

# Specific Learning Disorders, Motor Disorders, and Communication Disorders

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© Springer Nature Switzerland AG 2022 M. O. Bertelli et al. (eds.), *Textbook of Psychiatry for Intellectual Disability* and Autism Spectrum Disorder, https://doi.org/10.1007/978-3-319-95720-3\_18 18

#### 🔁 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 <a>Table 18.1</a>).

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 schoolaged children across different languages and cultures, and approximately 4% in adults. These percentages vary from country to

| <b>Table 18.1</b> SLDs in ICD-10 [5], ICD-11 [1], and DSM-5 [2] |   |   |   |  |  |
|---|---|---|---|--|--|
| SLD   | ICD-10  | ICD-11  | DSM-5   |  |  |
| Dyslexia  | F81.0<br>Specific reading<br>disorder                             | 6A03.0 Developmental<br>learning disorder with<br>impairment in reading                     | 315.00<br>Specific learning disorder with<br>impairment in reading              |  |  |
| Dysorthographia   | F81.1<br>Specific spelling<br>disorder                            | 6A03.1 Developmental<br>learning disorder with<br>impairment in written<br>expression       | 315.2<br>Specific learning disorder<br>with impairment in written<br>expression |  |  |
| Dysgraphia  | F81.8<br>Other developmental<br>disorders of<br>scholastic skills | 6A03.3 Developmental<br>learning disorder with other<br>specified impairment of<br>learning | 315.2<br>Specific learning disorder<br>with impairment in written<br>expression |  |  |
| Dyscalculia   | F81.2<br>Specific disorder of<br>arithmetical skills              | 6A03.2 Developmental<br>learning disorder with<br>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 indepth 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 metaanalysis 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

- 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 nonword 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 actiopathogenesis 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 practice, 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 threebrick 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 selfmaintenance) 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 selfinjurious 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  $\triangleright$  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 selfinjury 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  $\triangleright$  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, nonrhythmic 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 ASDspecific 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.
  - Monosyllabic whole word repetitions (e.g. '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)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.

- 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<br>responding to questions irrespective<br>of their content. For example, they<br>may answer 'yes' or alternatively 'no'<br>to all questions irrespective of their<br>contents. This may tell you they are<br>not understanding what you are<br>saying. | The tendency of the person with ID to<br>echo the last option can be minimized<br>by asking her/him to confirm both<br>options and choose one.  |
|---|---|---|
| Be aware that the person<br>with ID may have negative<br>experiences of the interview<br>process.   | Avoid closed questions and always use open questions.   | Avoid complex phrasing and those<br>involving abstract concepts such as<br>'extent', 'all the time', and 'low<br>spirits'.  |
| They may also be concerned<br>about the consequences of<br>the interview.   | Establish first the person's preferred<br>method of communication and<br>inform other professionals about<br>that.  | Ask the person with intellectual disabilities to clarify statements using examples.   |
| Wait for them to respond to<br>your question and assess<br>whether you feel they have<br>understood you. If<br>necessary, repeat the<br>question.   | Be flexible about the length of<br>interviews, such as using several<br>shorter interviews instead of one<br>long interview.  | Avoid double negatives. For example,<br>'You do not refuse medication, do<br>you?' may be usefully phrased as 'Do<br>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<br>get a time frame. For example, 'Are<br>you feeling sad since your birthday?'  |
| Interview in an unfamiliar<br>location may pose its own<br>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<br>the first question before moving to<br>the next.   | Watch out for non-verbal signs especially distress and discomfort.  |
| Remember that sensory<br>difficulties such as impaired<br>hearing or vision are likely<br>to affect the interview.  | Be aware of body language. For<br>example, during the discussion of a<br>recent bereavement, an autistic<br>person started to rock in his chair,<br>which is an early sign of distress.<br>You may need to stop the discussion<br>or change the topic.                            | You should be aware of autistic<br>masking where someone says<br>something, but it is not exactly<br>concise. The interviewer needs to use<br>insight and interpretation to establish<br>what the person actually means.  |
| Reduce background noise,<br>allow plenty of time and<br>ensure you understand each<br>individual's communicative<br>needs, and try to establish<br>eye contact to get their<br>attention. | You can use reverse wording. For<br>example, the question 'Do you have<br>trouble sleeping?' can be followed by<br>'Do you sleep well?' in the latter part<br>of the interview.   | For example, BJ, a 13-year-old boy<br>assessed for depression talked about<br>having his 'tummy cut'. His mother<br>clarified that this referred to his<br>preoccupation with his weight of late.<br>He had recently watched a programme<br>on television about surgical treatments<br>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-centredthinking-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-centredthinking-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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