From Kanner to DSM-5: Autism as an Evolving Diagnostic Concept

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Abstract

Seven decades have elapsed since Leo Kanner described the syndrome he termed early infantile autism. Over this time, and particularly over the past two decades, noteworthy changes have occurred in how the condition is conceptualized. Here we provide an overview of these changes, beginning with a brief discussion of the significance of classification in general before discussing Kanner’s original paper and subsequent changes. We touch on relevant issues, such as comorbidity, dimensional aspects of diagnosis and screening, and the complex issue of diagnosis relative to eligibility for services. Approaches to diagnosis have tended to swing from emphasizing overarching groups (lumping) to focusing on potentially distinct subgroups (splitting). Autism raises particular problems given the broad range of syndrome expression over age and developmental level. The most recent revision of the American Psychiatric Association’s diagnostic taxonomy marks a significant departure from its predecessor and has been the focus of much debate. It remains unclear which of the currently existing categorical approaches will ultimately be most widely applied. We hope to convey a sense of areas in which consensus has been achieved and areas of continued controversy.

Keywords

autism, autism spectrum, DSM-5, diagnosis, classification
ISSUES IN DIAGNOSIS AND CLASSIFICATION

Systems of classification are intended for a variety of purposes, including enhanced communication for clinical work, research, and policy planning. When diagnostic systems are used consistently, they can enhance research by enabling investigators in different locations to effectively communicate findings about similar groups. From the point of view of treatment, particularly in the United States, the use of a specific diagnostic label may result in special entitlements to services, such as special education. Official diagnostic schemes have a particular set of challenges related to consistent application across gender, culture, age, developmental or functional level, and other factors. Other considerations arise in terms of whether the official system is meant to be totally comprehensive or whether it is to be used in research only, in clinical work only, or in both clinical and research work. Challenges arise if the system is intended to be comprehensive in coverage since not all cases readily fit prototypic categories. Other challenges arise because of the impact of development on the expression of clinical disorders and the tendency of disorders to occur jointly (the problem of comorbidity; Rutter 2011, Rutter & Tuma 1988, Volkmar et al. 1991). Before considering some of the issues that arise for official classification schemes, it is important to emphasize that a diagnostic label is in fact only one facet of a comprehensive diagnostic process (Cohen 1976).

Official diagnostic systems, such as the American Psychiatric Association’s Diagnostic and Statistical Manual of Mental Disorders, 4th edition (DSM-IV) (Am. Psychiatr. Assoc. 1994) or DSM-5 (Am. Psychiatr. Assoc. 2013) and the World Health Organization’s International Classification of Diseases and Related Health Problems (World Health Organ. 1990), must strike a careful balance between being too broad or too narrow. Consistency in use and reliability are both important, and there is a recurrent tension between “lumping” and “splitting” because diagnostic systems lose value by adhering to categories that are too gross or too nuanced. Other challenges relate to the balance of clinical versus research needs. One issue, particularly for developmental and psychiatric disorders, is the degree to which impairment (including interference in learning/development for children) is an essential feature of the condition. An argument could readily be made [as indeed Asperger (1944, 1979) himself made] about the boundary between personality style and disorder (Rutter 2011). Tourette’s syndrome is an example of the issue of degree of impairment in that one may see a child who is effectively immobilized by severe tics and who has a father whose tics are noticeable to the trained observer but are so mild and noninterfering that he has never been
Some individuals with autism and related disorders may experience significant social difficulties but may see their problems more as a lifestyle/personality type and may wish to avoid the potential stigma of a diagnosis. Others may wish to share an awareness of their issues with individuals who experience similar challenges (Rutter 2011).

