as well as care when informal caregivers are no longer present) and the caregivers is likely to be high as well, but the services that exist are extremely limited or may be absolutely nonexistent. Also, disadvantages are greater in families with members with a disability (especially multiple disabilities or adjunctive health conditions), including poverty, health problems, and social isolation [30].

Internationally, independent of culture, mothers tend to be the caregivers of family members with disabilities, including children and adults with ID or ASD. In many lower- and middle-income countries, various members of the family, including the extended family, are often involved. Nevertheless, as for higher-income countries, when younger members of the family leave the home, the parents are almost invariably those who continue in the informal caregiver role, even when they get older. Some research indicates that in the absence of programs and services to support adults with ID and/or ASD, which also end up supporting the caregiver and the rest of the family, personal resources become increasingly important, including religion or spirituality and a positive outlook. These personal resources can be even more important in those cultural contexts where discrimination and stigma are greater [30].

In all cultures and contexts, informal caregivers report that their role involves burdens and stress but also satisfaction and gratification. Nevertheless, cultural differences do exist in terms of levels of depression and positive attitudes among caregivers, as research shows. One example is a study on Latina and Anglo mothers of children with ID, cerebral palsy, Down syndrome, or ASD, with pervasive support needs. In this study, the Latinas reported high levels of depressive symptoms (a good population-level indicator of depression), independently of the type of syndrome their child had (i.e., it would appear that the cultural difference was more important than the type of syndrome). Also, while negative impact of caregiving for children with ID did not differ by culture, Latina mothers reported more positive feelings and positive implications for their own life than Anglo mothers [31].

Although caregivers in lower-income contexts have low material and cultural support, research results indicate that mothers and others who provide care are still able to carry out their role for many hours a day and across years. These findings have prompted specific research interest on what psychosocial, spiritual, and other personal factors can make caregiving a positive experience and increase caregivers' resilience. Studies in this area have not yet produced significant results but may be available in the near future [21]. Particular attention must be paid

to ensuring that the identification of personal protective factors in caregivers does not unduly induce socio-health policy-makers to ignore the strong need for public services and support of these caregivers and persons with ID/ASD.

Research has tended to focus on the negative impacts of caregiving or portrayed it through a negative lens. While measuring the negative impact is important, for example, to estimate need for services, an exclusive focus on negative issues does not allow the development of services aimed at enhancing caregiver strengths [25]. In addition, there are methodological limitations of studies with an exclusively negative focus on caregiving that prevent establishing if those negative outcomes that are measured actually stem from caregiving [25].

Informal caregivers of persons with ID do not seem to have lower quality of life than other caregivers (i.e., people with mental health issues or dementia) but show a considerable risk increase of reporting poorer health status, even though poorer health is not necessarily to be attributed to caregiving [25, 26]. Actually, poor health status may have existed before caregiving began; for example, a person with poor health status may choose caregiving because (previous to beginning caregiving) they can not be employed outside the home.

On the other hand, caregivers of persons with ID, mental health problems, or dementia seem to experience more negative impact on their lives as compared to caregivers of persons with other conditions and carers of people with ID are more likely to have financial difficulties and a high caring load [20, 25].

Certain aspects of caregiving may vary by type of disability, while others may vary by culture, although main caregivers' needs seem to be rather stable across different cultures and contexts. For example, caregivers and families in both higher- and lower-income countries (with very different levels of services) seem to agree that family-centered approaches or home-based community support are of value both to themselves and to their family members with disabilities [21]. Also, the lack of employment opportunities or residential care later in life for people with ID or ASD adds greatly to the burdens of caregivers in lower-income countries, while those in higher-income countries also value these opportunities and may see the need for even greater options.

Policy specifically designed for informal carers is lacking in many cultural contexts, especially in low- and middle-income countries. At the policy (macro) level and at the provision (meso) level, caregivers' needs are often conflated with those of the person they are supporting and policy-makers often neglect the complex and sometimes conflicting dynamics of actual (micro-level) caregiving relationships [27–29]. For example, leisure or occupational opportunities provided to persons with ID/ASD may not always be in line with caregivers' needs and wishes. Furthermore, informal caregivers often face health and social problems themselves and therefore become providers and recipients of care at the same time, leading to adjunctive difficulties in constructing policies for their needs [29].

- In most low- and middle-income countries, most adults with ID and/or ASD are cared for by family members. Poverty and long caregiving hours seem to have the highest impact on caregivers' susceptibility to negative well-being. In the absence of programs and services to support caregivers and persons with disabilities they care for, personal resources become increasingly important, including religion or spirituality and a positive outlook.

- Some caregivers support strategies seem to be valued across different cultures and contexts such as family-centered approaches or home-based community support.

Tip

- When treating people with ID/ASD, it is important to take into account their informal caregivers (appreciation/recognition of the caregiving burden is essential);
- Caregiver or family supports/services are important for the quality of care/life of the person with ID/ASD, even though they are not provided directly to them (a family-centered approach);
- Needs of informal caregivers do not always coincide with those of persons with ID/ASD they care for; thus policies for the support of informal carers have to be assessed for such potential conflicts.

39.4 Organizational Culture and Context in Programs and Services

Both organizational culture and context affect the quality of care and outcomes achieved by services and programs for people with ID and/or ASD. For example, in order to develop an analysis focusing on culture in group homes for people with severe ID, Bigby and collaborators [1] used literature about staff performance that puts forward a differentiation between the formal culture of the overarching organization and the informal culture of the staff. The former refers to the culture on which different models of care are based and articulated in terms of processes, strategies, and goals; the latter describes what staff members think about these strategies and goals and how they, therefore, actually implement them in practice. In relation to evaluating and improving services or programs, the authors of these definitions [1, 25, 30, 32] concluded that when the informal culture of the staff is aligned with the formal culture expressed in the model of care, outcomes will be better. When the staff recognizes and takes on the utility of strategies and goals included in the model of care (understood to be part of the formal culture of the overarching organization), and, therefore, actually try to implement them, assuming that the model of care is evidence-based and effective, this alignment of the two cultures will improve outcomes. These two cultures have been termed formal and informal, the latter of which refers to organizational or staff culture as performed at the level of direct interaction with people [1]. Research has shown that it is more common for these two cultures to lack alignment than to achieve it, within support programs for people with ID [1, 31, 32].

Using culture as a construct to analyze the quality of care in services for people with ID and ASD (residential facilities, day centers, or other services, at staff level and/or general organization level) makes it possible to identify problems that do not derive from the model of care espoused by the organization providing the services. That is, when culture is examined and alignment of culture in different parts of an organization is studied (by applying this theoretical construct), issues in implementation of the model of care can be visualized, described, and analyzed in order to propose solutions. The model of care itself may be completely adequate, but obviously if it is not implemented as designed, it will not have the desired positive outcomes for services users (in this case, people with ID and ASD). Thus, analyzing the situation through the lens of culture can offer clarity about whether the problem is in the model of care itself, or (more likely) in how it is being implemented (with fidelity to the original model) [1].

Considering cultural aspects in evaluating the performance of services and programs for persons with ID/ASD can also favor the identification of a series of relevant issues, such as the following:

- How power and leadership are exercised at different levels within services and programs.
- Whether leadership or direction as well as practices in general are aligned with the values that underlie the model of care.
- Whether there is resistance to change, new models of care, new practices, or new ideas

among providers/staff or openness and receptivity instead.
- How service users (people with ID and/or ASD) are regarded; for example, as individuals or only as a group; as fundamentally different/"other" or as fellow humans.
- How the goals of care or services are perceived by providers or staff; for example, whether staff practices are oriented toward "doing things for" people with severe disabilities or "doing things with them;" whether staff practices lead to community presence or actual community participation; whether everyday practices address staff priorities or actual service users' needs [1].

Context is also an important construct with respect to services and supports for people with ID and/or ASD. Although there is no consensus on its definition, context is generally described as the set of personal and environmental factors that interact with health conditions such as ethnicity, socio-demographic characteristics (e.g., gender, age, place of residence), economic factors, cultural factors, education, family wealth, and nutrition [33]. Within the International Classification of Functioning, Disability, and Health [34], environmental or contextual factors represent major determinants for all life activities and participation (involvement in life situations).

When applied to the analysis of support needs for a person with ID and/or ASD, context can be considered as both an independent and intervening variable. As an independent variable it comprises personal (values, goals, and desires) and environmental characteristics, while as an intervening variable, it refers to the organizations, systems, and policies as well as sociocultural practices that take place in the person's environment. Thus, context can be used at three different levels: (1) to describe personal and environmental factors that need to be taken into account when planning for or evaluating supports; (2) to plan the actual support to be provided; and (3) more broadly, to develop policies related to support systems [2].

Contextual factors with the highest impact on the quality of support systems or other services provided to people with disabilities are represented by the level of consideration of person-centered planning and care in general, and the breadth of options provided by the caregiving organization as well as associated managerial strategies and practices.

In terms of policy development, contextual factors need to be taken into account at all levels that influence human functioning outcomes, that is, at the individual (microsystem), family (mesosystem), and societal (macrosystem) levels [2, 35]. A framework to guide policy development that focuses on an operational definition of context has been proposed by Shogren et al. [2]. It includes administrative principles such as collaboration, accountability, individualization, cultural responsiveness, and prevention or amelioration. This framework also proposes outcomes such as personal independence, productivity, social inclusion, and self-determination, as well as positive outcomes at the family and social levels. All these outcomes are also permeated by culture.

- Organizational culture can affect implementation of the model of care and quality of services.
- When the staff recognizes and takes on the utility of strategies and goals of a model of care (understood to be part of the formal culture of the overarching organization), and try to implement them, assuming that the model of care is evidence-based and effective, quality of care and outcomes achieved by services and programs improve.
- Analyzing the situation through the lens of culture can permit evaluation of the efficacy of the model of care itself and how it is being implemented within the support systems for people with ID and ASD.
- In terms of policy development, contextual factors need to be taken into account at all levels that influence human functioning outcomes, that is, at the individual (microsystem), family (mesosystem), and societal (macrosystem) levels.

Tip

- A common pitfall is assuming organizational culture has no effect on care or that it will be the same at any institution (big or small, e.g., a large institutionalized residential setting or a small group home);
- The concept of organizational culture can be used as an heuristic tool for improving quality of care or services; for example, it can be incorporated into evaluations of programs and services in such a way as to produce not just a score on how good or bad the services are but ways problems could be dealt with or in which quality could be improved.

Key Points

- Good data about the prevalence of ID and ASD can allow stakeholders to advocate for better coverage and better quality of services, including expansion of existing programs and establishment of new programs.
- There is a lack of a public health approach and population-level screening for developmental delays and other indications of the possibility of an ID and/or ASD.
- In many contexts, there are few psychiatrists per inhabitant and diagnosis may be carried out using tools that are not current or that are not evidence-based.
- Type and level of informal caregiving vary from culture to culture; the burden it implies also varies depending on sociocultural issues and on existing policies and services.
- Research on caregiving through a positive lens (focusing on positive effects instead of exclusively on negative impacts) is needed to provide inputs for services and programs that can build on caregiver strengths.
- The context of and organizational culture in support systems, programs, and services for people with ID and ASD can impact how well the model of care is implemented and quality of care.

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Spiritual Issues

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Contents

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Learning Objectives

- To become aware of the connection between spirituality and quality of life.
- To become aware of the importance of spirituality and religion for individuals with intellectual disabilities.
- To learn about spiritual concepts and expression of people with intellectual disabilities.
- To learn about models of spiritual care.
- To learn about characteristics of inclusive faith communities.
- To learn about ways to support people with intellectual disabilities and professionals in addressing spiritual needs.

40.1 Introduction and Overview

Spirituality as a fundamental human need is getting increasing attention in society and health care. Pierre Teilhard de Chardin addresses the indefinable complexity of the whole human nature: 'We are not human beings having a spiritual experience, we are spiritual beings having a human experience' [1].

The title of this chapter 'spiritual issues' indicates that the vast dimension of different facets of the human condition, which reaches beyond the here and now can be addressed only fragmentarily, not only due to the scarcity of empirical studies.

After a brief discussion of definitions of the terms 'spirituality' and 'religion', the relationship between spirituality and health with a special focus on mental health is addressed.

A brief introduction of cognitive and emotional aspects of spirituality leads into a section in which research findings of spiritual lives of people with intellectual disability (ID) are presented. This is followed by a part focusing on the relationship between quality of life and spiritual well-being.

The section on spiritual care for people with ID and/or autism spectrum disorder with high support need (ASD-HSN) also encompasses attitudes of different religions towards disability in general, aspects of inclusion in faith communities and policy statements, which call upon the 'spiritual rights' of people with ID and ASD-HSN.

40.2 Definition

The place of spirituality and religiosity in society is under constant change. The complexities and controversies in defining the concepts are shown in a perennial discourse. Although the terms spirituality and religion are frequently used interchangeably, in recent social science, the concepts are differentiated [2].

The broad concept of spirituality is understood for instance by Swinton as that aspect of life that gives it its 'humanness'. Generally, the term is linked to the personal quest for understanding answers to the ultimate questions about life, meaning and purpose. Further, it includes dimensions of the quest about relationship with the sacred or transcendent, as well as the 'holy amongst us'. Religion is seen as the more specific version of spiritual expression finding place in an organized belief system, practices, shared rituals as well as symbols which facilitate experiencing closeness to the sacred or transcendent [3–5].

Spirituality is often seen to be an individual quality whilst religion is linked to entities, with religious rites at the centre, but also usually characterized by membership and containing related social, cultural, service and economic activities and aims [6].

However, it is argued that the distinction between religion and spirituality should only be taken cautiously. It could implicitly denote the wrong assumption of spirituality being good and religion being bad. Further, the polarization tends to neglect that many spiritual experiences occur in, and are shaped by, organized religious contexts, where the difference between religion and spirituality is not perceived as such. Such a distinction additionally can lead to unnecessary duplications in concepts and measures [7].

In research, spirituality is often assessed either in terms of religion or by positive psychological, social or character states like questions asked about meaning and purpose in life, connections with others, peacefulness, existential well-being and comfort and joy. This approach can lead to a bias that spirituality – defined as good state of mind and positive psychological or social traits – is found to correlate with good mental health [8].

40.3 Spirituality and Health/Mental Health

Research findings display a considerable positive impact of spirituality and religiousness on physical and mental health, in general and in terms of mortality, morbidity, disability and better recovery from illness [9, 10].

As found by a systematic review examining the literature from the years 1990 to 2010, two-thirds of the studies showed a relationship between the level of spiritual/religious involvement and a lower prevalence of mental disorders, whilst only 5% have identified a negative relation. The positive effect was found in all studies on dementia, suicide and stress-related disorders and about two-thirds of the studies focusing on depression and substance abuse. Mixed results were found on the relationship between religion/spirituality and schizophrenia and no or negative association in studies on bipolar disorder [11]. A review on religion, spirituality and mental health in the West and Middle East shows the positive relationship with well-being, hope and optimism as well as meaning and purpose. Further, more internal personal control and a higher self-esteem were reported amongst the more religious/spiritual individuals [12].

40.4 Spirituality and Quality of Life

In various studies on healthcare practices for people with chronic illnesses, a fundamental and strong correlation between the concepts of quality of life (QoL) and spirituality could be shown [13].

Spirituality was detected as a predictor of high level of QoL and subjective well-being in the medium and long term, together with optimism, good mood, collaboration in care and social support even in studies on complex psychosocial factors.

Further, there seems to be a link between the importance given to spiritual life, the level of satisfaction and positive health and well-being, both physical and mental [14].

Considerable evidence exists that supports the association of spirituality with the improvement of cognitive performance, affectivity, impulse control, coping mechanism and functioning of the immune and endocrine systems, increase of the life expectancy and reduction of psychopathological vulnerability, suicide risk and cardiovascular diseases, which could also be relevant for people with ID and ASD-HSN [15, 16].

Spirituality is also presumed to manifest in various psychological domains, such as self-esteem, motivation and relatedness [14, 16].

The concept of spiritual well-being (SWB) is a recent attempt for an operationalization of the strong connection between spirituality and QoL. The focus lays on the way and the amount by which spirituality affects individual well-being and QoL [17]. Due to being a well-defined and objective concept, spiritual well-being is used in a number of studies in which a strong association with mental and physical health could be shown [18].

The spiritual well-being scale (SWBS) is the most popular tool used to measure spiritual well-being. Good psychometric characteristics have been proven for the instrument [19]. Furthermore, the validation in numerous countries makes it usable in many different cultural and religious backgrounds, both in healthcare and in general research [17, 18, 20, 21].

Through studies of the general population, which also included people with borderline intellectual functioning and milder forms of ID, a link between the level of SWB and personality traits could be seen, such as extroversion, easy adaptability and willingness to understand [22]. Furthermore, SWB seems to correlate positively with the ability to set goals, with motivation and with self-confidence as well as with participation in religious activities. SWB correlates negatively with individualism, loneliness and manifestations of unconditional personal freedom [23].

Further studies on the relationship between spirituality and QoL have been conducted by the World Health Organisation (WHO). This also led to the development of another widespread tool – the WHOQOL Spirituality, Religiousness, and Personal Beliefs (WHOQOL-SRPB [24]). Through the appli-

cation in 18 different countries (with a sample of more than 5000 people), it was found that the concepts of spirituality, religion and personal faith represented relevant areas of QoL in all of them. Spirituality correlates especially with the general QoL, psychological being and socialization. In a hierarchical and stepwise regression analysis of the data from a sample of people with health problems (N = 588), including a small percentage of individuals with ID and ASD-HSN, spirituality was found to be the third amongst four domains explaining most of the variance (52%), after the level of independence and the environment but before physical being [24].

