Chapter 1
A Brief History of Autism

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Abstract In the 70 years that have elapsed since the first description of infantile autism considerable progress has been made. The official recognition of the condition in 1980 stimulated further research and around the same time the strong neurobiological and genetic aspects of autism began to emerge. Subsequent research has focused on issues of early diagnosis and intervention, understanding the neural mechanisms of autism, and approach to treatment. Unfortunately the research base varies considerably by age group with much research on younger individuals and rather little on adults—particularly older adults. Recent changes in diagnostic practice may complicate the interpretation of past, and future, research.

Keywords Autism · Autism spectrum · DSM-IV · DSM-5 · Diagnosis

1.1 A Brief History of Autism

Since its initial description in 1943 (Kanner 1943) interest in, and research on, autism has increased dramatically. In the 20 years following Kanner's classic description fewer than 50 peer reviewed papers on the topic appeared, but they increased gradually and then dramatically between 1973 and 1982 over 1000 papers appeared, and between 2003 and 2012 with well over 10,000 papers were published. This dramatic rise reflects several factors including early confusion about the diagnostic validity of autism (particularly apart from childhood schizophrenia), confusion about social class association and etiology and confusion about basic aspects of phenomenology. In this chapter I succinctly review the development of autism as a diagnostic concept, advances in research and treatment, and social and public policy issues. It is, of course, important to understand that all these issues are inter-connected, e.g., advances in treatment may have important policy implications which in term may impact research. As noted subsequently a range of materials on autism are now available (including on line reference works and lectures); the interested reader should review the original sources. An excellent review of the history of autism is provided by Adam Feinstein in his recent book (Feinstein 2010).

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1.2 Development of Autism as a Diagnostic Concept

Kanner's work was a remarkably accurate description of the condition we know today as autism (or autism spectrum disorder) and continues to impact approaches to diagnosis as well as to research and treatment. As the first child psychiatrist in the U.S. Kanner reported 11 cases where the child seemed to come into the world with an inability to relate to other people. Kanner regarded this as one of two essential features of the condition and emphasized the child's lack of social interest in his use of Bleuler's (1911) earlier term for autistic (self-centered) thinking in schizophrenia (the use of this term also, unfortunately immediately introduced considerable confusion about the relationship of infantile autism and schizophrenia). Kanner was careful to site the work of Arnold Gesell (Gesell and Ilg 1943) on normative social development to highlight the significance of the social isolation. In addition to 'autistic aloneness' Kanner emphasized a second core feature which he termed insistence on sameness (or resistance to change). This referred to the often marked difficulties children with autism had in dealing with change in the environment. Over time the concept has come to include stereotyped motor mannerisms (presumed to help the child maintain sameness) as well as the unusual interests and idiosyncratic responses observed. In retrospect it was, and remains, quite striking that in many ways the child's lack of interest in the social world was so very different from the over engagement in the nonsocial world (a topic that has become an increasing focus of research).

In addition to the confusion caused by Kanner's use of the word autism his otherwise excellent initial description also included a few observations that mislead early investigators. For example, he suggested that there was apparently normal intellectual potential (given performance on some—usually nonverbal—parts of IQ tests), that other medical conditions were not apparently present (given the lack of obvious physical stigmata and attractive appearance of the children), and, in his original report, there appeared to be a strong association with social class (most cases coming from families where a parent was highly successful). A considerable amount of time was spent in the 1950's and 1960's correcting these misimpressions.

It became apparent that the profile on psychological testing in autism was unusual for marked scatter with nonverbal skills being relatively preserved but verbal abilities are often highly deficient and overall IQ scores frequently in the intellectually deficient range (Goldstein et al. 2009). As children with autism were followed over time it became clear that they were at markedly increased risk for epilepsy (Volkmar and Nelson 1990) and that autism was associated with some medical disorders particularly Fragile X syndrome and tuberous sclerosis (Rutter et al. 1994). Finally the association with socio-economic status of families came to be viewed as an aspect of referral bias (Wing 1980) and today there is recognition that apparent ethnic/racial disparities are likely to reflect under-diagnosis in disadvantaged populations bias against use of the label (Mandell et al. 2009). Probably most importantly a series of studies in the U.K. (Kolvin 1971; Rutter 1972) made it clear that autism was a distinctive disorder and differed from childhood schizophrenia in