Various approaches to the classification of disorders exist. In medicine, public health, and, to some extent, social policy, categorical models (the presence or absence of a condition) tend to dominate, although even here some conditions (e.g., hypertension, intellectual disability) exist on a continuum or dimension, and the determination of a specific categorical cut point is somewhat arbitrary (although increasingly required to be informed by empirical evidence). Dimensional approaches to classification provide, for some purposes, more useful information than categorical ones. Dimensional approaches can be used diagnostically and are exemplified in the use of screening approaches for autism and in increasingly sophisticated diagnostic measures that assess levels of function/dysfunction (see Lord 2013, Stone & Ibanez 2013). Issues of instrument development can be complicated when one moves from a more normatively based, standardized approach to one where atypical behaviors are rated. Different approaches to this problem have been adopted, e.g., in one diagnostic instrument, the Childhood Autism Rating Scale, second edition, behaviors in 15 categories are rated on a four-point scale ranging from normal to severely autistic. For this approach, the complexity has less to do with defining normal and more to do with defining levels of severity (Schopler et al. 2010). Normatively based approaches have also been used successfully. In one study, deviations from expected social skills based on a normative sample using the Vineland Adaptive Behaviors Scales (Sparrow et al. 1984) were predictive of autism (Volkmar et al. 1987).

One of the major lessons learned so far in the development of classification schemes is that theory-based approaches have potential pitfalls. Theory-based approaches guided the two versions of the DSM prior to 1980 and, as a result, those who did not adhere to the theoretical framework on which the criteria were based had difficulties applying the criteria. This situation changed dramatically with the advent of the research diagnostic criteria approach—one that emphasized observable features rather than theoretical concepts, i.e., “sticking to the facts” in diagnosis (for a discussion, see Spitzer et al. 1978). Other issues arise in terms of the need for balancing the report of history with direct observations/current findings as the basis for categorization and whether to encompass the level of severity or development. Such issues are particularly complex for autism given its early onset, and they become even more complex when clinicians are faced with adults who can provide little information about their early growth and development. Approaches to dealing with the complex issues of developmental change began in the late 1960s with use of a multiaxial approach to diagnosis (Rutter et al. 1969). This approach, with modifications, has continued to be used in the various DSMs since 1980, and DSM-5 attempts a rather complex set of changes to eliminate former distinctions between psychiatric, developmental, and medical disorders, with a provision for noting important factors relating to context/psychosocial situation (formerly Axis IV) and severity of disability (previously Axis V). The multiaxial approach was particularly useful for documenting important conditions that needed to be addressed, for example, co-occurring intellectual disability or seizure disorder, and this information will now be conveyed through a series of specifiers appended to DSM-5 diagnoses.

Developmental factors impact almost all conditions to some degree. For disorders with a very early onset such as autism, these factors can be of tremendous importance. A comprehension of developmental issues often deepens our appreciation of clinical phenomena, for example, in understanding the functions of echolalia in autism (Paul 2005). Developmental change and

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1In this review, we use the terms autism and autism spectrum disorder interchangeably.
complexity also present significant challenges, for example, around the problem of developmental interference and the effects of development on clinical manifestation over time. These factors can predispose individuals with a disorder toward additional disorders, or comorbidities (Rutter et al. 1976). Awareness of the high rates of some disorders in individuals with intellectual disability led to the development of the concept of diagnostic overshadowing, i.e., the presence of one disorder masks others (Reiss & Szyszko 1983). The problems raised by comorbidity are substantial and are approached differently in various official categorization schemes (Rutter 1997). It has become increasingly apparent that individuals with autism and related disorders have an increased risk of developing other problems (Brereton et al. 2006). More cognitively able individuals with autism, for example, have higher-than-expected rates of mood and anxiety problems (Lugnegard et al. 2011, Stewart et al. 2006, White et al. 2012). These issues may have important implications for treatment, although, sadly, basic data are lacking in many areas [e.g., work is absent on rates of suicidal thought/behavior in adolescents and adults with autism spectrum disorder (ASD)].

The role of etiology in treatment also presents some challenges for diagnostic schemes. Even for disorders with known etiology, such as those caused by a single gene disorder or other genetic condition, phenotypic heterogeneity is observed. Different approaches have been taken into account for these situations in diagnostic taxonomies. Rett’s disorder was included in the class of pervasive developmental disorders in DSM-IV (Am. Psychiatr. Assoc. 1994) even though it was not thought to be a form of autism (although there was some potential for confusion of Rett’s disorder with autism early in childhood); the distinct course and clinical features suggested a specific etiology, but it was considered important to recognize and distinguish disorders like autism and Rett’s with (transiently) similar presentations (Rutter 1994). Subsequent to Rett’s inclusion, a specific genetic etiology was found. The removal of the condition from DSM-5 reflects an intent to avoid distinctions between medical and psychiatric disorders (e.g., in the abandonment of the multiaxial system) and an awareness that, for this single gene disorder, autistic features are usually confined to a specific point in development (the preschool period). The presence of Rett’s disorder can be noted as an etiologic factor (along with fragile X syndrome) in the specifiers, but this shift in conceptualization may lead to more challenges in the future, as specific genetic contributions to autism are elucidated.