To date, both SWBS and WHOQOL-SRPB have not been used very much with people with ID even though the dimension of spirituality and QoL could be interesting dimensions in the planning of interventions [25].

Spirituality may have a high impact on the QoL of PwID/ASD. It represents a core element in the system of values of many persons with ID, across the various cultures and the different geographical and ethnic contexts. Thus, spirituality should represent a main reference area in intervention planning and outcome assessment.

40.5 Cognitive and Spiritual Development

Theories on the connection between cognitive and spiritual development draw different parallels focusing, for example, on the ego development, autonomy and self-awareness as they affect and are affected by relationship to others and the divine. Other theories aim to illustrate the development of moral understanding and consequent behaviour towards others or suggest that individual faith development reflects a 'meaning-making' process in which individuals seek the understanding of their own lives in relation to possible guiding values and commitments [26, 27].

Contrary to approaches which link cognitive and spiritual development, recent neuroscientific findings suggest a potential sense and development of an interior transcendent dimension even amongst people with the most severe neurodevelopmental disabilities [28]. The emotional functions which might be considered as the precondition for spirituality can be distinguished from the current conceptions of intelligence, logical and deductive skills upon which the diagnosis of ID is based primarily. These emotional functions are mainly located in subcortical ancient brain areas whereas neocortical areas are responsible for logical deductive thinking [29]. The role of music as universal language with a direct pathway to the human emotional and spiritual dimension deserves further investigation especially in people with ID [30].

In fact, people with disabilities are sometimes viewed as experiencing a greater intensity of spirituality or even as being 'closer to God', whilst rationality is considered an obstacle [31]. One needs to be cautious about equating spirituality with the capacity to reason [32, 33].

Recent neuroscientific findings suggest a potential sense and development of an interior transcendent dimension even amongst persons with most severe neurodevelopmental disabilities.

40.6 Spiritual Life in People with ID

In 2017 a systematic review article was published by Sango and Forrester-Jones on intellectual and developmental disabilities, spirituality and religion. In their extensive literature search of studies conducted between 1990 and 2015, the authors identified 57 publications. Only five articles met the inclusion criteria of being empirical and focusing on the adult population with ID directly. One of those had some quantitative measures. Additionally, to the studies, which were part of the review, the results of a qualitative research by Liu and colleagues interviewing 20 young people with autism and ID are reflected.

In the following, we address main findings regarding the importance of spirituality and

religion, religious concepts, expressions as well as the impact of religion and spirituality reported by the individuals with ID themselves [4].

40.6.1 Importance of Spirituality/Religion

A stimulating qualitative research on religious interest or background has been conducted in northwest England at the beginning of the new millennium, as part of a larger research project on users of adult services for people with ID [6]. Authors report the ability of people with ID of diverse faiths to express positive and strong religious identities. Further distinctions of one's own religion and its connected practices from other religions were made. Some of the participants even made clear that their faith did not root in childhood or family life, but was their own choice. Even against indifference or hostility from support providers, participants showed persistence in religious expression.

Shogren and Rye report 83% of participants with intellectual disabilities indicating a self-view of being religious. A belief in God was stated by 95% of participants. An even higher percentage of 95 stated that they thought about God quite frequently. Religion was described as somewhat (34%) or very important (56%) to them. A significant correlation between the ratings of importance and self-reported ratings of frequency of religious participation could be found [34].

Swinton reports about results from individual and focus group interviews with people with learning disabilities, support workers and service providers in Scotland and England without giving methodical details. He found that in contrast with minor indication of importance of spirituality by carers and support workers, the majority of people with disabilities expressed a deep spirituality [33].

Understanding and defining individual conditions of disability from a spiritual perspective is paramount when considering the impact of cultural components on mental health needs and care of persons with neurodevelopmental disorders [35, 36].

> Understanding and defining individual conditions of neurodevelopmental disability from a spiritual perspective is paramount when considering the impact of cultural components on mental health needs and care.

40.6.2 Understanding of Spiritual or Religious Concepts

The importance of friendship as a significant spiritual relationship became clear during many of the interviews in Swinton's survey. He reported that people's understanding of God was most often expressed in the language of friendship and tended to refer to God in human with terms as 'a friend', 'me mate', 'a good man', 'the gaffer'. God was seen as a natural extension of everyday social relationships and a vital form of social support. It has to be mentioned though, that these perceptions were evolving from a Christian context and that there is a need to explore the ideas of people with intellectual disabilities from other religious backgrounds [33].

From Shogren's study, 29 participants (71%) drew an anthropomorphic picture on the Draw God task. Abstract drawings in 17% showed a heart with written 'God loves me' or a church with the letters ILG which meant 'I love God'. There was no difference between participants with mild and moderate ID with respect to the degree of an anthropomorphic image of God [34].

In the same study, a religious picture identifying task was performed with 24 pictures from the three categories 'concrete', 'abstract' and 'non-religious'. These pictures were presented to the participants one at a time in random order with the question: Does this make you think about God or religion? A Bible (98%) and a church (98%) were chosen with the greatest frequency from the concrete scale. From the abstract religious scale, 78% chose

a heart with LOVE underneath. Interestingly, also money (27%) and muffins (24%) were associated with God and religion as they reminded them of occasions when they gave money or ate cake at church [34]. In Turner's 29 semi-structured interviews, many participants used religious language and expressions and were aware of their own religious affiliation as Catholic, Protestant or Muslim [6].

40.6.3 Expression of Spiritual Life in People with ID

Prayer proved to be a primary avenue of spiritual expression. This became evident in Turner's, Liu's and Shogren's interviews with individuals with ID. About 90% reported to pray regularly [6, 34, 37].

In the study by Shogren and Rye, 76% of participants reported that they attended worship services, with 49% of these individuals quoting at least weekly attendance. There were only few other activities participants engaged in more frequently on a weekly basis.

Other less prominent ways in which the young peoples' spirituality found expression was through rites of passage, through social connections or through acts of service or ministry to others [37].

40.6.4 Impact of Spirituality or Religion in People with ID

Spirituality seems to have a strong beneficial impact on the life of persons with ID/ASD, in terms of aid, friendship, love, warmth and protection. It also helps in providing meaning for life and coping with negative life events [6, 37, 38]. Spirituality and religiosity may also represent a unique resource in the understanding of the life and death cycle, as well as in bereavement [39].

Having a religious faith has been found to support personal growth and to increase the perception of life as active and satisfying. Catholic persons with ID defined christening, communion and confirmation as some of the significant experiences of their life and as extraordinary facilitators for the acceptance of their disability, since they made them feel loved, valued and understood by God [37]. Additionally, the benefit related to a sense of religious connectedness and belonging could be seen [6].

In people with Down syndrome, spiritual well-being has been associated with a general satisfaction towards life and with many other aspects of existence that are linked to a good QoL, such as psychological balance and personal control, building positive relationships, acceptance of life's difficulties and changes, internalization of purposes and dimensions transcending the individuality [40, 41].

The positive role of beliefs in the lives of people with ID could further be seen in the interviews of support providers in Shogren and Rye's study [34]. The proxy's descriptions indicated that religious participation supported the development and enhancement of positive social and personal values.

The impact of spirituality on mental health has been studied more than that on physical health, with around three quarters of research findings reporting a strong relationship between the level of involvement in spiritual activities and the lower prevalence of mental disorders, especially for pathological anxiety, depression, substance abuse and suicide. This has been ascertained for different religions, specifically Catholic, Protestant, Jew, Muslim and Buddhist [11, 14]. Carrying out spiritual activities in daily life, such as doing things that allow to feel at peace with themselves, helping other persons, celebrating anniversaries and special occasions or being an active member of a religious organization, may favour feelings of trust, safety and security as well as positively influence life expectancy and perceptions of themselves and of others [42].

Interviews with adolescents and adults with ID, integrated with information from their caregivers, have confirmed that being satisfied with prayer, inner faith and participation in religious group activities have positive effects on mood [37, 43].

Interestingly, research on the potential negative impact of religion/spirituality on people with ID is lacking. The limited literature available indicates that sometimes

religious dogmas, precepts and icons can represent a liability to mental ill-health more than a protective factor. Hallucinations, obsessive thoughts and delusions with religion contents are frequent in clinical practice and can give rise to problem behaviours. In more vulnerable individuals, warped interpretations of religion and practices may strengthen neurotic tendencies, enhance feelings of fear or guilt, limit socialization and being used in primitive and defensive ways to avoid making necessary life changes [8, 13].

Spirituality seems to have a strong beneficial impact on many aspects of many PwID/ASD's lives, such as aid, friendship, love, warmth, protection, meaning for life, coping with negative life events, understanding of the life and death cycle and bereavement.

40.7 Family Spirituality

The experience of spirituality of family members in the particularities of marriage, sexual intimacy, procreation, parenting, building and sustaining relationships, providing, sheltering and feeding transcendental experiences represents a fundamental reference for the person with ID/ASD in the development and consolidation of his or her own spirituality, especially whilst living in the family [13]. Spirituality in the family is also relevant for the identification of spiritual needs of the person with ID/ASD and the provision of satisfactory support in their living environments [32, 44]. Persons with ID/ASD living outside their families seem to need more concrete personal relationships and to rely more on transcendent sources of help than those who live with their family. Nevertheless, the extent of the impact of family spirituality on spirituality of the person with ID/ASD and associated aspects of QoL is yet to be determined [13, 45]. To date, the few studies addressing this issue have focused primarily on the extent to which parents find spirituality and/or religion to be an important resource for their life [46–49]. Religious faith was described as important or very important by the vast majority of parents of children with ID, ASD and other developmental disabilities in different cultural contexts [50–52]. However, the level of actual involvement in the array of activities offered in or through a local congregation has not been examined in depth. Available literature shows that only six of ten religious parents attend religious services at least weekly, and three of ten attend yearly, or even less frequently or never [45, 53].

Persons with ID/ASD living outside their family seem to need more concrete personal relationships and to rely more on transcendent sources of help than those who live with family [40].

High levels of spirituality or religion in parents were found to have a positive impact on the QoL of the whole family as well as on problem behaviours and support needs of the person with ID/ASD-HSN, especially during transition to adulthood [54]. For mothers of children with ID/ASD, the maintenance of a spiritual or religious dimension has been associated with stress reduction, containment of mood drops, increase in self-esteem, general life satisfaction, optimism, and self-control [55–57]. Spiritual and religious beliefs have been found to also improve parents' response to disability, understanding of disability, coping and family adaptation and to provide a source of meaning, hope, and strength [48, 58].

However, attaching great value to spirituality across their life does not exempt parents and other family members from the risk of not providing their child or relative with ID/ASD with all the opportunities they need to appropriately develop their own sense of spirituality. In fact, the birth of a child with ID/ASD-HSN and/or the persistence of adaptive or behavioural difficulties across years can lead parents and other family members to a spiritual crisis as well as the great burden of daily practical needs and future planning can determine a neglect of the spiritual dimension [41].

Anecdotally, many families will share stories of experiences of rejection or neglect by faith communities, but there has been little research on the extent of those experiences. Hoping and trusting a religious community to

be welcoming is indeed a risk, with the potential for great benefits but also disappoint [59].

As individuals, families of PwID/ASD may experience different impacts of spirituality and religiousness on their QoL on the basis of the sociocultural context, which can also be determinant for acceptance of disability, quality of diagnosis and care and community inclusion [13]. For example, in sociocultural contexts where prevalent religions interpret children with disability as a divine gift, mothers seem to maintain a higher QoL than in those contexts whose religions consider disability as a divine punishment [60, 61].

The support that parents and other family members of PwID/ASD draw from their spirituality, religious involvement and congregational connections is another important point, which has been scarcely addressed by research [62–64]. The few available findings show social support to be rather easy to achieve and to be associated with family quality of life improvement and highlighted sense of strength [65]. Lower rate of religious/spiritual social support has been reported in case of urgent need of help, greater disability severity and higher frequency and intensity of challenging behaviours [45].

Spirituality/religion is a relevant aspect of the lives of parents and caregivers whose family members have ID. It is also very important for the identification of spiritual needs of the person with ID/ASD and support in satisfaction. Congregational involvement, spiritual beliefs and practices and religious/spiritual social support comprise most important areas of inquiry. Services and supports can be improved by better understanding individual needs and preferences.

40.8 Spiritual Care for People with ID

Understanding the importance and the place of spirituality in people with ID inevitably leads to implications for service providers. In fact, it should become an urgent service priority. This includes that religious interest is identified proactively. Staff training to ensure that religious issues become routinely addressed within service practices is recommended. Further, the development of individual plans addressing people's religious needs and preferences is suggested [6].

According to Swinton [33], who interviewed support workers and service providers, the importance of spirituality was not always seen and given attention to. Whilst some apparently have dealt with this issue, some stated never really having thought about the relevance of spirituality, or, if they did, that it was not very relevant for their daily practice. As the issue never occurred in any training, amongst many it was only brought up when they were particularly religious or spiritually orientated themselves.

Frequently, wariness is shared about the ability of people with ID to have a spiritual life due to their cognitive limitations and the perception that spirituality and faith in God require a higher level of abstract thinking as well as the presumption of a cognitive threshold for the membership in a faith community. Further, some are sceptical of religion and a possible enforcement of unhelpful values and morals on a group of people, which might be particularly vulnerable to psychological manipulation [33].

Also, Carter mentioned that the intersection of faith and disability is sometimes marked by hesitation and uncertainty for service providers. Concerns about state-supported proselytization can lead some staff to ignore the spiritual needs of those individuals they serve, whilst eagerness to encourage religious connections can lead other staff to trump people's preferences [66].

In a study by Sango and colleagues, the perceived and operationalized spiritual and religious aspects of care for people with intellectual disability in a stated 'faith-based' service (Adam's House) and another service stating 'no-faith' (Greenleaves) were examined [4].

They chose a mixed-methods approach interviewing 42 support workers, as well as gaining information through observations of service participants over a time span of 6 months in each service. Furthermore,

information about the self-assessment of 'spiritual competence' and attitudes towards providing spiritual care to individuals with ID was gathered using the 'spiritual care competence scale' (SCCS) and the 'spiritual care perspectives scale' (SCPS).

Eighty-one per cent of Adam's House staff stated that their belief and spiritual background played a role in their daily lives and their approach to work. Only 62% of staff working in Greenleaves who had a religious background expressed its influence on their lives and jobs, especially during difficult times. The effect of a 'code of practice and conduct at work' was mentioned by the others.

In terms of attitudes towards spiritual care, as for example being positive about the residents' spirituality, no differences between the care providers of the two sites could be found. There was also no difference between the two staff groups regarding the question if enough attention was devoted to residents' spiritual care, or participants' own spiritual care competence and experience.

Regarding training, significant differences became evident, as 62% of Adam's House staff reported of receiving training in spiritual care as part of their formation, whilst 90% of Greenleaves claimed of not having received any training with that perspective.

Though both groups reported support of the residents' attendance of church service, further aspects of spiritual care were approached differently.

The homely functional care approach of Greenleaves focuses on the individuals' physical needs, promotes empowerment and independence as well as it respects and supports personal choices. Even though individuals expressed interest in and requested spiritual and religious activities, activities of spiritual nature were missing in the daily schedules.

Adam's House shows a structure which allows the staff and service users to share their daily life experiences, as for example the tradition of eating together, which is also connected to the Christian value system. This serves as ground for spiritual intimacy between service providers and users, enabling individuals to reveal special spiritual needs as the request for prayers. Further, the less institutional setting seems to lead to apparently more familiar attachments and care relationships with a personified dimension of spiritual community. In Adam's House, alternative ways of communication like religious and spiritual pictorial images were used for simplifying prayer and praise sessions.

Over 50% of staff of both services agreed that barriers included themes and categories such as 'communication issues', 'shortage of staff' and 'staff perception of spiritual understanding'.

Similarly, Swinton [33] indicated that communicational barriers prevent caregivers from exploring the emotional and spiritual aspects of the lives of people with ID.

Assumptions of staff were found to be the main barrier in providing spiritual care. As for instance 50% of Greenleaves staff suggest that residents 'might not have a spiritual need' or did not comprehend spiritual matters due to their 'mental capacity'. A lack of knowledge or belief in 'spiritual things' and absent confidence on how to provide religious or spiritual care were stated as further obstacles. This reinforces Swinton's statements.

Although empirical research of different faith-based and secular care models is not available, the model of L'Arche as a Christian faith-centred shared community living for individuals with and without disabilities attracts worldwide attention [67, 68].

Inspiration of spiritual care provision can be drawn from the example of the therapeutic living communities for deaf people with multiple disabilities set up by the first author of the present chapter [69]. In the regular morning meetings in the three facilities with 16–30 residents each, members with disabilities and professional staff participate voluntarily and actively. During these morning meetings, stories from the Gospel of Jesus are shared through real-time painting, role plays and discussion in sign language. By this therapeutic and adaptive principles like conflict resolution and forgiveness, overcoming hardship and trauma and being an active and positive member of the community are conveyed.

Tip

In the current organization of health and social-educational services, the consideration of the spiritual needs of PwID/ASD is scarce. Specific staff training and individual intervention plans are recommended for future policies. High quality research evidence also has to be promoted.

40.9 Inclusion in Faith Communities

40.9.1 Religious Approaches to Disability in General

Even within different religions, the ways how disabilities are seen and addressed vary widely [70]. Some researchers have noted how 'disabling theology' links disability with impurity and sin and proposes suffering as a way to gain heavenly rewards [71, 72].

Most religions emphasize the responsibility of faith community members to recognize the plight of persons with disability and to improve their condition and state, which, to some extent, nurtures overprotective and paternalistic stances [73].