terms of its onset, clinical features, and associated family history. By the late 1970s it was clear that autism was a distinctive disorder and attempts were made to develop better approaches to diagnosis. Rutter's approach (Rutter 1978) emphasized delayed social and language development (not just due to any overall developmental delay) along with unusual interests/behavior with an onset before 30 months of age. The National Society for Autistic Children (NSAC) NSAC definition (NSAC 1978) emphasized other aspects of the condition (disturbances in rates/sequences of development, hyper/hyposensitivity).

As a result of these advances a decision was made to include autism (as "infantile autism") in the landmark 3rd edition of the Diagnostic and Statistical Manual (DSM-III) (APA 1980). In DSM-III a new term, Pervasive Developmental Disorder, was coined for the class of disorder to which autism was assigned. In addition to autism other concepts were also included, e.g., 'residual' infantile autism for individuals who once met criteria but no longer did so. While including autism as an officially recognized condition was a distinct advance many aspects of the approach chosen were problematic (see Volkmar and Klin 2005), e.g., the definition lacked a strong developmental orientation and the notion that people 'grew out' of infantile autism to 'residual' autism was problematic. As a result many changes were adopted in DSM-III-R including a stronger developmental orientation (Volkmar and Klin 2005). This approach had its own problems—particularly with over-diagnosis of autism among the more cognitively impaired (Volkmar et al. 1992).

DSM-IV appeared in 1994 and for autism a large international field trial was conducted in conjunction with the pending revision of the international classification of diseases (ICD-10) (Volkmar et al. 1994). The DSM-IV approach included social, communication, and restricted interests/behaviors criteria, was polythetic, and did a better job of diagnosis autism regardless of IQ. The DSM-IV/ICD-10 approach also recognized explicitly a number of other PDD subtypes (Asperger's, Rett's, Childhood Disintegrative Disorder, and subthreshold autism). The convergence of DSM-IV with ICD-10 likely significantly advanced research.

DSM-5 has appeared in 2013 (APA 2013). It has adopted a number of changes including a new overarching diagnostic concept—autism spectrum disorder, eliminating previous subtypes such as Asperger's, and using criteria derived from research diagnostic assessment instruments. DSM-5 has proven unusually controversial as it appears that some individuals will 'lose' their diagnosis complicating both service provision and research (Volkmar and Reichow 2013). The final impact of DSM-5 remains unclear although a potential lack of convergence with ICD-10 seems problematic.

1.3 Related Diagnostic Concepts

Both before, and after, Kanner's classic description of autism in 1943, other clinician-researchers described diagnostic constructs with some points of similarity to autism. Indeed it is likely that cases of autism may first have appeared in some of

the descriptions of so-called "feralwild" or "feral" children (thought to have been raised by animals) and indeed it has now been recognized that in addition severe deprivation can also be associated with 'autistic like' behaviors (Rutter et al. 1999; Wolff 2004). Some of these disorders have been officially recognized while the validity of others remains debated. In general their major point of similarity to Kanner's autism has to do with the presence of severe social disability.

In 1908 the Viennese special educator Theodor Heller described children who developed normally for some years (typically 3 to 4) but then had a marked regression with the development of what now would be viewed as an autistic-like clinical syndrome (see Heller 1908, 1930) and for an English translation see (Westphal et al. 2013). His original term for the condition "dementia infantilis" reflected the then common notion of regression being a form of dementia; subsequently the term "disintegrative psychosis" was used. We now know (Volkmar et al. 2005) that the condition differs from dementia in that the loss of skills is not progressive and that it is not a psychosis (the latter term arose in the context of broad views of childhood psychosis current in the first decades of the twentieth century (Volkmar and Tsatsanis 2002). Although it resembles autism once it is established the regression is highly unusual, much later than is typical in autism, and the outcome, sadly, is worse (see Volkmar et al. 2005). While included in DSM-IV the condition has been dropped from DSM-5 complicating potential research.