Classification systems, like all human constructions, can be misused (see Gould 1996, Hobbs 1975). One of the most egregious mistakes is to equate people with their disease label, for example, “autistics” or “diabetics.” The use of categorical terms fails to capture the tremendous range in expression of the syndrome. From a scientific viewpoint, there is great danger when a label becomes an explanation. Finally, a major potential outcome of misuse of classifications is stigmatization and a resulting minimization of opportunities for maximal development and full community integration for individuals with disabilities.

The value of diagnostic systems for autism and related disorders is reflected, to some degree, in a review of the peer-reviewed scientific literature. Prior to DSM-III, autism was not a recognized diagnosis, and as a result, the studies that did appear on autism were difficult to interpret because no consistent diagnostic approach was used across studies. Thus, much of the early work on autism is difficult, if not impossible, to interpret. After DSM-III (Am. Psychiatr. Assoc. 1980) included autism, papers on the topic appeared with greater frequency. By 1993, the year before DSM-IV (Am. Psychiatr. Assoc. 1994) was published, there were approximately 250 peer-reviewed papers; 1,500 to 2,000 papers have appeared annually during the past several years. From the clinical perspective, shared and useful diagnostic systems can facilitate treatment and policy planning. Changes in diagnostic systems present important opportunities and challenges. Opportunities arise given the potential for making data-based changes that will improve the systems; challenges arise because major changes have the potential to disrupt ongoing studies, complicate the interpretation
of older work, and, potentially, complicate treatment (e.g., if mandates for service change or if dramatic shifts in epidemiology require reconfiguration of services or planning).

FROM KANNER'S REPORT TO THE DSM-III

Kanner's Report

It is likely that reports of so-called feral children represent the first reports of children with autism (Candland 1995, Wolff 2004). Although intellectual disability had been recognized since antiquity (Harris 2006), interest in it and child development in general began to increase with the Enlightenment and the debate of the role of nature versus nurture in child development (Hunt 1961). By the mid 1800s, interest in psychiatric problems in children grew, and continuities with adult forms of mental illness were suggested (Maudsley 1867), although little provision was made for recognizing the relevance of developmental factors in syndrome expression. The late 1800s, in particular, was a time of great activity in psychiatry taxonomy, with the recognition of dementia praecox (now termed schizophrenia; Bleuler 1911) and manic-depressive illness (bipolar disorder; Kraepelin 1921). These concepts were rapidly extended to children (e.g., dementia praecossima; de Sanctis 1906). The early tendency to equate severe psychiatric disturbance in childhood with adult schizophrenia later became a problem for recognizing the validity of autism as a distinct diagnostic category.

Leo Kanner, an émigré from Austria, was recruited to Johns Hopkins Hospital to bridge the fields of psychiatry and pediatrics. He wrote the first textbook on child psychiatry and in 1943 published the classic paper “Autistic Disturbances of Affective Contact.” This paper presented 11 children who Kanner believed entered the world without the predisposition to be social. He framed his description developmentally, noting Gesell’s (1934) description of early infant social development; by a few weeks after birth babies typically were smiling responsively and were socially engaged. Kanner provided careful, factually based descriptions of the cases and then attempted a synthesis in his overall description of the condition. He believed that in addition to autism (being cut off from the world of people), the subjects also had trouble in dealing with change in the nonsocial world. Kanner termed this “resistance to change” or “insistence on sameness.” In his view, some of the unusual autistic self-stimulatory behaviors (hand flapping and body rocking, for example) were attempts on the child’s part to “maintain sameness.” Although he did not recognize communication problems as an essential feature of autism, he did observe that three of the subjects did not speak at all and that the others had language problems. These difficulties included echolalia, idiosyncratic and extremely literal language, and difficulties with pronoun use. His observations were prescient in many ways, as recent research efforts in autism have focused on the apparent paradox of overengagement with the nonsocial world and lack of interest in the social world (Klin 2005a,b; McPartland et al. 2012). Kanner indicated that he believed the condition to be congenital (but did note a history of apparent regression in one of the cases).