The wide diversity of perspectives even within a religion is reflected in a qualitative research amongst Roman Catholic leaders. Patka and McDonald identified five religiously anchored narratives of intellectual disabilities ranging from viewing individuals with ID as having a special relationship with God, being eternal children, needing to conform to existing settings, being defective to them, being a natural part of human diversity [31]. Surveys amongst parents indicate similar assignments to their children's disabilities as: gifts from God, abundant blessings, disabled by design, tests of their faith, punishment for sin etc. [74].

Interactions between personal experiences, several cultural practices and religious views may explain the variety of approaches in perceiving disability [75] which mainly influence the inclusion of people with intellectual disabilities in the respective faith communities.

40.9.2 Inclusion and Participation in Faith Communities

Inclusion in community activities has relevant implications for quality of life of most people with ID/ASD. Several position statements have been made by major organizations acknowledging the rights of people with disabilities to access supports, opportunities and relationships in this dimension of their lives [76]. According to TASH (The Association for persons with Severe Handicaps), an international organization advocating with and on behalf of people with significant disabilities:

> 'Supports shall enable children and adults with disabilities to participate in every aspect of socio-cultural life including, but not limited to, home, school, work, cultural and spiritual activities, leisure, travel and political life.' [77].

Participation in faith communities is one community activity that is often difficult for people with disabilities [78]. A survey of the US Kessler Foundation and the National Organization on Disability conducted in 2010 shows that there is a gap of approximately 14% between people without and with severe disability attending faith community services at least once a month ([65, 79, 80]).

A survey by Carter amongst 12,700 adults with ID in the US reported a similar high number of around 50% attending religious services in the past month, with variations related to individual and contextual factors with respect to involvement. The difference in the rate of participation, however, does not necessarily originate in the lower importance of faith in the individuals lives. A survey conducted by the US National Organization on Disability determined that faith was considered important by 84% of people with and 87% of people without disabilities. Consequently, the disparities in attendance cannot be due to the lack of interest [81].

Studies which interviewed individuals with intellectual disabilities themselves clearly indicate the high importance of attendance in faith community activities [34, 37].

The meaning of the term 'inclusion' and the way it should be increased in faith settings

has been and still is debated by many religious leaders and congregants through position statements, expert opinion papers and some studies [59, 65, 82–87], but a consensus definition of an 'inclusive faith community' has not yet been reached.

The topic of accessibility was addressed in a study by Larocque and Eigenbrood, who aimed to examine the current physical and attitudinal accessibility of religious congregations for people with disabilities. The results show that the majority of the 91 participating congregations were in the phase of planning for the inclusion, with only a small number of congregations (4.4%) reporting that they had 'not started'. Most of the congregations also indicated that they are on their way to enhance inclusion through specific actions. Apparently medium (501-1500) and larger (1501 and more) size congregations rated themselves significantly more accessible than smaller congregations [88].

In the United States of America, there has been significant debate about whether a congregation should have a 'special ministry' with people with disabilities versus including them in all aspects of a congregation's life, or a combination of the two [32].

In the last decade, the debate has progressively moved from accessibility to inclusion to belonging [32], with an increasing focus on how to make connections basing on shared identities, regardless of ability [32, 78]. Carter proposed 10 dimensions of belonging, ranging from being invited to being needed and loved and their salience to inclusion [59].

40.9.3 Characteristics of Inclusive Faith Communities

There is only a small number of empirical studies aiming to explore the characteristics and factors related to successful inclusion of people with ID in faith communities.

Griffin and colleagues conducted a survey on characteristics of faith communities related to greater inclusion in the United States. In total, 160 individuals responded to the web-based survey, including people with disabilities, their family members and leaders of faith communities and congregants [79].

The Survey on Inclusion in Faith community comprised 145 items. Besides questions related to the respondents' characteristics as demographics, quantity of attendance in services, demographic characteristics of faith communities as, for example, size, location, distance travelled to attend or number of congregants with disability, disability-related characteristics of faith communities were collected.

The survey questions concerning inclusive outcomes converged on three factors, which accounted for 65.78% of the variance.

Factor I related to how welcoming and inclusive the faith community was to people with disabilities. It accounted for 47% of the variance and included activities like the assistance provided for people with disabilities to become members, the provision of accommodations and adaptations for full inclusion, support for their families and the adoption of a welcoming attitude.

Factor II related to the roles of people with disabilities, which was quite specific and asked if people with ID for instance were included/encouraged to serve on committees, as church officers, greeters/ushers or to participate in outreach programs.

Factor III of the community's physical accessibility subsumed the approachability of for instance the restrooms, classrooms or fellowship areas.

Further, five independent predictors related to the three outcomes were identified: commitment of leadership, educational resources, positive portrayals in religious teachings, closer relations to disability organizations and commitment to social justice. The most significant of the variables was the community leaders` commitment to inclusion. The faith community's demographic characteristics, such as the community's size, location or number of congregants with disabilities did not play a significant role.

Ault and colleagues reported qualitative findings from a mixed-methods study exploring the perspectives of 416 parents on the welcome and support they have experienced within faith communities, both for themselves

and for their sons or daughters with developmental disabilities [50].

Most parents emphasized the important role of accommodations and specific support provided by a faith community to have impact on themselves and their children's participation. Examples given were about the support during religious education classes received by peer tutors, volunteers or hired assistants. It further was presented positively when faith communities gained information about adaptations appropriate for their children, including information about the child's behaviours and needs. However, the majority expressed their dissatisfaction about the lack of support. This includes the non-existence of accommodations for the child as for instance the access to sign language interpreters or available visually accessible materials as well as the lack of understanding of the behavioural supports needed for their children. Some parents also reported either explicit non-acceptance into programmes or inappropriate placements in religious education programmes. Parents and/or siblings often were the ones responsible for providing the support their children needed to participate in programmes.

Another major factor was the children's traits. For example, parents (93.7% of the comments) described their children as having behavioural characteristics such as 'being noisy in church', 'unable to sit still', 'touching others', 'overwhelmed by the noise', 'aggressive with others', 'unable to understand religious education lessons' and 'overstimulated'. These behaviours led to results ranging from embarrassment of the parents, to parents being told to not to bring their child back to services or activities. How important a particular person, as for instance a congregational leader or an individual acting as advocate, can be in including the family in the faith community, was another main finding [50].

In accordance with Griffin's predictors [79], the use of educational resources and links to disability organizations, the need for expertise and knowledge was emphasized by the parents. They described ways in which education and training were needed and how people who were educated about disabilities could be particularly helpful to the family.

Church leaders might be encouraged by a study by McNair and Sanchez who found in a survey for the National Organization on Disability's 'Accessible Congregations Campaign' amongst church leaders that people with disability were not regarded as being expensive to the church, driving potential members away, nor taking excessive time away from other potential areas of service [71].

In a recent systematic review, Miller and Skubik-Peplaski [89] identified the following specific supports to increase participation for individuals with disabilities in faith communities:

- Welcoming/positive attitude towards all members
- Accessible environment
- Education and training to members
- Parental support
- Spiritual counselling
- Special worship services for people with special needs
- Accessible materials
- Support groups
- Advocate support for individuals with special needs
- Peer tutors, volunteers or hire assistants.

Although people with ID want to be part of faith communities, the reality of inclusion is lagging behind. Religious attitudes towards disability influence the extent to which inclusion of people with disabilities is accomplished in faith communities. A welcoming atmosphere, active involvement of persons with ID and physical accessibility are characteristics of inclusive faith communities. Leaders of congregations have a major responsibility for the process of inclusion.

40.10 Conclusion and Policy Statements

Recent policy statements endorse tactful consideration of individuals' spirituality and religious practice.

The World Psychiatric Association (WPA) asks psychiatrists not only to understand spirituality and religion in their relationship

to aetiology and treatment of psychiatric disorders but also to get specific training in order to approach religion and spirituality in a person-centred way and to collaborate in a multidisciplinary way with faith communities [36].

The professional position should in no terms be used to proselytize for spiritual or secular worldviews. But in contrast, awareness, respect and sensitivity to spiritual/religious beliefs and practices are requested and should be demonstrated both in relation to the patients as well as staff.

Both the beneficial and the harmful potential of religious, spiritual and secular worldviews and practices should be addressed and information should be shared with the wider community for supporting the promotion of health and well-being.

In the joint position statement of the American Association on Intellectual and Developmental Disabilities (AAIDD) [76] and the Arc [67], it is emphasized that spirituality, spiritual growth and religious expression as well as the choice not to participate in religious or spiritual activities are rights and should be paid respect to by service systems and faith-based communities.

Key Points

- Spirituality is an important dimension of human beings exceeding cognitive capacities. In individuals with ID, openness to spiritual experiences and interest in religious issues are frequently not attended by professional care and support workers, which may be due to uncertainties and hesitation about ways of adequate provision.
- Different models of spiritual care range from faith-based communities to individual approaches of inclusion in faith communities. Inclusive faith communities are characterized by accessible environment, education and training for all members, parental support as well as special worship services for people with special needs. Spirituality comprises a dimension where professionals can learn from people with ID.

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Further Reading

Resources

http://friendship.org/ (Friendship Ministries, 2017).
https://www.autismspeaks.org/family-services/your-religious-community (Autism Speaks, 2018).
https://opwdd.ny.gov/accessibility (Office for People with Developmental Disabilities. New York State, 2018).
http://www.faithinclusionnetwork.org/ (Faith Inclusion Network, 2012).
https://www.aapd.com/advocacy/interfaith/ (AAPD, 2018).

www.autism-society.org/faith (Autism Society, 2016).
http://www.qualitymall.org/directory/dept1.asp?deptid=19 (Regents of the University of Minnesota, 2008).
http://www.vibi.at/en – visual bible with drawings, international sign videos and easy to understand text (VIBI, The Visual Bible, 2022).
https://s3.amazonaws.com/poacwebsite/wp-content/uploads/2016/06/04184324/DimensionsofFaith2009.pdf (The Boggs EM. Center on Developmental Disabilities, University of Medicine & Dentistry of New Jersey, Wood Johnson R. Medical School, Department of Pediatrics, 2009).
https://aaiddreligion.org/activities (American Association on Intellectual and Developmental Disabilities - Religion and Spirituality Interest Network, 2022)

Forensic Issues in Intellectual Disability and Autism Spectrum Disorder

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Contents

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This chapter provides an overview of offending in adults with ID and/or ASD covering epidemiology including risk factors for offending behaviours, offence type along with clinical presentation, risk assessment, interventions and a brief description of UK-based forensic/secure services as it relates to this population when in contact with the criminal justice system.

Learning Objectives

1. Knowledge of prevalence rates for offenders with ID and/or ASD across the criminal justice system.
2. Understanding of main types of offences committed by adults with ID and/or ASD.
3. Awareness of approaches to risk assessment for use in forensic settings working with people with ID.

41.1 Introduction

Both ID and ASD are neurodevelopmental disorders listed in the Diagnostic and Statistical Manual DSM-5 [1]. The American Association of Intellectual and Developmental Disabilities defines ID as follows: 'Intellectual disability is characterized by significant limitations both in intellectual functioning and adaptive behavior as expressed in conceptual, social, and practical skills. This disability originates before age 18' [2]. Whereas ASD is characterized by a triad of impairments in social communication, interaction and imagination. The rates of ID in people with ASD are estimated between 41% and 51% [3, 4] with the prevalence of ASD increasing with the greater severity of ID [5]. People with ID and ASD are more likely to suffer comorbid conditions such as mental illness and other neurodevelopmental disorders such as attention deficit and hyperactivity disorder (ADHD) [6–8].

The relationship between the diagnosis of psychopathology and offending is poorly understood in people with ID and ASD, with recent studies taking a wider perspective to understand those with neurodevelopmental disorders and offending, for example, the link between ADHD, conduct disorder [9], or foetal alcohol spectrum disorder [10] to offending. Few studies have examined and compared offenders with ID and ASD to non-offender groups [11] so limiting our understanding of this at risk group in terms of outcomes and development of community-based services to meet their needs. The focus of this chapter is on those offenders with ID and to some extent ASD and their pathway through the criminal justice system (CJS).

41.2 Epidemiology

The worldwide prevalence of ID is estimated to be 1% in high-income countries and higher in middle and lower income countries [12], whilst severe and profound ID is uniformly distributed geographically, irrespective of socio-economic status, mild ID rates are open to variation and are influenced by socio-economic factors [13], parental lifestyle, for example, foetal alcohol syndrome. ASD affects 1% of adults [5]. Often disorders such as ID and ASD will go unrecognized in adults and in high-risk populations such as those in the prison system [14] as is the case with other neurodevelopmental disorders. Historically, mental deficiency (MD), an early term for ID, was put forward by the typological school of criminology. It was thought that hereditary biological inferiority or degeneracy and those with MD made up a significant proportion of the criminal population, particularly Moral Imbeciles [15]. Whilst early studies showed a correlation between intelligence and crime, there was no evidence of a causal relationship. As today, many of the studies were flawed in some way whether due to location bias, a lack of standardized instruments being used and the definition or level of MD. There was in early research the assumption that offenders were biologically different, to non-offenders [16]. ASD and offending have a more recent history, even so, it is often put forward that ASD is overrepresented in offender populations, Aspergers syndrome has been reported at a rates of 1.5% in high secure hospitals in the United Kingdom [17]. For all

substypes of ASD, a review of inpatient services reported a range between 2.4–9.9% [18] and 2.7–8.3% in the dangerous and severe personality disorder unit at Whitemoor Prison [19]. Despite this, there is limited research on the prevalence of ASD and offending and what there is, has often been in highly selective samples, with methodological issues such as uniformity in diagnostic procedures and limited evidence of the reliability and validity of measures in forensic settings [20].

The detection of offenders with ID and ASD has not been a priority across the CJS, which has concentrated on identifying offenders with severe mental illness. The evidence to date indicates that people with ID and ASD may be overrepresented in the CJS [21] and the presence of ASD and ID within the CJS is associated with poorer outcomes [22], increasing the likelihood of future recidivism, or custodial or more restrictive or punitive sentencing [23]. Often there are methodological flaws in studies of offenders with ID although authors acknowledge these limitations. There is a need to place results in context as these are often misquoted or taken out of context for the subpopulations they report. To provide an insight into the prevalence and risk of offending by people with ID whole population studies overtime is required.

Estimates of prevalence of offenders with ID vary across settings. In a study of 237 people in community ID services, 49 had prior contact with the CJS, while the other 188 had histories of anti-social behaviour but no CJS contact. Although most of the offender group were more able, mild ID was not a statistically significant predictor of contact with the CJS [24]. In a larger study of 1326 adults known to ID services in South West England, 7% had contact with the CJS, with 3% having a conviction and 0.8% serving a current sentence [25].

Responses to those committing a crime by the CJS can vary by country and within a country and the interpretation of national and local legislation and policy. A study based in Cambridge, United Kingdom, of 385 adults with ID found that 2% (n = 7) in residential homes or attending day services had been in contact with the police in the preceding year with further action being taken in only one case where the person was cautioned [26]. Whereas in a study in London, United Kingdom, of 180 people identified with an ID, 17 people had contact with the police in the last 12 months, with nine people having been arrested, received a caution, appeared in court or been convicted [27].

Most recent studies of prevalence have been identifying offenders with ID and ASD aided by screening tools aimed at people with ID and ASD and have included studies in court settings. These include the Rapid Assessment of Potential Intellectual Disability (RAPID) [28], Hayes Ability Screening Index (HASI) [29], Learning Disability Screening Questionnaire (LDSQ) [30] and the Kaufman Brief Intelligence Test (K-BIT) [31] for ID and the AQ-10 for ASD [32]. There is very little research into the prevalence of ID and ASD in the court setting and, therefore, varying estimates from 3 to 23% are reported for ID and ASD at this point in this pathway. In a sample of 60 defendants appearing at four Magistrate's Courts in Australia, 2 (3.5%) were estimated to have an ID, with this figure rising to 12% for those with IQ up to 75 [22]. Reporting on studies at six Magistrate's Courts, 208 individuals appearing at court were examined, 47 individuals (23.6%) were labelled ID using the KBIT [31], with another 28 (14.1%) in the borderline range. Those with ID were also more likely to present with mental health problems. Despite differences in IQ assessment, many of those with mild and borderline ID had similar characteristics [33], which were different to defendants/offenders without ID [34]. Given current evidence, it is difficult to estimate the prevalence and it is suggested that many defendants/offenders with ASD and ID are missed at the court stage, with reports of 58% of probation detainees not being identified by police [35].

An international review of 12,000 prisoners estimated prevalence rates for those with ID ranging from 0% to 2.9%. Results were highly dependent on the assessment and criteria used to define ID, for example, in two studies where basic screening techniques were used, rates of ID increased as high as 11.2% [36]. However, many who reach the screening cut-off for ID with borderline intellectual

functioning (BIF) will still suffer a degree of cognitive and social impairment and have unrecognized or unmet needs. This increases their vulnerability as those with BIF will not normally meet eligibility criteria for health or social care services, therefore receiving little support or intervention. The No One Knows reports [37, 38] estimated 7% of prisoners had an IQ of less than 70 with a further 25% had an IQ of below 80. In ASD, the evidence suggests that they have lower rates within the prison population and in some areas, are not overrepresented. A study of referrals to ID offender services reported 10% of referrals had ASD, and showed similar offending patterns of those without, with less contact sex offences, leading the authors to conclude that ASD was not a risk factor for offending [39]. A study of Scottish prisoners estimated the prevalence of ASD at 1% [40]. However, a screening study in a North American male maximum security prison reported rates of 4.4% [41]. In a study of Prisoners in South London, 7.5% [19] were screened for then diagnosed with ID with a further 15 having BIF. The prisoners with ID were more likely to be from a white ethnic background, be under 35 years of age and be single, homeless or unemployed before coming into prison. They were significantly more likely to have mental health problems with 25% have thought about suicide in the last month more likely to be housed in the vulnerable prisoners' wing [42]. In the same study, 10% screened positive for ASD, with 2% meeting diagnostic criteria [43] along with high rates of co-existing ADHD [8, 23].