In 1966 the neurologist Andreas Rett (1966) observed a group of girls who exhibited some autistic features as well as unusual motor mannerisms (peculiar hand washing stereotypies), marked loss of skills, breathing difficulties, and orthopedic problems. Although he originally speculated that this condition was an autism variant in fact the 'autistic-like period' (of relative social withdrawal) is relatively brief and confined largely to the preschool years (Van Acker et al. 2005). This condition, recognized in DSM-IV but dropped from DSM-5 has now been shown to be due, in general, to the presence of a specific gene defect (Van Acker et al. 2005). Apart from its clinical significance the research significance of the condition has arisen from the potential for animal models (via knock-out approaches) for this disorder.

In 1944 Hans Asperger, then a Viennese medical student, described a group of boys with marked social vulnerabilities and motor problems who also exhibited unusual specialized interest (that interfered with functioning); well-developed language (though not good social-communication) skills (Asperger 1944). Unaware of Kanner's 1943 report, Asperger termed the condition autistic psychopathy (or, in probably a better translation, autistic personality disorder) using the word autism as had Kanner to convey marked social vulnerability. This condition received little attention until Lorna Wing's (1981) influential review and case series. Subsequently interest increased and data from the DSM-IV field trial was used, in part, to support inclusion of the condition as one of the pervasive developmental disorders.

Unfortunately the definition adopted was not ideal and problems were quickly noted (Miller and Ozonoff 1997). Subsequent work has noted the importance of including features like circumscribed interests and motor delays if robust patterns of difference are to be observed (Klin et al. 2005). The unfortunate decision in DSM-IV to adopt a precedence rule (i.e. that a diagnosis of autism should take

precedence) also created problems (Miller and Ozonoff 1997) and could have been remedied had clinicians been allowed to choose the more appropriate diagnosis. Although including Asperger's in DSM-IV markedly increased research in the area, the lack of consensus on definition has led to a proliferation of views on diagnosis (Sharma et al. 2012) leading to difficulties in interpretation of research. These difficulties also have contributed to the decision not to include Asperger's in DSM-5 see (Lord and Jones 2012). This seems unfortunate given the potential for significant research and clinical implications of the condition, e.g., relative to differences from autism in neuropsychological profile (with implications for treatment; Klin et al. 1995; Lincoln et al. 1998), associations with other psychiatric problems and differences in family psychiatric history (Klin et al. 2005) and in outcome (Szatmari et al. 2003).

Atypical Autism/PDD-NOS: Despite the change in name from Pervasive developmental disorder to Autism Spectrum Disorder and despite the marked increase in the 'broader autism spectrum' (reflected in the media and epidemiological studies) research on this condition (or group of conditions) has remained limited (Towbin 2005).

1.4 Advances in Research

The mistaken belief in psychological causation of autism (and the confusion with childhood schizophrenia) during the 1950's and 1960's significantly impeded research. This situation began to change in the 1970's. Work on the phenomenology of autism in England by Kolvin (1971) and Rutter (1972) strongly supported the validity of the category. In addition the strong brain basis of autism began to be appreciated, e.g., given the high rates of seizure disorder observed with onset both early in life and in adolescence (Volkmar and Nelson 1990) and the very strong genetic basis of the condition (revealed in the first twin study (Folstein and Rutter 1977). Parents, some of whom also became early researchers, were critical of this effort. For example, Bernard Rimland authored an influential early book (1964) arguing for a neurobiological approach to understanding (Rimland 1964) and studies using his first diagnostic instrument (Rimland 1971) also supported the validity of autism as different from schizophrenia. Early work on intervention began to show the importance of highly structured educational program (Bartak and Rutter 1973). Early research on intervention also revealed the importance of structured treatment programs (Bartak and Rutter 1973) and the role of behavioral, rather than psychodynamic, interventions (Ferster 1989; Lovass and Smith 1988). Early research on the neurobiology of autism centered on different systems including various brain systems (Ornitz and Ritvo 1976) and neurochemical correlates, particularly relative to the neurotransmitter Serotonin (Hanley et al. 1977). In addition a range of pharmacological interventions were evaluated by Campbell and colleagues at NYU (Campbell 1975). The first Journal, in the field the Journal of Autism and Developmental Disorders, was established in 1971 with Kanner as the initial editor. Over

time research interest has dramatically increased with over 2000 peer reviewed publications on an annual basis at present and with a number of other journals now focused on autism.