Screening rarely equates to diagnosis and, as a result, screening studies can be over-inclusive. However, those screening positive and not meeting diagnostic threshold will have similar needs in many cases. In a study of 87 prisoners who screened positive for neurodevelopmental difficulties, compared to neurotypical prisoners, reported neurodevelopmental difficulties group had significantly higher rates of current mental disorder and more likely to have thought about self-harm and suicide in the last month [44].

Higher rates of psychiatric disorders, self-harm and suicide-related thoughts and behaviours in prisoners with ID, ASD and other neurodevelopmental disorders than in those without neurodevelopmental disorders have also been reported in previous studies [45–50]. Psychosis and substance misuse seem to be the common issues, but depression, mania or hypomania, generalized anxiety disorder, social phobia, obsessive compulsive disorder, post-traumatic stress disorder and anti-social personality disorder are quite frequent [44–46].

- Estimates of prevalence of persons with ID and ASD in prison vary across the pathway of forensic services. The estimated international prevalence rates for ID range from 0% to 2.9% and up to 7% in a UK study. For prisoners with autistic traits, the estimated prevalence range is from 1% to 4%.

- Prisoners with ID and/or ASD are more likely to have co-morbid mental disorder, self-harm or suicide-related thoughts and behaviours than those without neurodevelopmental conditions.

41.3 Risk Factors for Offending Behaviour

The Cambridge Delinquency Study, of 411 working class boys living in South London born in 1953, reported several predictor variables of future criminality by the age of 32 including poor socio-economic status characterized by increased poverty, poorer education and academic achievement, higher rates of unemployment and the presence of neurodevelopmental disability such as ADHD [51, 52]. Prediction of future offending was based on a measure of the vulnerability of children aged between 8 and 10, specific predictors within the measure included low family income, large family size (five or more children), a convicted parent, poor child rearing and low non-verbal IQ (90 or less), 46 of 63 (73%) assessed with three or more of these adverse factors in childhood had displayed criminal behaviour by age 32.

One hundred and ninety-two people with ID from the 1953 Swedish birth cohort study were studied retrospectively. The study reported increased rates of offending by women with ID who were four times more likely and men with ID were three times more likely to offend than the rest of the population and 25 more times more likely to commit a violent offence [53]. Similar results were repeated in a Danish population study of over 300,000 people born between 1944 and 1947, women and men with ID admitted to psychiatric facilities were 5.5 and 6.9 times, respectively, more likely to have committed an offence than those who had not been admitted. However, the definition of ID used were unreliable in that it covered those in special school who may or may not have had ID.

Offending rates in females with ID have been reported as compared to the general population [54]. Although referrals to services for offending and anti-social behaviour have been reported at 40% for women, compared to male referrals of just over 60% and at a greater rate than in the general offending literature relating to women [24]. Similar to forensic populations without ID, the severity and types of crimes associated with males and females are different. The Wheeler study found that women with ID were more likely to have committed offences or have convictions against children such as neglect and cruelty. The danger is that a group of women with ID are being criminalized for crimes that would have not been committed if adequate support had been put in place for mother and child. Although females have been included in a limited number of studies, there is a lack of specific research relating to women with autism [55].

One theory of offending often put forward is linked to the characteristics that may be considered to be more associated with ID and ASD and so may predispose a risk for contact with the CJS such as:

- Anxiety, anger and impulsivity.
- Mental health problems.
- Peer pressure and exploitation.
- Social naiveté or misunderstanding of social situations.
- Communication difficulties.
- Change in environment or disruption to routines.
- Lack of capacity to distinguish between right or wrong.
- Lack of empathy.

41.4 Offence Types

Findings from a systematic review [55] suggest that there are relatively few differences in types of offences between the ASD offenders and the non-ASD offenders, aside from lower rates of driving offences and drug offences although there are three types of offences have had a historical association with offenders with ID and ASD, namely, fire-setting behaviour, sex offending behaviour and crimes involving violence.

41.4.1 Fire-Setting

Historically, abnormal psychosexual development, poor impulse control, being of weak mind and being predisposed to crime by having low intelligence have been put forward to explain fire-setting behaviour in adults with ID [56]. An association that ID is predictive of repeat fire-setting along with disorders such as schizophrenia and antisocial personality disorder has also been put forward [57–60]. However, the assumption that individuals with ID are more likely to be firesetters is being challenged with insufficient evidence to support a clear association between ID and fire-setting [54, 61].

In a study across services for people with ID, fire-setting accounted for 2.9% (2/69) of index offences in community settings, 21.4% (15/70) in medium/low secure services and 6.8% (5/73) in high secure services, with those with a previous history of fire-setting accounting for 11.6% (8/69) in community services, 27.1% (19/70) in medium/low secure services and 11% (8/73) in high secure services [62].

Several studies have put forward specific characteristics associated with people with ID who are firesetters. For example, being male, have emotional and behavioural characteristics in childhood related to temperamental difficulties and conduct disorder, increased likelihood of relationship difficulties, introversion, previous psychiatric treatment and a history of self-injury [63].

The Functional analysis theory [64] is widely used and outlines five factors thought to underlie fire-setting:

1. Psychosocial disadvantage.
2. Life dissatisfaction and self-loathing.
3. Social ineffectiveness.
4. Fire experiences.
5. Triggers.

This model is designed to allow clinicians to establish emotional, cognitive and situational antecedents that may predispose, precipitate and perpetuate fire-setting behaviour through a detailed analysis of the behaviour itself [65]. Within treatment, there are several key targets for treatment including communication, anger, poor assertiveness, low self-esteem, social isolation and frustration. One example showing the practical application of cognitive behavioural therapy (CBT) using a single case study of a young man's assessment and treatment of fire-setting is reported [66]. Research has moved from the reporting of case information to intervention-based studies [67]. A case series of six female with a history of fire-setting behaviour, using a CBT approach reported improvement in clinical measures relating to motivations for fire-setting behaviour and no reports of fire-setting 2 years following completion of the programme [68]. In a comparison of firesetters (n = 38) with non-firesetters in a UK forensic ID service, from a sample of 138 patients over 6 years, found fire-setting was associated with psychosis and personality disorders. In terms of criminality, 14 (47%) had a conviction for arson and were more likely to convicted of violent offences [69]. In terms of treatment, this suggests that offence treatment aimed at arson in some cases may be a secondary consideration and whether treatment needs differ from firesetters without intellectual disability [70].

41.4.2 Sex Offending

Like fire-setting behaviour, there is growing evidence that offenders with ID are not over-represented among sex offenders and paraphilics in general [71]. In a New South Wales prison, 3.7% of those with ID had committed a sexual offence compared with 4% of offenders without ID [72]. Increasingly, there are more and more community studies of sex offenders. A 12-year follow-up community Scottish study compared three groups, male sex offenders (N = 121), other male offenders (N = 105) and female offenders (N = 21). In terms of characteristics and propensity to future recidivism, the sex offender group was more likely to be older and has problems with relationships and daily living skills, whilst those most at risk of higher reoffending were male offenders (59%) followed by male sex offenders (23%) and female offenders (19%) over 12-year period [54]. An essential part of treatment for sex offences is often adapted CBT programmes to address the impact of cognitive impairments for ID/ASD offenders to reduce the risk of sexual reoffending [73–75]. More recent studies have suggested that treatment success might be affected by the severity of the offence [76]. A diagnosis of ASD in the past has been associated with worse outcomes and more likely to engage in further sexually abusive behaviour. The SOTSEC-ID project (Sex Offender Treatment Services Collaborative – Intellectual Disabilities) [77] reported on a 1-year cognitive behavioural therapy group, evaluated 13 groups, consisting of 46 men with mild to borderline ID with 'sexually abusive behavior'. Both during the programme and 6 months post intervention, there were no non-sex offences committed reported. Three men (6.5%) were responsible for eight incidents of non-contact sexually abusive behaviour during the project, and at 6-month follow-up, four (8.7%) men were responsible for nine incidents of non-contact sexually abusive behaviours and two incidents of contact sexually abusive behaviour. The three (75%) who were involved in sexually abusive incidents had a diagnosis of ASD with poorer pre-post-

and follow-up scores on measures of cognitive distortions suggesting implications for future treatment and monitoring.

41.4.3 Violence

Crimes involving violence have also had a long association with ID, with widely different rates being reported between studies and settings. Rates of between 20% and 60% have been reported across community and inpatient settings [78, 79]. Gray and colleagues [79], compared a group with ID of 118 (81.4%) men and 27 (18.6%) women, and a non-ID group of 843 (85.6%) men and 153 (15.4%) women. The ID group was reconvicted at a significantly lower rate, with a reconviction rate for violent offences after 2 years of 4.8% compared to the non-ID group of 11.2%, while for general offences, the rate of reconviction was 9.7% for the ID group compared to a rate for the non-ID group of 18.7%. Estimating prevalence rates of offenders with ID committing violence is difficult and can be examined from several different perspectives. A lack of comparison studies involving ID and non-ID offenders makes this direct comparison difficult [80]. Violence has been put forward as the most common offence perpetrated by people with ASD [81]. The motivation and function of violence are varied, and it among the targets for intervention is anger, which can precede violence. A systematic review of two randomised controlled trials (RCTs) and four pre-post unmatched groups suggests that anger management is an effective intervention in this group, a lack of robust methodological controlled studies means more evidence is required to support such a conclusion [82].

There are few differences in types of offences committed by offenders with ID or ASD when compared with other offenders, aside from lower rates of driving and drug offences. The most common offence for those with ID or ASD requiring a therapeutic intervention is violent behaviour.

41.5 Risk Assessment

Risk assessment for offenders with ID and ASD is usually supplemented using standardized tools developed for the wider population. Concerns have been voiced as to whether conventional risk assessment tools can capture the reasons and motivation for offending in these groups. A study of ASD violent offenders ($n = 21$) motivation compared to ASD sex offenders ($n = 12$) reported that the violent offenders were more likely to have severe mental health problems but intellectually more able than the sex offenders. In terms of motivation, six (29%) were motivated by revenge and five (24%) thought to have been motivated by misunderstandings or idiosyncratic ideas, five (24%) were thought to be reactive. Revenge, misunderstanding, rigidity, special interest and idiosyncratic ideas were not identified in the sex offender group, whose offences were found to be instrumental [83]. An individual formulation is often recommended to supplement risk assessment tools [84], for example, with ASD, although conventional risk factors may be present the assessment may not highlight the specific difficulties associated with ASD that might increase the individual's vulnerability to offend [55, 85]. Offenders with ID will share similar risk predictors with non-ID offenders, for example, being young, male, substance abuse, antisocial behaviour, higher unemployment, poorer socio-economic status and histories of abuse [86–88]. Of the 477 people with ID referred to forensic services, 35% (165) had experienced abuse or neglect as a child and started offending at a young age [89].

The shared characteristics between ID and Non-ID offenders have seen increasing use of risk assessment tools validated in non-ID forensic samples. The majority used will support a structured professional judgement such as the HCR-20 (Historical, Clinical, Risk-20 factors) [90] which has been reported as an effective predictor of violence for individuals with ID discharged from medium secure services [79] and a significant predictor across different levels of security (high, medium and low), [62]. In another study of individuals

with ID in high security, 60 people were followed up with the HCR-20 performing better than the Hare Psychopathy Checklist-Revised (PCL-R) [91] in predicting institutional aggression over a year [92]. Although good predictor value has been reported, the usefulness [93] with differences in subscale scores between ID and comparison groups also reported [94]. Risk assessment tools are now incorporated into existing assessment protocols as a matter of course to screen for and offer a more reliable estimate of risk.

There are also many specialist risk tools to measure the likelihood of risk involving specific offences, such as the:

- STATIC-99 [95] which is predictive of sexual risk.
- The Rapid Risk Assessment for Sex Offence Recidivism (RRASOR) [96].
- Assessment of Risk and Manageability of Intellectually Disabled Individuals who Offend Sexually (ARMIDILO-S) [97].

There is less evidence of the effectiveness of specialist tools. In a study of 88 sex offenders divided into two groups (1) ID to borderline intellectual functioning (IQ 70–80) and (2) those with no ID found the ARMIDILO the best predictor for sexual reconviction for those with ID compared to the RRASOR, the Risk Matrix 2000-non sexual Violent (RM2000-V), the Sexual Violence Risk-20 (SVR-20), with the first two measures reported to be no better than chance for the special needs group [98].

There is little in the way of bespoke ASD risk assessment measures. Often cognitive or sensory difficulties in generic risk assessment are not considered. Therefore, the initial assessment should focus on specific difficulties of those with ASD that traditional risk assessment tools find difficult to capture, which may be why people with ASD are detained longer in secure hospitals than other offenders [88]. As a result, the evidence of risk assessment in ASD is less developed.

Consideration to the predisposing, precipitating and maintenance factors for violent and offending behaviour can often focus on what underlies the risk behaviour being assessed and targets for intervention:

- Inability to seek an appropriate course of action in response to perceived or actual difficulties caused by others and reacting to such difficulties by using violence which the individual thinks is appropriate justice. This is exaggerated by a difficulty to seek solutions by meaningful discussions.
- Inability to appreciate social boundaries, and engaging in inappropriate behaviour, sometimes combined with inappropriate sexual or other unusual interests/preoccupations. Misinterpreting events, rules and situations.
- Overriding obsessions, misjudged relationships, innate lack of empathy, lack of ability to recognize fear and difficulties in accepting justice and taking matters into their own hands due to limited social understanding.
- Passive aggression by refusing to cooperate, eat, speak, dress or attend self-care as a way of protest.
- Limited awareness of the consequences of their actions to themselves or others.
- The presence of co-morbidity such as depression, anxiety or other mental disorders [99, 100].

Risk assessment for offenders with ID and ASD who commit violent offences involves using structured professional judgement tools such as the HCR-20 as used in the wider population. The evidence to date indicates that the HCR-20 is an effective predictor of violence in people with ID.

41.6 Services in the United Kingdom

Forensic services for adults with ID or ASD in the UK are made up of both community and secure services. Secure hospital services for people with ID/ASD are divided into three levels of security in the United Kingdom. High secure care for people with ID is provided at the national centre at Rampton Hospital, whilst medium secure [101] and low secure care are provided by both the NHS and

independent sector with the majority of patients being in a low secure unit.

Aside from high security services for ID, the development of less secure services has a common beginning, starting with the Butler report [102] which introduced medium secure services. The Reed report [103] for the first time highlighted the needs of the ID group. The Bradley report put ID on an equal footing with mental health in terms of what providers of services need to consider and called for better cooperation between CJS and health providers to reduce delays between and within services encouraging joined up working and pathways between agencies. In terms of best practice to support people outside of health and CJS services, the Mansell Reports, outline best practice guidance for developing and managing services for people with ID and challenging behaviour in the community [104, 105]. Mansell advocates care within community settings wherever possible with the development and expansion of person-centred services. However, abusive practices and unacceptable standards have been highlighted by several recent inquiries despite a policy of least restrictive practice [106, 107].

The provision of specialized secure/forensic inpatient services for offenders with ASD is a recent development over the last 5 years as historically this group would have resided in services for those with mental illness or ID. A comparison of men with and without ASD in low secure services has found those with ASD being significantly less likely to have a personality disorder or history of drug abuse. One-third had the additional diagnosis of schizophrenia or related psychosis, an indication of their complex presentation within secures hospitals [108]. Differences in ASD and ID offending group have also been noted in other studies. In a study of 138 patients in a forensic ID service [109], the 42 with a diagnosis of ASD were more likely to be detained without restrictions and on a civil section and less likely to have been convicted in the past for violent offences, sex offences and arson. However, in a subset of 114 for whom data were available, the ASD group was more likely to display self-harm and institutional aggression and was significantly more likely to be subject to physical intervention.

An evaluation of completed admissions of offenders (n = 45) vs non-offenders (n = 41) within a London based, assessment and treatment service for adults with intellectual disability found the non-offender group lacked a history of custodial sentences or previous high secure admission but were more likely to be involved in assaults on staff and other patients and to use weapons, whilst the offender group had a higher rate of personality disorder and self-injurious behaviour (SIB) and less likely to be admitted from or be discharged into the community. Another study of 138 patients (77 discharged and 62 inpatients) examined short-term outcomes of a medium secure unit and reported around 90% of patients going to lower levels of security and a third going to community placements (Median length of stay 2.8 years) [101]. In terms of readmission to secure services, a specialist ID service, containing both low and medium secure provision, of 40 male patients, 27 patients (67.5%) were discharged into the community with 14 patients having sole support from the community ID services and 4 from the community forensic services. Of these, 20% were readmitted within the study period and 22.2% received further convictions via the CJS following discharge [110].

A lack of local or regional specialist services offering a high degree of structured community and secure services for people with ID has given rise to out of area placements particularly in the United Kingdom [111, 112] and added to the problem of difficult to discharge patients [101]. Chaplin and colleagues reported on and contrasted the characteristics of 44 people with ID placed out of area compared to those placed locally, near their community [111]. Violence against the person was the most likely event to precipitate and maintain out of area placements.

Women's secure services have developed at a much slower pace, given demand. Facilities are based upon the same model with the omission of women's' high secure services which

have been replaced by enhanced medium secure facilities, with women who offend, generally more likely to be managed at lower levels of security, than men [113].

The move towards treatment and care being delivered in the least restrictive environments has been somewhat thwarted by a lack of appropriate community provision for this group. The emphasis of future service research is likely to focus on recognizing people with ID and/or ASD earlier in the criminal justice pathway and diverting them to more appropriate treatment services if cost-effective. The responses to those committing crime from the criminal justice system vary by country and within countries. The provision of community forensic services is still underdeveloped for offenders with ID and ASD across the United Kingdom, but this is not the case in other countries [114]. Where there are services, deficits in ability, understanding and functioning associated with ID and/or ASD often means lifelong involvement from multiple agencies for those who have contact with the criminal justice system. For many, diversion from prison has meant long periods in hospital. However, there is a tendency that treatment and care are delivered in the least restrictive environments, although currently, the provision of community forensic services is still underdeveloped for offenders with ID and/or ASD, as it in prisons and the entire criminal justice system. Services within prisons and across the entire criminal justice system need to be developed to be effective in meeting both the vulnerabilities and risks of individuals with ID and ASD.

Tip

- Research into offenders with ID has suffered from wide methodological disparities, but there is increasing use of screening tools such as the Rapid Assessment of Potential Intellectual Disability (RAPID) [28], Hayes Ability Screening Index (HASI) [29], Learning Disability Screening Questionnaire (LDSQ) [30] to improve identification across the criminal justice system and so inform future research.
- Risk assessment for offenders with ID and ASD is usually supplemented with standardized tools used in the wider population such as the HCR-20 [90], but in more recent years, specific tools for people with ID have been developed such as Assessment of Risk and Manageability of Intellectually Disabled Individuals who Offend Sexually (ARMIDILO-S) [97].

This chapter has outlined the current evidence relating to the advances in understanding of a number of areas linked to people with ID or ASD who commit offences. These include how the characteristics of the individual are used to understand clinical presentation to inform treatment. There is a move towards least restrictive practice principles in managing this group of offenders. We have also shown that although research has increased, it is still an under researched area when compared to offender research into the wider population. There is a need for further research to include larger population studies which looks at the spectrum of neurodevelopmental disorders.

Key Points

- It is estimated that 7% of prisoners had an IQ of less than 70 with a further 25% had an IQ of below 80.
- There are three types of offences that are commonly reported for offenders with ID and/or ASD, namely, fire-setting behaviour, sex offending behaviour and crimes involving violence.
- Offenders with ID will share similar risk predictors with non-ID offenders, for example, being young, male, substance abuse, anti-social behaviour, higher

unemployment, poorer socio-economic status and histories of abuse.
- Specific risk tools have been developed for use with offenders with ID, but there is little in the way of bespoke ASD risk assessment measures.

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Human Rights

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Contents

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Learning Objectives

- To understand disability as a human rights issue
- To recognize human rights standards related to disability, according to United Nations Convention on the Rights of Persons with Disabilities
- People with disabilities worldwide experience discrimination and inequalities particularly concerning self-determination, participation in society, quality of life, and healthcare provision.
- The United Nations Convention on the Rights of Persons with Disabilities represents a legal, political, and development instrument, with the aim to promote, protect, and ensure the full and equal enjoyment of all human rights and fundamental freedoms by all persons with disabilities.
- It adopts a social concept of disability and guarantees the universal recognition of the intrinsic dignity of every person.
- Human rights provide a robust framework for strengthening the voices of people with disorders of intellectual development.
- The United Nations Convention on the Rights of Persons with Disabilities does not allow exceptions or distinctions based on disability, promoting the adoption of a bio-psycho-social model founded on respect for human rights and more effective social inclusion.
- Persons with disabilities have a central role in monitoring activities, which includes reporting, corrective action, and follow-up.

42.1 Introduction

The United Nations (UN) define human rights as being inherent to all human beings, regardless of race, sex, nationality, ethnicity, language, religion, or any other status. They include the right to life and liberty, freedom from slavery and torture, freedom of opinion and expression, the right to work and education.

They apply from birth until death, irrespective of place of origin, personal beliefs, or how people choose to live their lives.

Human rights may also be regarded as defining the minimal conditions required to lead to worthwhile lives [1], particularly in terms of achieving personal goals. From this perspective, all individuals must have the opportunities to put their ideals and desires into actions and to give meaning to their lives, regardless of whether they have a disability or not.

Nevertheless, throughout history, there has been the tendency to downgrade the value and the dignity of persons with disorders of intellectual development (DID) and subject them to injustices and neglect.

Persons with disabilities and their associations have largely used the concept of human rights to argue for greater equality and for receiving dignified and respectful treatment.

Gewirth [2, 3] states that freedom and well-being are essential conditions for the achievement of goals. In fact, without freedom to act individuals would not be able to achieve their own objectives and in order to act people require a certain level of well-being, both physical and mental, and the access to various material, social, and psychological resources.

Actions by the UN on the issue of the human rights of persons with disabilities have included the UN Standard Rules on the Equalization of Opportunities for Persons with Disabilities [4] and the Convention on the Rights of Persons with Disabilities (CRPD) which was adopted in New York on December 13, 2006 [5]. While the first document essentially represents a set of guidelines, indicating the UN endorsement of persons with disabilities' human rights, the CRPD embodies a significant "turning point" in that it represents a legal, political, and development instrument, elaborated and monitored together with people with disabilities. It adopts a social concept of disability and guarantees the universal recognition of the intrinsic dignity of every person with this condition. The CRPD does not allow exceptions or distinctions based on disability, promoting the

adoption of a bio-psycho-social model founded on respect for human rights and more effective social inclusion.

Tip

The condition of people with specific characteristics depends on a complex combination of bio-psycho-social factors which can be modified on both the social and individual levels. Removing or reducing disability is a responsibility of States and society as a whole.

42.2 Rights of Persons with Disorders of Intellectual Development and the UN Convention on the Rights of Persons with Disabilities

The link between the condition of people with disabilities, including DID, and human rights arises in part from the criticism of the medical model of disability prevalent in the 1970s and 1980s and from the first reflections of the UN, begun in 1981 with the work of the Sub-Commission on the Human Rights of Persons with Disabilities. The CRPD stems from the observation that there were 650 million people with disabilities in the world, 80% living in poor countries that were not able to meet their needs and that worldwide people with disabilities faced barriers in their participation in society and lower standards of living.

It was noted that while the existence and universal nature of previous treaties may have been expected to include people with disabilities, without a specific focus this population was often "legally invisible" in their societies and internationally.

The approval of a convention on human rights represents a significant moment of political and social recognition of the will to recognize, uphold, and protect the rights of the interested parties. A convention sets a new theme within the global and national agenda, creating the conditions for changing policies and legislation. The primary impact concerns governments, parliaments, national and local institutions. Its cultural impact is equally important since it influences the whole society and offers new approaches to social vision that is often attributed to groups subject to human rights violations. This impact can be supported by initiatives, such as public awareness campaigns, involvement of communication media, and appropriate cultural and social mobilization.

In ratifying the Convention, States undertake to comply with its articles through national legislation and policies. The United Nations Conventions prevail over all other forms of legislation, and therefore, the principles and obligations therein must be known and interpreted, in order to guarantee the highest level of protection of human rights at national and local level. In addition to the implications for information and communication systems, the Convention must also have an impact on training in various areas of expertise, encouraging at all levels, the acquisition of knowledge and skills consistent with the aims of the Convention. The Convention, in many ways, represents a unique instrument. It has been developed and monitored together with people with disability. It is a legal and political instrument which considers disability as a matter of human rights, adopting a social concept of disability itself. The Convention ratifies the universal recognition of the inherent dignity of all individuals with disability and does not admit exceptions or distinctions based on their condition. This approach considers people with disability as subjects of rights, able to demand their rights, to make their own decisions about their life, and to actively contribute to society. The general obligations of the Convention include guaranteeing the fulfilment of all the rights for all people with disability, adjusting statutory legislations and policies, and taking the necessary steps to ensure appropriate supports and reasonable compromises (Article 4). Specific obligations include taking effective and appropriate measures to guarantee full legal capacity and necessary supports to exercise it (Article 12), to supply adequate residential and community services (Article 19) as well as educational and

professional opportunities (Article 24, 27), and to provide for physical, psychic, social, and occupational rehabilitation of people with disability (Article 26). Rights guaranteed to persons with disabilities are summarized in the table below (Table 42.1).

The main purpose of conventions is to promote and safeguard human rights all over the world—human rights are for all. They are not new and special rights specifically for people with DID or other particular disabilities.

Table 42.1 Rights guaranteed to persons with disabilities

Article 10: Right to life	Article 21: Freedom of expression and opinion, and access to information
Article 11: Situations of risk and humanitarian emergencies	Article 22: Respect for privacy
Article 12: Equal recognition before the law	Article 23: Respect for home and the family
Article 13: Access to justice	Article 24: Education
Article 14: Liberty and security of the person	Article 25: Health
Article 15: Freedom from torture or cruel, inhuman or degrading treatment or punishment	Article 26: Habilitation and rehabilitation
Article 16: Freedom from exploitation, violence, and abuse	Article 27: Work and employment
Article 17: Protecting the integrity of the person	Article 28: Adequate standard of living and social protection
Article 18: Liberty of movement and nationality	Article 29: Participation in political and public life
Article 19: Living independently and being included in the community	Article 30: Participation in cultural life, recreation, leisure, and sport
Article 20: Personal mobility	

The human and civil rights of all people with DID must be honored and protected but nowadays, as throughout history, have often been unjustifiably limited or denied. These rights include autonomy, dignity, family, justice, life, liberty, equality, self-determination, community participation, property, health, well-being, access to voting, and equality of opportunity and others recognized by law or international declarations, conventions, or standards. The presence of other characteristics such as age, gender, race/ethnicity, sexual orientation, cultural or linguistic diversity, economic status, or severity of disability exposes people with disabilities to increased risk of violations of their rights.

In reality, many individuals, state and local government agencies, and other organizations ignore the human and civil rights of this population. As a result, people with DID face unique challenges, including a history of discrimination, segregation, and exclusion from meaningful choice and participation in society, cultural attitudes of devaluation, and social failure to provide the supports needed for the improvement of participation, equal opportunity, or independent living.

The CRPD promotes the adoption of a bio-psycho-social model founded on respect for human rights and more effective social inclusion. A human rights-based approach to disability implies that persons with disabilities need to participate in all spheres of society on an equal basis with their non-disabled peers.

42.3 Fundamental Principles

The fundamental principles of the CRPD are expressed in article 3 and refer to "respect for inherent dignity, individual autonomy including the freedom to make one's own choices, and independence of persons, non-discrimination, full and effective participation and inclusion in society, respect for difference and acceptance of persons with disabilities as part of human diversity and humanity, equality of opportunity, accessibility, equality between

men and women, respect for the evolving capacities of children with disabilities and for their right to preserve their identities" [5].

42.3.1 Equality, Non-discrimination, and Dignity

The concept of equality alludes to the creation of conditions which ensure full participation and equal opportunities for all persons. This right derives in turn from the principle that diversity (including neurodiversity) represents a characteristic of humankind and a resource for its maintenance and improvement.

In some cases, achieving this goal requires additional adaptations and supports. For example, people with DID might need assistance in making choices and in exercising their legal capacity on equal terms with others [6].

Non-discrimination refers to the fact that rights are assured to everyone without distinction or limitations based on disability or on origin, sex, language, religion, or any other status. It should be noted that article 3 refers to discrimination considered in all its forms.

Inherent dignity refers to the worth of every individual, to his/her innate right to respect and ethical treatment. When the dignity of persons with disabilities is respected, it means that their experiences and opinions are valued [6].

The medical model of disability has led to the dissemination of different approaches and treatments compared to those addressed to other people, developing solutions and interventions that have often generated violations of human rights of persons with disabilities. According to the UN Standard Rules on the Equalization of Opportunities for Persons with Disabilities (v.s.) "persons with disabilities have the right [...] to receive the support they need within the ordinary structures of education, health, employment and social services. [4]" In support of this, States have produced anti-discrimination laws, which also protect people with disabilities, prohibiting any discrimination based on their condition through a legal basis that provides for the removal of discriminations by means of "reasonable accommodation" (Article 5) [5]. Discrimination affects people on the basis of characteristics that are subject to different treatments, prejudices, and barriers to full social participation. When characteristics of gender, race, culture, religion, political opinion, age, and disability condition combine with each other, the result is a multiple discrimination that leads to people who have these characteristics even more vulnerable. A typical example may be represented by women with disabilities, who are subject to severe limitations with respect to access to rights, services, and social participation in respect of relationships, sexuality, childbearing, etc. Disabled people are more likely to experience exclusion, isolation, and discrimination [7, 8] and face a range of other marginalizing factors and there is a universal need for reformulating a new concept of justice for people with disabilities, based on inclusion, empowerment, cultural and social change, respect for human rights, and promotion of human diversity. Exclusion is a process which expresses itself through the denial of opportunities; inclusion is a complex process which requires willingness to "change the rules" and to actively seek feasible solutions.

42.3.2 Autonomy and Self-Determination

Respect for the individual autonomy of persons with disabilities entails that they are enabled to exercise their own decisions about their lives, with support where needed. Applying this principle, for example, to a person with mental disabilities should result in them being offered a range of alternatives for maintenance of their mental health together with the liberty to make decisions on the basis of individual preference.

People with DID have the same right to self-determination as everyone else. They must have opportunities and supports to exercise control over their lives and to advocate on

their own behalf. Nevertheless, many individuals with DID have not been afforded these opportunities and often others make decisions not only about key elements of their lives but also with regard to the everyday activities of daily living. This may be compounded by a lack of experiences that would enable the learning of decision-making skills and making choices, the lack of opportunities impeding participation as valued members of their communities, as well as from making decisions about important aspects of their lives.

People with DID therefore should be given: the assurance that their desires, interests, and preferences will be respected; opportunities to acquire skills which better enable them to make general and specific choices; the lead in decision-making about all aspects of their lives; the opportunity to self-direct their own supports and services and allocate available resources [9].

42.3.3 Independent Life and Social Inclusion

All people with DID benefit from full inclusion and participation in society. Nevertheless, they are not treated equally and often they are separated from their community and labeled as having "a disability," experiencing several forms of exclusion. Negative views can often be encountered about the economic costs of inclusion particularly where public resources are severely constrained in low- and middle-income countries. Morgon et al. [10] in their review of studies describe "pathways" to economic impact of exclusion and inclusion in the areas of education, work and employment, and health concluding that exclusion can lead to a significant economic burden for individuals and their families and also to society as a whole. Such evidence may be helpful when advocating for inclusion in national or local policy or strategy; however, it must be understood that inclusion is a human right and, as with other rights, does not have to be justified.

Inclusion is a right based on the full participation of persons with disabilities in all the fields of life, without discriminations, respecting the dignity and giving value to human diversity through appropriate interventions.

Article 19 of the Convention [5] sets out the right of all disabled people to "live in the community, with choices equal to others," requiring States to enable disabled people to be fully included and participate in society. According to the text of the article:

(a) Persons with disabilities have the opportunity to choose their place of residence and where and with whom they live on an equal basis with others and are not obliged to live in a particular living arrangement.
(b) Persons with disabilities have access to a range of in-home, residential, and other community support services, including personal assistance necessary to support living and inclusion in the community, and to prevent isolation or segregation from the community.
(c) Community services and facilities for the general population are available on an equal basis to persons with disabilities and are responsive to their needs.

People with disabilities should be included in all aspects of our society and receive the supports they need to participate actively in community life. They should have the opportunity to have relationships with other individuals in the community, live in a home where and with whom they choose, have access to the supports that they need, engage in meaningful work in an inclusive setting, and enjoy the same leisure activities that are available to the general public [9].

For participation and inclusion to be achieved, public and private dimensions of society must be organized in order to enable all person to take part effectively and to be recognized as equal participants; full and effective inclusion is achievable only through accessible physical and social environments.

According to Article 26: *States Parties shall take effective and appropriate measures, including through peer support, to enable per-*

sons with disabilities to attain and maintain maximum independence, full physical, mental, social and vocational ability, and full inclusion and participation in all aspects of life.

This necessitates the provision of comprehensive services and programs for habilitation and rehabilitation particularly in the areas of health, employment, education, and social services, and ensuring that these:

(a) *Begin at the earliest possible stage, and are based on the multidisciplinary assessment of individual needs and strengths;*
(b) *Support participation and inclusion in the community and all aspects of society, are voluntary, and are available to persons with disabilities as close as possible to their own communities, including in rural areas*

Article 26 also emphasizes the promotion of training for professional staff in such services and also the availability knowledge and use of assistive technologies [5].

Article 3 identifies key principles of a human rights approach to disability, including respect for inherent dignity, individual autonomy, non-discrimination, inclusion in society. Fundamental principles are very important for interpreting the rights and other articles in the Convention, and to assist States in understanding and implementing its provisions effectively.

42.3.4 Quality of Life

People with DID are entitled to a life of quality but they often do not have the adequate supports and personal relationships they need or desire in order to achieve this[1]. A meaningful quality of life exists when they receive, at all stages of their lives, the support, opportunity, and resources to explore and define how they want to live, choose the services and supports they need, have a life enriched by friends and family and have opportunities for intimate relationships, experience life-long learning and develop decision-making skills, and enjoy the same rights and respect for their dignity and privacy, as do people without disabilities [9]. Hence, quality of life and the preservation of human rights are closely related.