1.5 Treatments

The earliest efforts to employ a psychotherapeutic intervention model for autism (Bettelheim 1967) gave way to more structured educational intervention programs (Bartak and Rutter 1973). It became clear that structured behaviorally based intervention had a particular role in building skills and helping the child generalize and maintain skills learned. Over the last several decades a range of approaches have been developed (see National Research Council 2001 for an excellent summary). These include more behaviorally based programs using applied behavior analysis, to more developmentally modeled programs, e.g., the Denver model developed by Rogers (1996) as well as other programs that combine aspects of both the developmental and behavioral approaches (Koegel et al 2001). Still other programs are more eclectic and designed to address the specific learning styles seen in individuals with autism (Schopler et al. 1995). Many of these programs have now become strongly evidence based (Reichow et al. 2011).

To briefly summarize a complicated literature on the theoretical basis of treatment, it has become clear that, in essence, lacking the social 'frame' that most infants have, those with autism develop idiosyncratic and nonsocially focused ways of learning about the world. As a result they have very different styles of learning reflected, early in life, by difficulties in early emerging language skills (prosody, social language, rule taking) and then by delayed vocabulary and language acquisition with an unusual 'gestalt' style of learning (reflected in the tendency to echo heard language and in the use of idiosyncratic language (Schoen et al. 2011; Volkmar and Wiesner 2009). Similarly unusual interests and preoccupations (presumably also arising because of lack of primary social orientation) can pose obstacles for learning and are reflected in other differences in learning style (Volkmar and Wiesner 2009). There has been an increased awareness of the importance both of early diagnosis for ultimate outcome (National Research Council 2001) and of peer exposure and the importance of a focus on social skills (Prendeville et al. 2006) and in mainstreaming in the acquisition of social skills as well as supporting teachers and parents as they, in turn, support the child's learning (Venn et al. 1993). On balance, with earlier intervention and more sustained and targeted intervention outcome apparently has gradually improved with more individuals with autism now able to be independent and self-sufficient (Volkmar and Wiesner 2009) although sadly research on older individuals and appropriate treatment programs to assist them have been minimal. For some individuals, even with good programs from early in life, outcome remains poor (Howlin 2005). It should also be noted that although more interventions can now be regarded as evidence based there are many complementary and alternative treatments in frequent use (Volkmar and Wiesner 2009). Issues in the use of such

treatments become particularly complex when there is some risk to the child (either directly in terms of safety or in terms of removing the child from treatments shown to be effective).

1.6 Research Directions

As noted previously early research efforts were impeded by a lack of consensus on diagnosis. As this began to change in the late 1970's more useable research began to appear. Parents and advocacy groups also formed to support research. As with other areas of medicine and behaviour much of the significant research conducted has been in the context of college/university settings with their strong commitment both to research and training. As noted above there has been a major increase in the research enterprise within the U.S. and abroad based both on an increased governmental commitment, strong support from advocacy groups, and, particularly in the U.S., the engagement of philanthropy (Autism Speaks, the Autism Science Foundation, and the Simons foundation have been notable contributors in this regard). Important obstacles and gaps in research remain, e.g., the more or less total absence of work on aging in autism and the minimal amounts of work on intervention for adolescents and adults. Treatment research has been particularly challenging to fund. On the other hand advances in the basic research on genetic factors (Abrahams and Geschwind 2008) and in our understanding of the social brain (McPartland and Pelphrey 2012) have been very significant. This basic research is beginning to be joined with treatment research, e.g., in studies showing brain changes in response to treatments (Voos et al. 2012). Research on pharmacological interventions has also significantly advanced including new work focused on the core social disability (Scahill and Martin In Press). The multisite study of risperidone has served as an excellent example of collaborative work that established the efficacy of this agent (McCracken et al. 2002).