In recent years, increased research attention has been devoted to person-centered outcomes and quality of life. The latter in particular has repeatedly been defined an essential reference in the intervention planning and evaluation toward DID, where the recovering of neuropsychic functions cannot be pursued as in other mental or physical conditions in the general population. Evaluation of quality of life aims to identify how the individual might achieve a satisfying life and experience participation, opportunities, and the possibility of making independent choices. It is a multidimensional concept, involving several definitions and applications [11–13]. There is general agreement on the fact that Quality of Life measurement should be based on both qualitative and quantitative variables [12, 14], from both subjective and objective positions [15]. The Quality of Life model is a useful conceptual parameter in maintaining the life aims of people with DID aligned with those of other non-disabled people. The model has been shown to have a number of positive implications for care and intervention for those with neurodevelopmental disorders, including caregiver burden, supports, health services, and health policies [16]. There remain still many challenges in reaching an optimal implementation of the concept of Quality of Life into daily practices, such as matching with the non-reductionistic approach of the phenomenological tradition and the very specific cognitive impairment.

There is a close relationship between the quality of life domains and the articles of the Convention. Focusing on enhancing quality of life and human rights encourages organizations to provide individualized supports and involve people with DID in the decision-making of their own lives.

1 For further reflections on Quality of Life in people with DID, see ▶ Chaps. 14 and 15 on this volume.

42.4 Access and Quality of Physical and Mental Health Care

People with DID have higher rates of morbidity and mortality than the general population. Cooper and colleagues [17] in looking at multiple morbidity in those with DID concluded not only is the greater burden, occurring at a much earlier age, with a different health profile but also that specific initiatives are required in order to reduce inequalities.

The National Learning Disability Mortality Review program (LeDeR) [18] in reviewing the deaths of 1081 people with a learning disability aged 4 years and over between 2016 and 2018 recorded a median age of death for males with a learning disability (intellectual disability) of 60 years compared with 83 for men in the general population and median age of death for females of 59 years compared with 86 for females in the general population.

Yet more disturbing was the finding that 15 people had their cause of death recorded as "learning disabilities," and the term "learning disabilities" or "Down's syndrome" had been recorded as the reason for a Do Not Attempt Cardiopulmonary Resuscitation order.

The LeDeR report concluded that 71 adults had been subject to failings in care that was far short of good practice and that had had a significant impact on their well-being or been a direct contributing factor to their cause of death.

It might be inferred that such discrimination and inequalities in health could be addressed by bringing a human rights-based approach in the pathways of access to healthcare provision.

A government led initiative, The Human Rights in Healthcare Programme was established in England in 2005 to embed human rights into public services. The Programme was a collaboration between the Department of Health, the British Institute of Human Rights, and, over time, by eight National Health Service (NHS) trusts.

The program defined five principles, known as the PANEL principles, as being fundamental to a human rights-based approach:

- People's right to participate in decisions that affect their lives;
- Accountability of duty-bearers to rights-holders;
- Non-discrimination and prioritization of vulnerable groups;
- Empowerment of rights-holders;
- Legality: the express application of the 1998 Human Rights Act (UK).

A review of the impact of this program by Dyer [19] concluded that the Human Rights Act in the United Kingdom, which attempted to enshrine the European Convention on Human Rights into UK law, had not resulted in the anticipated cultural change in health services. While there was evidence that the development of human rights-based resources could have a beneficial effect, the program had not addressed the "big human rights questions about health care" nor adequately engaged a multidisciplinary approach.

In respect to mental health, which is the object of the present textbook, the right for care (Article 25) of people with DID is still challenged by many issues in most countries across the world.

The World Health Organization recently carried out a project addressing mental health, human rights, and standards of care in institutions across the European Region [20]. Phase 1 was comprised of a survey of institutions that provide long-term care for adults with psychosocial and intellectual disabilities across 31 participating countries. In Phase 2, observance of human rights was assessed in a sample of institutions using World Health Organization Quality Rights toolkit. This toolkit has 25 standards within five interrelated themes referenced to related article of the CRPD:

- Theme 1. The right to an adequate standard of living (Article 28 of the CRPD);
- Theme 2. The right to enjoyment of the highest attainable standard of physical and mental health (Article 25 of the CRPD);

- Theme 3. The right to exercise legal capacity and the right to personal liberty and the security of person (Articles 12 and 14 of the CRPD);
- Theme 4. Freedom from torture or cruel, inhuman or degrading treatment or punishment and from exploitation, violence, and abuse (Articles 15 and 16 of the CRPD);
- Theme 5. The right to live independently and be included in the community (Article 19 of the CRPD).

Assessments were carried out by government nominated national assessment teams in 75 facilities across 24 World Health Organization Member States and also Kosovo.

The findings showed that long-term institutional care for people with psychosocial and intellectual disabilities is significantly below standard in many European countries and there were many examples of violations of people's fundamental rights including their legal capacity, autonomy, dignity, liberty and security of person, physical and mental integrity, and freedom from torture and ill treatment and from exploitation, violence, and abuse. Concerns were also raised about use of mechanical and pharmacological restraints to manage difficult behavior, a "culture of impunity with regard to reported cases of sexual abuse" and "numerous irregularities concerning informed consent, discrimination and barriers to access to high-quality care for general and reproductive health, lack of alternative or complementary mental health treatment options and a general lack of opportunities for meaningful daily activities within or outside the institutions."

The clinical approach to people with DID still largely neglects the therapeutic relationship, diagnostic evaluation, and outcome measures of the interventions, and many people often encounter difficulties in receiving adequate assistance. Literature shows a considerable amount of evidence on the lack of specific services for people with DID, particularly for those who suffer from additional mental health problems. In fact, various studies have pointed out that to date the health needs of people with DID remain largely unsatisfied, both in qualitative and quantitative terms. At the European level, the greatest difficulties have been identified in relation to access to treatment, diagnostic evaluations, psychopharmacological prescriptions, treatment of psychiatric disorders in comorbidity, and reception and support offered. Particularly important is the difficulty that clinicians may face in detecting and evaluating symptoms in people with DID, due to communication problems, poor quality of informant accounts, and the diagnostic overshadowing of disability. This and poor quality of care in the choice of interventions are mainly due to the lack of specific training and often linked, in turn, to the presence of prejudices and beliefs that further reduce the effectiveness of practices. The high psychiatric needs of people with DID are not adequately addressed by mental health services and delays in detection, assessment, and treatment often lead to a progressive deterioration of psychological and behavioral conditions. Furthermore, there is an excessive prescription of antipsychotics, neuroleptics, and psycholeptics [21]. Literature indicates that some pharmacological treatments have been shown to be effective on certain behaviors, but it is not possible to define specific therapeutic relationships [22].[2] Although their efficacy has not been proven, mood stabilizers, neuroleptics and benzodiazepines, as well as selective serotonin reuptake inhibitors, and new generation antipsychotics, are often prescribed. Polypharmacy is also common in people with disabilities even if it is not supported by evidence. Great attention must be paid to the ethical implications of drug treatments in people with DID. Research in this field has been severely limited by ethical concerns related to the inability of people with most severe DID to consent to participate in randomized and controlled trials for treatments. People with DID end up being deprived of the right to receive therapies based on strong evidence and being subject to unwanted effects of medications prescribed for unjustifi-

2 For further reflections on psychopharmacology in people with DID, see ▶ Chap. 11 on this volume.

able reasons constitutes infringements of article 25 (and on occasions article 15) of the CRPD.

To date, health needs of people with DID remain largely unsatisfied, both in qualitative and quantitative terms. Discrimination and inequalities could be addressed by bringing a human rights-based approach in the pathways of access to healthcare provision and improving specific trainings for healthcare professionals.

42.5 Monitoring the Rights of Persons with Disabilities: An Overview

When monitoring the rights of people with DID, it is essential to start communicating directly with the person, if necessary through the use of augmented communication, tools, or support persons. The frequent exhortation from the disability activism of the 1990s of "nothing about us without us" is a cry for not only inclusion but also empowerment and enablement in being active agents in the determination of the quality of their own lives.

Monitoring the rights of people with disabilities requires us to ask what society has or has not done which is impeding the full enjoyment of their rights. For example, when monitoring the right to education, recognizing that negative attitudes and prejudice or inaccessible schools may underlie the person being unable to enjoy the rights to education [6].

Monitoring processes should be in place in order to evaluate whether States are respecting their obligations in relation to the rights set out in the Convention; the use of the World Health Organization Quality Rights toolkit quoted above is one example. Monitors should ensure that they understand the Convention's relevant articles and apply the "respect/protect/fulfill" framework accordingly [6].

Comprehensive and effective monitoring includes reporting, corrective action, and follow-up. There should be a process of assurance that the monitoring report has provided a complete and adequate summary together with clarity of recommendations and responsibilities for subsequent corrective action.

It is fundamental to bear in mind the central role of persons with disabilities in monitoring activities. The Convention states that this role must be continuous, requiring States parties to "closely consult with" and "actively involve" persons with disabilities in decision-making processes related to them (Article 4). According to article 33 of the Convention, "civil society, in particular persons with disabilities and their representative organizations, shall be involved and participate fully in the monitoring process" [5]. Besides being included, opinions and experiences of persons with disabilities must be central in the monitoring process since they are the experts on their own situation. It is also important to ensure that the monitoring of the rights of persons with disabilities involves all persons regardless of their types of disabilities and from all socio-economic and ethnic backgrounds or age groups.

Tip

Monitoring process should not strengthen an approach that considers persons with disabilities as intrinsically different from others. The *voice* of *people with disabilities* produces important reflections in the field of human rights protection and significant examples of monitoring disability policies.

42.6 Conclusion

Due regard to human rights highlights the fact that people with disabilities too often experience segregation and social exclusion in society, they are subject to discrimination, and in mental healthcare are in receipt of differentiated, poor quality, and degrading treatment. There is a pervasive violation of their human rights.

The protection of human rights is linked not only to respect for individual freedoms but also to the cultural and social construction of inclusive societies, where prejudices

and barriers are eliminated and everyone can live without his personal characteristics being stigmatized. This opens up a new field of cultural and political action that involves society as a whole: a way of including human diversity within societies and within models of economic and social development. The specific theme of the reformulation of the cultural and social vision of disability becomes a paradigmatic example for the comparison with ideologies that have become "common sense," built on secular practices of segregation and exclusion [23]. The new social universe must be based on a pervasive approach to the construction of environments, goods, and services, on the basis of equal opportunities for all citizens and elimination of any form of discrimination.

Key Points

- The adoption of the UN Convention on the Rights of Persons with Disabilities reinforced a key paradigm shift with the move from the medical model to the social and rights-based model
- A human rights-based approach ensures that rights such as dignity, non-discrimination, participation and inclusion, respect for difference, equality of opportunity, and accessibility are upheld and secured for all.

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Teleassistance and Telerehabilitation: COVID-19, Other Epidemic Situations and Future Prospects

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Learning Objectives

This chapter's primary objective is to provide information on the use of telehealth, specifically teleassistance and telerehabilitation, for PwID/ASD in a condition of epidemic emergency and in general. Most frequent TA and TR activities are reviewed, together with their prerequisites, ways of use, objectives, efficacy, and limits.

43.1 COVID-19 Outbreak and Associated Vulnerability and Support Needs for People with Intellectual Disability and Autism Spectrum Disorder with High and Very High Support Needs

Intellectual disability (ID) and autism spectrum disorder with high support needs (ASD-HSN) represent the two neurodevelopmental disorders with the greatest negative impact on communication skills, self-insight skills, and individual functioning. Furthermore, these conditions have a high co-occurrence of physical and psychiatric disorders [1], significantly higher and with earlier onset than the general population. They can often be found together in the same individuals [2, 3], thereby causing an even more complex vulnerability [1].

People with ID and/or ASD-HSN (PwID/ASD-HSN) belong to one of the populations most vulnerable to the COVID-19 epidemic and related stressors due to several factors: multimorbidity, low levels of health literacy, the difficulty in following complex hygiene rules, the high need to maintain daily routines, and the dependence on other subjects for their health care.

Physical multimorbidity includes endocrine diseases, hypertension, respiratory problems, cancer, and other conditions associated with a higher risk of severe acute respiratory distress syndrome and other COVID-19 (SARS-CoV-2) complications.

Research on previous respiratory viral epidemics, including influenza A virus (H1N1) and respiratory syncytial virus (RSV), indicates that people with genetic syndromes counting ID and ASD-HSN are more likely to develop complications and need hospitalization than the general population [4–6].

A preliminary study about COVID-19 sequelae risk rate was initiated in mid-March by the Italian Society for Neurodevelopmental Disorders (SIDiN) in reference to the whole Italian territory.

The data collected so far concern 660 cases of only six regions (Tuscany, Lombardy, Emilia-Romagna, Piedmont, Campania, and Sicily); data indicate the presence of severe-critical symptoms in a much higher percentage than the general population (19–51% vs. 15.6%). Variability mainly depends on the epidemic area and housing condition.

Another recent study conducted mainly on the USA territory through the TriNetX COVID-19 Research Network platform has contributed to validate the notion that COVID-19 can present a greater risk to people with intellectual and developmental disabilities [7]. The study has been conducted to explore the trends in comorbidities, number of cases and number of deaths, and case-fatality rates among patients with intellectual and developmental disabilities who received a diagnosis of COVID-19 through May 2020. The results have shown that this population had a higher prevalence of specific comorbidities associated with poorer COVID-19 sequelae. Moreover, distinct age-related differences in COVID-19 trends were reported, with a higher concentration of COVID-19 cases at younger ages [7].

For what concerns psychiatric vulnerability, PwID/ASD-HSN have a very high rate of psychiatric disorders, with a lifetime prevalence of up to 44% or even higher when the two conditions are associated. Anxiety and affective disorders are the most frequent disorders. The disorders' symptoms are often quite different from the general population, especially in people with low or absent verbal communication skills, who express themselves only by problem behavior [8]. Therefore, the comorbidity of unidentified psychiatric disorders is also very high in PwID/ASD-HSN, with prevalence rates estimated above 50%, even in specialized support settings.

Because of difficulties in understanding and communicating, the high need for routine and for predictability, low adaptability, and the probability that the environmental and social changes associated with the pandemic may cause psychological distress, or even psychopathological problems in PwID/ASD-HSN is very high. Previous epidemic studies indicate that the drastic changes in the daily routine (e.g., restriction or interruption of regular activities, places, and interpersonal contacts), forced coexistence, loss of support, and extended perception of loneliness can determine regression, loss of ability, psychological distress, and physical disturbances in these subjects [9–12]. Furthermore, the risks associated with the epidemic may also relate to the appearance or aggravation of problem behaviors, such as stereotypies, self-stimulation, self-injurious, aggressive behaviors toward objects or other people, and oppositional behaviors. The mere reduction of interpersonal contact can exacerbate problem behaviors [13]. The high prevalence of problem behaviors in PwID/ASD-HSN has been associated with a proportionate frequency of psychiatric disorders [14–19]. In fact, in PwID/ASD-HSN with severe cognitive and communication difficulties, psychiatric symptoms are expressed with significant behavioral changes [20–22], including the presence or the aggravation of problem behaviors. Obviously, problem behaviors can also be related to many other factors, such as physical pain or unspecific emotional reactions to stress.

The symptoms of mental suffering related to the COVID-19 epidemic during the lockdown phase were the subject of a survey of the Italian Society for Neurodevelopmental Disorders (SIDiN) conducted throughout the national territory since April 2020. Two adaptations of an online questionnaire (short and easy to complete) were anonymously submitted to caregivers and family members of persons with ID living with family or in small apartments in the community, and to operators of social-health residential services for PwID/ASD. In total, 56.9% of caregivers/family members who answered the questionnaire referred to persons with co-occurrence of ID and ASD, 27.7% to PwID, and 15.4% to PwASD. On the other hand, most healthcare professionals referred to PwID (66%), 30% to people with co-occurrence of ID and ASD-HSN, and only 4% to PwASD.

Preliminary results on persons living with family or in small apartments in the community indicate an increase in agitation (53.8% of the sample), irritability (52.3%), sleep disturbances (54%), increased appetite (43.1%), stereotypies (43.1%), soliloquy (30.8%), and social withdrawal (27.7%). Moreover, 29.2% needed the activation or modification of psychopharmacological treatment for the management of problem behaviors. This data support experts' opinions, which were already being expressed since the early days of the mass quarantine on a risk increase of behavioral and mental issues [23, 24].

Preliminary results on persons living in social-health residential services indicate an increase in irritability in 21.4% of the sample and agitation in 14.5%. Other behavioral changes were present in less than 14% of the sample. Many operators declared that significant changes were not observed in about 50% of cases during the mass quarantine in the free annotations section of the questionnaire. In total, 93.9% of the cases did not require the activation or modification of pharmacological interventions.

In general, the behavioral changes detected could reflect an increase in anxiety associated with psychopathological and non-psychopathological syndromes. In fact, irritability, psycho-motor agitation, and stereotypies increase have been listed among the typical behavioral equivalents of anxiety in PwID/ASD-HSN [8]. Many behavioral changes associated with affective disorders, such as increased closure to others, changes in appetite, and insomnia were also found in the sample of people living in families or small apartments in the community [8, 25]. These alterations could be related to the loss of important activities – before the mass quarantine, 52.3% of the sample attended day centers and 75.4% recreational associations.

PwID/ASD-HSN families have a greater vulnerability to distress compared with families of the general population [26, 27], especially if the PwID/ASD-HSN has entered adulthood since a long time [28].

These are often single-parent families and/or with elderly parents and/or with autistic traits [29]. These parents find it challenging to offer social support to their sons, have low coping strategies, and present a high risk of developing affective disorders [30, 31].

Problem behaviors, interpersonal conflicts or tensions, and lack of social networks are frequent even in ordinary life and negatively impact the quality of PwID/ASD-HSN's family relationships.

For all these reasons, remote assistance could usefully provide various support needs, especially through their integration, as far as possible, with traditional interventions. During the COVID-19 emergency, for the purpose of health protection, methodologically rigorous and easy-to-use interventions through the use of technology were requested.

People with ID and/or ASD-HSN belong to one of the populations most vulnerable to epidemics and related stressors. Risks may also relate to the appearance or aggravation of problem behaviors, such as stereotypies, self-stimulation, self-injurious, aggressive toward objects or other people, and oppositional behaviors. Problem behaviors can reflect an increase in anxiety or mood alteration associated with psychopathological syndromes.

43.2 Care Needs Manageable Through Telemedicine Services

Persons with developmental disabilities represent potential users of telemedicine services together with persons suffering from psychiatric disorders, chronic diseases, rare diseases requiring constant contact with healthcare structures and health practitioners, people who need special non-deferrable supports, including psychological supports, or people who require long-term treatments that are usually managed by territorial services or residential structures.

During the 2020 COVID-19 health emergency, service providers had to manage the activation and continuous implementation of interventions for many PwID/ASD-HSN in quarantine or fiduciary isolation. These interventions were necessary to guarantee continuity of care even during the emergency period due to the pathological conditions and the frailties described. All these needs have been addressed in sanitary emergency conditions, with a lack of material and human resources and the extreme obligation to respect the anti-contagion rules for the protection of staff, PwID/ASD-HSN, and their families. Therefore, remote interventions were indispensable [32]. Many leading technical-scientific bodies of different countries have indicated that these telehealth services must be provided primarily through updated digital and telecommunication technologies that can ensure more appropriate support opportunities than the previous technologies.

The realization of remote interventions requires first to consider the relational needs of people with disabilities with the social-educational-health system.

The isolation within one's home or the inability to access services in person makes it particularly desirable to be able to count on a service that can be easily used remotely, being able to quickly access the interview with health professionals and educators, as needed.

PwID/ASD-HSN and/or his family or caregivers may expect to receive the solution of their problem already through the first telematic contact or at least to perceive the concrete possibility of being assisted effectively and safely for any need. In case these expectations are not met, they will tend not to trust the proposed system and not to use it, especially when they are faced with urgent needs.

This kind of relationship dynamics between users' needs and resources offered are very frequent in all assistance and health services; in remote services, they are even more intense due to the interposition of technologies and related relational limits, including the possible difficulty in the use of technologies by PwID/ASD-HSN and their families.

It is essential to set up the remote service by analyzing the different possible situations of its use and the different types of needs of the persons to whom the service is addressed.

The literature indicates a higher possibility of direct fruition by people who have residual cognitive and communicative skills, while people with higher levels of impairment require a constant mediator/assistant's support [33, 34].

In order to counter the spread of the infection and to monitor any clinical aggravations related to COVID-19, the national health system of many countries has identified four types of people for whom it was necessary to provide isolation and home health checks:

1. Asymptomatic person who had contact with a positive COVID-19 case (quarantine up to 14 days from the last contact with the positive case)
2. Paucisymptomatic person who had contact with a positive COVID-19 case, with a negative COVID-19 test (isolation up to 14 days from the last contact with the case)
3. Paucisymptomatic person with positive COVID-19 test (isolation until tests become negative and symptoms disappear)
4. Person discharged from the hospital as clinically healed but still positive to SARS-CoV-2

These epidemic contrast actions can be performed by health services that operate in telemedicine with numerous advantages, such as (a) monitoring at home, the evolution of clinical and health conditions of suspected asymptomatic subjects or subjects presenting mild or moderate symptoms of illness; (b) considerably contain the risk of contagion even in situations where the quantity of positivity tests for COVID-19 is insufficient; and (c) reduce any difficulties of the health system.

Telemedicine interventions may be appropriate for PwID/ASD to manage some care needs related to their condition (impairment and disability), physical and psychological co-occurring problems, including symptoms of an epidemic disease and for the limitation of contacts and access to services determined by the epidemic emergency. However, it is important to clarify that telemedicine should not be used to perform home medical treatments for people with severe illnesses who instead require intensive hospital care.

During an epidemic emergency, telemedicine can be appropriate for PwID/ASD to manage several health, rehabilitation, and assistance needs, in respect to both the prevention and treatment of the epidemic illness and the continuity of care necessary for the condition of developmental disability and co-occurrent physical or mental disorders.

43.3 General Objectives of Teleassistance and Telerehabilitation Services

In telemedicine, although innovative, technology is used properly only when it allows progress in healthcare practice. During an epidemic emergency, the main objective of teleassistance (TA) and telerehabilitation (TR) services for PwID/ASD is to provide health, rehabilitation, and assistance interventions to persons in isolation in order to proactively monitor their health conditions concerning both the prevention and treatment of the epidemic illness and the continuity of care necessary for the condition of developmental disability and associated disorders.

This general objective is useful for directing organizational actions within the framework of evidence-based methods, which can also facilitate implementations and optimization of service delivery. However, further aims have to be taken into account to meet every service user's specific needs. Programming telemedicine services requires to identify diagnostic, therapeutic, educational, and care practices appropriately that can be carried out remotely, through technologies that are available and usable by persons with different levels of disability.

The Health and Socio-Educational Team (HSET) in charge of treatment must choose from time to time what setting and technology are the safest and effective for the person to care. It should not be thought that PwID/ASD-HSN can find in technology, however advanced, a unique solution to their problems. People can achieve concrete improvements through technology only if used appropriately within a clinical approach

aimed at the care of the individual and not at the use of technology [32].

TA and TR objective must be aligned with the more general objective of every individualized therapeutic and rehabilitation plan, which is to promote and favor the quality of life of the PwID/ASD-HSN.

The individualization of the objectives, tools, and methods of service delivering should be based on a generic (whole-person) Quality of Life model, both in reference to the person with ID/ASD and their family. Most comprehensive Quality of Life models and tools are those that envisage: (a) a broad articulation of the area of Being/Well-Being, which includes spirituality; (b) a multidimensional assessment of individuality, including at least the dimensions of importance and satisfaction; and (c) an integration of direct assessments on the PwID/ASD, when possible, with assessments carried out through external informants (at least two and at least one of which should be represented by a family member) [35].

Generic quality of life represents the best framework for defining and sharing therapeutic and rehabilitative goals by technicians, PwID/ASD, family members, and other significant figures, upon which to establish a good therapeutic alliance and through which to promote good compliance with interventions.

TA and TR objective must be aligned with the more general aim of every individualized therapeutic and rehabilitation plan, which is to promote and favor the quality of life of the PwID/ASD.

43.4 General Principles for Teleassistance and Telerehabilitation Services Implementation

Teleassistance (TA) is a support modality, operated remotely by professionals with direct or indirect interaction on the local system. Direct interaction occurs when the assistant can directly operate in full autonomy with the PwID/ASD, sending instructions, indications, or comments and analyzing their responses without any local intermediation. This type of remote assistance is possible only with PwID/ASD who have fair cognitive and communication skills and are able to use technological devices. Productivity is high in the direct interaction setting, as the duration is limited to the time strictly necessary to carry out required tasks.

TA with indirect interaction takes place with the intermediation of a subject on the local side, which can be represented by a family member, a habitual caregiver, or a technician, depending on the type of assistance requested. For indirect TA, the remote assistant needs good technical and relational skills to correctly provide and interpret instructions even in the presence of technical problems. It is worth underlining that this modality of interaction is often preferred by users, as the human presence is perceived as an added value. In the indirect interaction setting, productivity can be significantly lower than in the direct one, due to the non-determinability of the time to spend with the remote assistant.

Telerehabilitation (TR) is a form of telemedicine that allows providing remote rehabilitation services directly at the patient's home or in another place chosen by them. TR uses different types of telecommunication technologies, including telephone, video, websites, and computer programs, to support the patient in the type of rehabilitation required. In some cases, virtual reality and robotics are also used [36].

TR activities include psychological exercises, learning tasks, physical movements, and behavioral techniques, which are defined by the HSET based on individual needs and carried out through most suitable devices, software, and IT applications. These applications are often integrated with video cameras that allow the HSET to monitor patients' health conditions and progress. TR allows the continuity of some positive characteristics of the normal educational-therapeutic relationship. The patient can continue to receive feedbacks for their needs and to achieve positive treatment outcomes, and the HSET can continue to use a personalized approach to care, adapting structures and interventions to the needs and abilities of the user.

In case the HSET needs to choose between different care and rehabilitation options (i.e., getting on with home care or hospitalizing the patient after an episode of severe self-injury or aggression toward family members), the following relevant technological aspects should be considered:

- Preliminary conditions for making telemedicine service possible
- Health responsibility during telemedicine activities
- List of the elements and tools necessary to carry out home services
- Effectiveness of telemedicine services
- Initial steps to activate the service

Further information on the correct use and problem management of digital and telecommunication technologies is available on the documents produced by experts in the field [37, 38].

TA and TR are operated remotely by professionals with direct or indirect interaction with the PwID/ASD. Direct interaction is possible only with PwID/ASD who have fair cognitive and communication skills and are able to use technological devices. Indirect interaction takes place with the intermediation of a subject on the local side, which can be represented by a family member, a habitual caregiver, or a technician. TR uses different types of telecommunication technologies, including telephone, video, websites, computer programs, virtual reality, and robotics. TR activities include psychological exercises, learning tasks, physical movements, and behavioral techniques, which are defined by the HSET based on individual needs and carried out through most suitable devices, software, and IT applications.

43.5 Effectiveness and Limits of Teleassistance and Telerehabilitation

Studies on TA and TR's efficacy for PwID/ASD-HSN are limited in quantity and quality, especially concerning adulthood. Most of the studies are represented by case reports or case series, only a few relatively large-scale cohort studies have been produced. No randomized and/or controlled studies were found. Almost all the studies used pre-post evaluation, questionnaires, and interviews to determine interventions outcome. No comparisons were made with traditional "face-to-face" interventions.

More than 90% of the studies used the satisfaction of service users (people with disabilities, family members, other habitual caregivers, professional assistants, clinicians who assist the person with disabilities in the use of the service, etc.) as the only outcome measure. Other outcome measures were also used in some studies based on specific objectives, including physical functioning, mental state, communication skills, self-care skills, cost and time savings, duration of service, number of hospital admissions, and objectives achievement. The duration of the interventions varies across different studies from a few hours to 2 years.

Most of the available studies report a high level of satisfaction from professionals, family members, and other caregivers [39–42]. One of the main positive factors has been identified in the possibility of avoiding travels to reach face-to-face services, often requiring complex organization, personnel, and equipment, and identified as a cause of distress and/or problem behaviors in many PwID/ASD-HSN.

TR showed efficacy in improving cognitive and adaptive skills in children and adults with ID and/or multiple disabilities [41, 43]. In collaborative occupational therapy with parents of children with ASD, it was able to increase carryover of home programs by providing opportunities for parents to ask questions, review sensory techniques, and understand the therapist's clinical reasoning [44]. In aging persons with Down's syndrome, it has been usefully inserted in programs for the prevention of cognitive decay [45].

TA has been found to be quick to obtain and easy to perform, families often receive rapid reassurance with respect to doubts of symptoms or prompt advice for the management of problem behaviors [39, 40]. This helps

family members and caregivers to become more confident in their ability to independently determine the severity of various difficult situations and when professional intervention is needed. Many family members and caregivers have shown good skills in using the technology for TA and TR or good learning skills of how to use them [39].

When preceded by face-to-face interventions (as in the case of normal rehabilitation practice before a mass lockdown), TA and TR interventions showed the ability to maintain a good relationship with the HSET and to keep on working on simple rehabilitation targets, both directly with the PwID/ASD and through the mediation of their caregiver as well as to provide useful suggestions and advice for the management of ordinary and extraordinary activities or critical episodes [33, 46].

Especially with adult users, family members have often moved from a position of delegation and dependence with respect to professional figures to a proactive position, discovering new empowerment skills in the relationship with the PwID/ASD, expanding the range of educational strategies, and promoting positive and collaborative interactions. This active participation of the parent in the definition of the enabling objectives for the PwID/ASD and of the intervention projects to achieve them has often allowed him/her to understand their importance better, renewing and strengthening the alliance with the operators. However, it has also fostered a wider capacity for recognizing and enhancing the characteristics and potential of the PwID/ASD-HSN, mitigating concerns for the future. Regular video calls have been shown to have a reassuring effect for the user and the caregiver and give continuity to the relationship with the HSET and the previously done rehabilitation work. They also reduce the distance between the rehabilitation center and the home, allowing the center's professionals to witness some dynamics and activities of domestic life and to widen the knowledge of the PwID/ASD and his family [39–42].

Even the interventions based on applied behavioral analysis principles seem to be well achievable through the TA and TR systems. Some researchers and clinicians have defined specific techniques and processes in this regard [47, 48].

The main limit is represented by the lack of all aspects of the therapeutic relationship related to physical interaction, so TA and TR have to be considered integrative and not substitutive of traditional (face-to-face) care. Other important limitations are represented by PwID/ASD-HSN's and their caregivers' ability to use the technology for TA and TR and the availability of the technology itself (lack of devices, bad connection quality, etc.). Further possible problems include privacy, informed consent, the definition of costs, and the cultural attitude of an entire community context [39, 49].

The presence of family members or other stimuli associated to the home environment can be a distracting factor and reduce the effectiveness of TR interventions compared to those in presence and appropriate setting. In respect to assessment and clinical interventions for psychopathological aspects, some studies have revealed difficulties in carrying out remotely [50] and favoring pharmacological rather than holistic approaches [51]. Other studies did not find significant differences or even suggested aspects of superiority compared to traditional in-person visits [39, 52–54]. Telepsychiatry has satisfied almost all users and professionals included in specific trials, has been associated with a marked reduction in access to emergency services and hospitalizations, and has resulted in costs for services provision significantly lower than those of traditional psychiatry [51, 54, 55].

Studies on TA and TR's efficacy for PwID/ASD-HSN are limited in quantity and quality, especially concerning adulthood. Available literature shows effectiveness on (a) improvement or maintenance of PwID/ASD's cognitive, adaptive, and occupational skills; (b) management of PwID/ASD's ordinary and extraordinary activities or critical episodes by family members or other caregivers; (c) empowerment of caregivers' skills in the relationship with the PwID/ASD, expanding the range of educational strategies, and promoting positive and collaborative interactions.

Compared with traditional face-to-face services, main advantages have been identified in higher availability and accessibility and shorter physical and psychological distance between the rehabilitation center and the person's home. The main limits are represented by the lack of all aspects of the therapeutic relationship related to physical interaction, possible poor ability to use technology, availability of the technology itself, privacy issue, and distracting factors associated to the home environment.

43.6 Prerequisites for Teleassistance and Telerehabilitation Services

In the short time allowed by the health emergency, the preliminary conditions' assessment takes the place of the phases of analysis and study preparatory to a detailed plan of the telemedicine services. This assessment must be straightforward and capable of being done quickly; it clearly has significance for the fact that the services have been and must be implemented within a short timeframe.

The preliminary conditions' assessment needs to be carried out with reference to the service provider, the user, and the territory where they are located. It must also consider the technological, health, and regulatory aspects, reporting territorial information and user needs. The decision about the possibility of providing TA and TR services during an epidemic emergency should be made on the conditions described in the following paragraphs and summarized in Table 43.1.

43.6.1 Conditions Referring to the Telecommunications Infrastructure

The technologies for delivering TA and TR services can be assigned to three main groups:

1. Telephone calls, text messages, interactive voice response (IVR) systems, and e-mail
2. Video calls (VC) using technologies such as Skype or WhatsApp
3. Specific applications for telemedicine services

Telephone calls, text messages, IVR systems, and e-mail can help organize, encourage, and remember TA and TR activities, but not for carrying out the activities themselves. They are also much more challenging to use for most individuals with ID/ASD-HSN [40, 56].

In more than 80% of the available studies, the assistance and treatments were provided

Table 43.1 Prerequisites for teleassistance and telerehabilitation services

Prerequisite	Teleassistance specification level	Telerehabilitation specification level
Connectivity to the domicile	5	5
Connectivity of the work station from where the socio-educational/healthcare personnel will take action	5	5
Accessibility of the service	5	5
Time schedule for providing the service	5	2
Security of personal data	5	5
Autonomy in the use of digital systems (by the recipient)	2	2

The authors of this document's specification levels have been indicated based on a Likert scale from 1 (minimum) to 5 (maximum)

through VC, with different technologies based on the type of assistance/treatment and suitability for the user. In the remaining studies, a website-VC combination or a store-and-forward technique was used [40]. The latter approach provides that information, even in the form of video, which is sent to an intermediate station where it is stored and transmitted later to the final destination or another intermediate station.

It is important to emphasize that the recipient's ability to connect represents the actual technological limit for any service employing information and communication technologies (ICT). In recent years, ICT and VC systems, which are the basis of telemedicine services, have developed significantly in Italy, but problems of availability and operation are still frequent among individuals with ID/ASD-HSN [40]. Poor connectivity can be caused by various infrastructural factors or the type of service made available by the telecommunications operator. For example, in-home users often experience a marked difference between the download transmission speed compared to the upload speed, with the consequent possibility that an in-home user will receive much more data within a specific unit of time than what he can send. A common consequence of this problem is a low quality of images coming from a patient in his home – a quality insufficient for use. Simultaneously, the observation of mimicry, gestures, and behaviors is critical for the technicians providing remote services [32].