1.7 Social Policy Issues

The predominant model of intervention in the 1950's was psychodynamically based. Public schools often declined to provide educational programs for children with autism despite the growing body of work on the impact of special education and behavioral treatments. This situation markedly changed with advent of Public Law 94–142 which mandated, in the U.S., that schools receiving federal funding had to provide services to students with disabilities—including autism (see National Research Council 2001). Other important social policy advances included the organization in the U.S. and U.K. of national parent advocacy groups. These groups advocated for research as well as for treatment programs. This work culminated in the 1990's with the establishment of a number of federal programs supporting network

of investigators. These included the Centers of Excellence for Programs in Autism (CPEA) led through the National Institute of Child Health and Human Development by then Director Dwayne Alexander and program officer Marie Bristol-Power and were then succeeded by support from multiple NIH institutes. Subsequent efforts have included the Studies to Advance Autism Research and Treatment (STAART) and Autism Center of Excellence Programs. The impact of these initiatives both on research and well trained investigators has been enormous.

Despite the clear role of legal entitlements to education, policy issues regarding school intervention program and appropriate practice continue to be addressed. In the U.S. the entitlement to education is now codified as IDEA (Individuals with Disabilities Education Act) which mandates free appropriate public education (FAPE) to children with a range of disabilities (Mandlawitz 2005). Follow-up federal legislation and judicial decisions have impacted the ways the IDEA is implemented. In addition another statute, the Americans with Disabilities Act of 1990, can apply to children as well as to adolescents and adults (particularly for young adult in college where it mandates against discrimination). On the other hand a source of confusion is that this law mandates supports when appropriate but does not establish program participation as an absolute right, e.g., in college (Wolff et al. 2009). It is important to note that decisions about intervention also have important economic implications with major expenditures in the U.S. and U.K. which might be minimized by improved diagnosis and treatment and better functional outcome (Knapp et al. 2009).

Interestingly some areas of work relevant to policy have received comparatively much less attention in the literature. Cultural aspects of autism remain an area where little work has been done. The issues here seem less to have to do with the presentation of autism (which likely is rather similar around the world) but with philosophies about intervention (Brown and Rogers 2003); study of ethnic and cultural issues is a priority as is the study of potential differences in presentation based on gender and socio-economic status. There is some suggestion that, in the U.S., children living in poverty, may be more likely under-diagnosed. (Mandell et al. 2009)

1.8 Training and Dissemination of Knowledge

As noted above the strong connection of research (and to some extent treatment) efforts to University settings has had a fortunate impact in developed countries of increasing the number of individuals trained in methods of research and knowledge about autism. Over time this work has been disseminated in various ways including new approaches in recent years including the internet (http://www.youtube.com/course?list=EC27FAF837577D180A) with some publications now including hypertext links to more rapidly update information and expand the range of knowledge rapidly available (Volkmar 2012).

1.9 Summary and Challenges for the Future

Although much progress has been made, many areas of challenge remain. From the research side the potential for connecting genetic and brain mechanisms is now on the horizon (Kaiser et al. 2010) and has the potential to significantly advance treatment research. The advent of specific genetic mechanisms will also allow for development of much more sophisticated animal models. New approaches to early diagnosis may facilitate more rapid screening—relevant particularly to at risk siblings. Similarly the ability to observe brain and other changes in response to treatment programs may help clarify the important issue of understanding which children do, and do not respond to various treatments.

Challenges remain. Particularly in the current economic climate, research funds are limited. Much of the research work has focused on young children with adolescent and adult research receiving short shrift. Given the potential for helping many young people become productive and self-sufficient adults this is especially unfortunate. Even for school age children important questions remain about best approaches to adopting treatments relevant to the specific child. Considerable variability exists in the U.S. between (and sometimes within) states despite overall Federal mandates.

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