In emergencies, it is sufficient to check to determine:

- If the person has a WiFi or cable connection, compatible with good data exchange
- If the person is at a correct distance from the router during connections
- If the person has digital devices that can be connected to the network and the type(s) of devices (e.g., smartphone, tablet, laptop, and/or desktop)
- Whether the person's computer or other digital devices have a video camera, microphone, and headphones
- If the person also has digital medical devices and their level of connectivity (WiFi or Bluetooth) on the network or with other devices that can be connected on the network

Services providers should advise users to download apps compatible with all smartphones and with simple log-in procedures.

Within the social-healthcare facilities, the operational work stations usually enjoy optimal connectivity. However, tests are recommended to check connection speed and verify the local network's real possibilities to support data traffic for the average volume of simultaneous download and upload requests. Such tests will be useful for: (i) ensuring the proper functioning at the time the services are rendered and (ii) documenting the quality of the work done during the emergency period. It is also recommended that the social-healthcare facilities equip themselves with various communications systems (e.g., SMS, e-mails with encrypted texts, video communications, and so forth). Video calls should be made with systems that are easy for the patient to use, directly from the app, preferably with web-based systems that do not require any installation of software on the device used.

43.6.2 Conditions Referring to the Treatment Services Provider and the Territorial Healthcare Organization

A TA service inclusive of treatments upon request should entail the same hours of operation generally provided before the implementation of the COVID-19-related precautionary measures. In doing so, the service provider can help people in isolation or at a distance for as long as possible and encourage a better quality of use by the service recipients. Experience and research indicate that the certainty of having a service available for extended periods usually prompts people to make more rational and less emotional use of the service [32].

It follows that, if the remote service is well structured from the start, with different activities sensibly offered over an extended time interval, it is possible to calibrate and orga-

nize the workload in a sustainable manner without the need for a huge number of operators. If it is not possible to offer the remote service over the usual time interval, or if it is deemed inappropriate to adopt the usual time interval, then it would become significant to explain to the user, from the very first contact, not only the rules of access and use by electronic means, but also a clear and exhaustive description of the alternative procedure to be followed at times when the service is not available.

For TR, it is possible to predefine with the user the times when the services will be rendered. TA and TR services require the acquisition of new skills by the professionals rendering the services. Therefore, it is useful to conduct training as soon as possible to address the use of telecommunications tools and the understanding of the peculiarities of the remote communications/interaction dynamics for two parties and groups, with particular reference to the treatment relationship. Teamwork should also be redefined and tested for specific needs.

The acquisition of trust and confidence in TR on the part of the professionals who must deliver the service is critical to the treatment's success, especially during the phase of building a treatment relationship.

43.6.3 Conditions Referring to Personal-Data Security and Digital Devices at the User's Home

The management of cybersecurity and data processing issues cannot be entrusted to improvisation, or even worse, abandoned, without excessively increasing the risk of intrusions, criminal activities, and improper use of patients' health data.

The socio-educational/healthcare workers who engage in telemedicine are required to observe the rules concerning the correct processing of patients' data and to avoid any behavior that can facilitate cyber-attacks. In this regard, it is noted that the behavior required by professionals to protect patients' personal data is simple and do not presuppose specific technical knowledge.

Concerning cybersecurity, the treatment facilities' remote-control systems must ensure the best possible IT security. Using impromptu solutions with social media platforms should be reserved for situations where it is impossible to pursue any other method and should be limited to the shortest possible time.

On the other hand, high standards of cybersecurity cannot be expected from the patient. In making remote services possible, particularly during this emergency, the telemedicine system must necessarily leverage the use of the devices at the patient's home, addressing the related risks as best as possible.

Adequate disclosures are needed to inform patients at home, and without needless red-tape, of the procedures concerning the risks for data security and cyberbullying in relation to the system used.

The commercially available systems dedicated to healthcare are practically all compliant with the EU General Data Protection Regulation ("GDPR") No. 679/2016 to which they had to adapt before the health emergency. Should particular situations arise as a result of the health emergency status, whereby the application of the data-processing rules would prejudice the provision of services, it would then be advisable to agree on a specific solution with the regulatory authority responsible for the personal data processing. Given the market presence of GDPR-compliant systems, there is no justification for not applying the rules, even in an emergency.

The rules relating to the certification of the medical devices that ID/ASD patients can use at home as part of TA and TR services are complex, have relevance for the sector's technicians, and are not detailed in this document. According to the primary reference standard (EC Directive 93/42), software programs that provide healthcare professionals with useful elements for making decisions for the patient are considered to be medical devices and, therefore, they must have adequate certification to be used. The certification mentioned above provides for different classes that correspond to increasingly stringent quality and safety requirements consistent with the

intended use. When delivering medical care procedures remotely, health professionals remain liable for the use of any software that is not certified for the use or is of unknown and non-verifiable origin (as could happen for software downloaded indiscreetly).

43.6.4 Conditions Referring to the In-Home Patient's Capability to Cooperate

In addition to sophisticated technological systems, telemedicine services also require the capacity of individuals to interact with these systems. For a telemedicine service to function correctly, the patient must be trained in its use. Over the years, the practical importance of networking has been better understood. Nowadays, many individuals with ID/ASD have better learned how to use network services, with various forms of tutoring. The service provider's staff needs to verify the extent to which the patient can interact with the necessary technologies. However, many ID/ASD patients are unable to use internet connection devices, obviously not only because of training deficiencies but because of the primary impairment of their learning and relationship skills. In such cases, the person needs a mediator for interacting with the service provider. Sometimes even family members or other habitual caregivers can have difficulty managing connections, both with regard to external parties and devices in the home.

Because of these problems, it is advisable that graphic interfaces are as intuitive and straightforward as possible, that the instructions are simple to follow and available both verbally and in writing, and that the staff in charge make the first contact with the recipient of the service, or with his caregiver, by means of a phone call. This first contact makes it possible to explain the type of service offered and indicate the most appropriate way to connect via video call for further and more detailed explanations and instructions. Numerous technological solutions available for video calls are easy to use, even from smartphones and tablets. During the dialogue in the first video call, information may also be requested to assess the extent to which the person can work with the available system, particularly with respect to sending the data to the referring doctor. Although many telephones, ADSL and fiber operators currently offer contracts with Internet traffic that is unlimited or high-volume and thus more abundant than what a person needs typically in everyday use, it is advisable to notify any patient who will receive TA and, more importantly, TR, that data traffic will necessarily be higher than usual with the activation of these services.

In general, the contents and the methods for delivering TA and TR must be defined from time to time based on the characteristics of the users of the service (e.g., the age and the degree of cognitive skills) and the service providers (e.g., the level of understanding of individuals with ID/ASD).

It is advisable to optimize the comfort levels by paying attention to the "sensory dimension" and the "clarity/comprehensibility" of the experience [57].

The expression "sensory dimension" refers to the sense-perceptive elements, especially visual and auditory sense-perceptive elements, which could distract or disturb an individual with ID/ASD. Elements to be considered include shaky images; continuous changes in the background; excess brightness; flashing screens; and excessively loud or confusing noises. The use of monitors needs to be carefully assessed, with respect to the type and the duration for the many individuals with ID/ASD-HSN who also have photosensitive epilepsy.

With the "clarity/comprehensibility" of the experience, reference is made to the extent of the structuring and the organization of services. Precise sequences, clear steps, and routines can foster a sense of predictability and control. The words, the contents of the communication, and the interactive methods (e.g., in terms of length of the statement, the tone used, the speed of speech, etc.) must be aligned with those that have proven to be the most effective over time for each ID/ASD patient.

The sensory dimension can be controlled through the choice of devices in relation to

the skills of people who use them. For example, the images captured by a smartphone or tablet in the hands of a person who tends to move about will be much shakier or blurred than those of the same devices fixed to a support or a PC. The stimuli that can pollute the sensory field of remote interaction can also be controlled by choosing the place and time for making the connection.

43.7 Type of Teleassistance and Telerehabilitation Services

When planning TA and TR services, it is crucial to do a specific-needs assessment using ad hoc tools. The assessment is necessary to become familiar with the ID/ASD patient's clinical-functional conditions and those of his family unit and the family's social support network. The assessment becomes the basis for arranging calibrated activities, according to an order of priority dictated by the urgency and seriousness of each situation.

TA and TR services can fulfil the following functions:

- Psycho-diagnostic assessments during the emergency
- Management of urgent situations
- Psycho-education
- Individual and/or family support
- Direct and indirect treatment [40, 58–61].

TR can be done individually or in small groups.

Concerning the psycho-educational function, it is essential in the emergency phase to provide the ID/ASD-HSN patient and his family members/caregivers with information about: the virus and the epidemic; hygiene and health standards; restrictions concerning routine activities; new ways of relating to others; physical and/or psychological symptoms that could arise due to distress (e.g., neuro-vegetative symptoms, insomnia, appetite changes, panic, depression, etc.); and services available (e.g., grocery delivery, online medical prescriptions, and assistance in hygienic/sanitary practices) through local services, residential structures, and day-care centers.

The psychological support function can be addressed to the individual with ID/ASD-HSN and those who interact with the individual, thereby ensuring the continuity of the relationship with the key-contact professional and possibly with the treatment facility frequented by the individual. Through this approach, it will be possible to moderate the feeling of loneliness, to ensure the availability of the professional's involvement in the event of an emergency that is difficult to manage, to explain information from the mass media that is not always comprehensible, and to avert distress through a series of therapeutic strategies.

The treatment function can be remotely implemented directly not only by the professional but also through the training and telemediated support of the caregivers (family members and in-home occupational therapists who work together on the ID/ASD-HSN patient's treatment program).

Informational, educational, and treatment techniques that are specifically used are:

- Social histories
- Various types of documents translated into alternative augmentative communications (AAC) forms
- Video-modeling or video-coaching (direct or indirect, individual, or group)
- Video tutorials (direct or indirect, individual or group)

There is a diversified array of telerehabilitation activities, along with numerous objectives. The activities can involve the individual with ID/ASD, as well as his caregivers and family members. Below is a list of the most frequent, which is based on literature and the practices of the authors of this document:

- Communicate and converse to favor the reception and the appreciation of messages of affection, acknowledgment, consideration, encouragement, and counsel and to obtain answers to questions
- Perform tasks together (collaborate) through VCs

- Carry out activities to enhance coping skills and resilience to distressors and to reduce the risk of stress and other psychiatric disorders
- Parent-training initiatives
- Interviews aimed at assessing the quality of life
- Provide support for the self-compilation of tools for assessing the quality of life
- Conduct assessments and applied behavioral analysis, with particular reference to instructions and help to carry out an assessment of the ID/ASD-HSN patient's preferences and to define a hierarchy of rewards (e.g., tangible, social, symbolic, and dynamic)
- Practical indications for identifying the psychological/behavioral suffering of the ID/ASD-HSN patient in relation to changes linked to the epidemic, evaluating symptoms, and problem behaviors through diagnostic tools, functional analysis cards, or supervision of video documents
- Help in compiling the aforementioned psycho-diagnostic and functional analysis tools
- Help in identifying specific reinforcing stimuli to motivate the ID/ASD-HSN patient to carry out hygiene practices, personal and domestic care activities, possible educational activities, etc.
- Supply indications for managing states of psychological and behavioral distress
- Maintain interpersonal ("friendly") contacts with both the ID/ASD-HSN patient and his caregivers (family members or in-home occupational therapists involved in this phase) to reduce the possible feelings of solitude and the sense of abandonment
- Help to build daily and weekly visual agendas to organize the ID/ASD-HSN patient's time and to maintain a daily routine
- Indicate how to increase the skills of the family unit, for example, through video monitoring of relationship dynamics
- Provide support for the continuity of the previously defined treatment plan, with a definition of objectives to be pursued within the home environment and related strategies
- Engage in a shared structuring of educational, occupational or recreational materials for time management
- Supply indications for the acquisition of new skills
- Provide indications for reducing the sense of ineffectiveness and powerlessness
- Provide indications for mobilizing prosocial and spiritual resources that may have been subdued to rediscover pleasant family experiences

43.8 Other Indications for Organizing and Delivering Teleassistance and Telerehabilitation Services

In addition to the guidelines set out in the previous paragraphs, the authors of this document have formulated further indications to be followed for organizing and managing the TA and TR services provided to individuals with ID/ASD-HSN. They are shown below:

- Follow the planning criteria and outcome measures focused on the ID/ASD patient; the identification of the objectives, tools, and methods for delivering the services must be based on a generic quality of life (whole-person) model
- Consider schedules, expectations, lifestyles, and needs about the ID/ASD patient's entire family
- Check in with the ID/ASD patient, his family members, and other significant caregivers/professionals, at regular and predefined intervals to determine if the activities carried out are meeting the needs and expectations and, if necessary, renegotiate the objectives and tools to achieve them, based on the assessment of the individual quality of life
- Have frequent interdisciplinary discussions about the objectives and progress of TR, with the inclusion of the case manager and the psychologists coordinating the program

- In the case of individuals with ASD, it is essential that the operator knows of a possible tendency to avoid eye contact and how to manage it. It might be useful to avoid being too close to the camera (so that the operator's face does not appear too large on the patient's monitor) or to avoid a continuous search for eye contact, for example, by asking the person to turn his attention to something close to him or by sharing a screen of the operator's device to show the ASD patient an image that can be watched together

43.9 Indications for Delivering Psychological Support

The service is designed to allow the person in isolation to have contact with a psychologist or psychotherapist. The video call is activated at the request of the patient and/or family members, regular care providers, or legal representatives, using a procedure that allows for the service as soon as possible, based on the resources available, directly at the patient's home.

By definition, these types of activities cannot be standardized. Therefore, the professional care provider is urged to seek references to optimize delivery, ensure safe conditions, and apply scientifically correct methods.

The service does not entail medical assistance but solely consists of a conversation with a psychologist, which aims to support people in isolation suffering from discomfort, limitations on relationships, and fears caused by the specific situation. The board-certified psychologist interacts via video call with the people who request the service, and can agree on further video call sessions with the person once the first contact is established and where he deems it necessary. The conversations will nonetheless be subject to the Order of Psychologists' Ethics Code and will not be recorded.

Before starting a session, the psychologist should identify the person with whom he is conversing, using the most appropriate procedure.

Given the nature of the individual conversation (with the use of no equipment) and in consideration of the healthcare emergency, the video-call system for psychological counseling only requires functional connectivity to allow the psychologist to comprehend both verbal and non-verbal language during the session.

If amenable to the service users, sessions can be conducted with groups, with activities, for example, based on sharing facts, experiences, or moods.

Professional care providers must seek references to optimize delivery, ensure safe conditions, and apply scientifically correct methods. If amenable to the service users, sessions can include group activities, such as sharing facts, experiences, or moods.

43.10 Future Prospects

This work has been conceived in view of the COVID-19 health emergency. Consequently, the authors did not focus specifically on the implications of using telemedicine services beyond epidemic conditions.

Although TA and TR services for individuals with ID/ASD have been shown to be feasible and effective, it is unlikely that they will be able to replace traditional practices, at least in the near future. However, they could represent a supplement to an integration of or a temporary alternative to conventional practices. For this purpose, numerous insights would be necessary, especially regarding the indications of the various activities, the methodology for evaluating the different services, and the various methods of implementation in the integration of public services and publicly subsidized private services. The adaptability of the TA and TR services at the local level and the assurance of their operational stability over time are two other factors requiring further analysis.

Part of the future research will have to focus on the use of artificial intelligence, machine learning, and interactive avatars [62].

Tip

Future research should provide insights on indications, efficacy assessment, contextual implementation, and operational stability over time of specific TA and TR activities. Further investigations should address the use of artificial intelligence, machine learning, and interactive avatars.

Key Points

- People with ID and/or ASD-HSN belong to one of the populations most vulnerable to the COVID-19 epidemic and related stressors due to several individual and environmental factors.
- The risks associated with the epidemic may also relate to the appearance or aggravation of problem behaviors.
- Persons with developmental disabilities represent potential users of telemedicine services as other patients who need continuous support.
- During the 2020 COVID-19 health emergency, remote interventions were indispensable to guarantee continuity of care for many PwID/ASD-HSN in quarantine or fiduciary isolation.
- It is essential to set up a remote service by analyzing the different possible situations of its use and the different types of needs of the persons to whom the service is addressed.
- Programming telemedicine services requires to identify diagnostic, therapeutic, educational, and care practices appropriately that can be carried out remotely, through technologies that are available and usable by persons with different levels of disability.
- TA and TR objectives must be aligned with the more general aim of every individualized therapeutic and rehabilitation plan, which is to promote and favor the generic quality of life of the PwID/ASD and his family.
- TA and TR are operated remotely by professionals with direct or indirect interaction with the PwID/ASD. Direct interaction is possible only with PwID/ASD who have fair cognitive and communication skills and are able to use technological devices. Indirect interaction takes place with the intermediation of a subject on the local side, which can be represented by a family member, a habitual caregiver, or a technician.
- Studies on TA and TR's efficacy for PwID/ASD-HSN are limited in quantity and quality, especially concerning adulthood, even if the available literature shows some areas of effectiveness (e.g., improvement or maintenance of PwID/ASD's cognitive, adaptive and occupational skills; management of PwID/ASD's ordinary and extraordinary activities or critical episodes by family members or other caregivers) and some advantages (e.g., higher availability and accessibility and shorter physical and psychological distance between the rehabilitation center and the person's home).
- Compared with traditional face-to-face services, main limits are represented by the lack of all aspects of the therapeutic relationship related to physical interaction, possible poor ability to use technology, availability of the technology itself, privacy issue, and distracting factors associated to the home environment.
- The decision about the possibility of providing TA and TR services during an epidemic emergency should be made on evaluating the service provider, the user, and the territory where they are located as well as technological, health, and regulatory aspects.
- When planning TA and TR services, it is crucial to do a specific-needs assessment using ad hoc tools.

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