Some aspects of Kanner’s description came to be sources of early confusion in the field. His use of the word autism did convey the aloneness of the cases. It also confused autism with schizophrenia, in which “autistic thinking” had been described some years before. His impression of normal intellectual potential in autism was based on an awareness that, on some parts of tests of intelligence (typically nonverbal, memory-based tasks), children with autism did well, but on parts involving language and symbolic thinking, they did poorly. Kanner’s impression was that if children performed as well on the latter areas as the former, they would function in the normal range. It took many years before psychologists appreciated that for children with autism, marked scatter in skill development was the rule, poor test performance reflected lack of ability rather than “negativism,”
and overall IQ scores (even while representing the average of very discrepant abilities) often fell in the intellectually deficient range (Goldstein et al. 2009). In a minority of cases, an unusual islet of high ability might be seen (Hermelin 2001). Kanner noted the attractive appearance of his subjects and emphasized that they were had no obvious physical indicators or dysmorphology; only as the children with autism were followed did it become clear that they were at increased risk for developing seizure disorder and that autism was associated with a small number of highly genetic conditions (Volkmar et al. 2013c). Finally, in Kanner’s initial case series, the parents of children with autism tended to be highly successful and accomplished people; there was little awareness of the potential for selection bias (i.e., that only well-informed and successful parents would be able to find the one child psychiatrist in the country). As a result, the impression in the 1950s and 1960s was that autism was a disorder of well-educated and high-status families, and this in turn led to an impression that experience or deviant parenting might play a role in pathogenesis (Bettleheim 1967). Subsequent work clarified that children with autism are observed in all socioeconomic status groups (Wing 1980) and that autism is a brain-based disorder with a strong genetic component (Rutter & Thupar 2013).

From Kanner to DSM-III

Interest in autism gradually increased following Kanner’s (1943) original report. Unfortunately, one major line of work focused on the potential for parental psychopathology to cause autism (Bettleheim 1967). This work was inspired, in part, by the then strong interest in psychoanalysis and noted effects of experience on children’s development, as well as—to some extent—Kanner’s observation of high levels of parental achievement in his original sample. In the first decade or two after Kanner’s description there was also a strong movement to view autism as the earliest manifestation of childhood psychosis or schizophrenia. However, several lines of evidence challenged this view.

The early pioneering work of Bernard Rimland (1964) focused on new approaches to objective diagnosis and provided a hypothetical neurobiological mechanism for autism. Studies of clinical phenomenology by Kolvin (1972) and Rutter (1972) made clear that autism was different from schizophrenia (even from childhood-onset schizophrenia) in terms of onset, clinical features, and family history. As children with autism were followed over time, there emerged strong evidence of a distinct course in autism and an elevated risk for developing recurrent seizures (epilepsy), suggesting a neural etiology (Volkmar & Nelson 1990). By the late 1970s, the first studies of twins suggested a strong genetic basis for the condition (Folstein & Rutter 1977). Studies of effective treatments (Bartak & Rutter 1973) also began to suggest the importance of structured, behaviorally focused treatments (rather than the unstructured psychotherapy advocated by prior prevailing theoretical views), and a growing body of work on behavioral interventions supported the use of methods from behavioral psychology and special education in guiding intervention programs (Ferster 1972).

By the late 1970s a consensus had begun to develop on the importance of studying autism independently of schizophrenia. A series of texts attempted to update Kanner’s definition with more formal diagnostic guidelines. Two notable examples were provided by Rutter (1978) and Ritvo & Freeman (1978). Rutter’s diagnostic approach markedly influenced DSM-III two years later. In Rutter’s diagnostic model, difficulties must have been evident at early onset and include marked social problems (not just due to intellectual delay), language difficulties or absence of speech (also not just due to intellectual delay), and unusual behaviors and rigidities subsumed by Kanner’s notion of resistance to change/insistence on sameness. The autism definition by the National Society for Autistic Children emphasized other aspects of the condition, including
unusual rates or sequences of development and hyper/hyposensitivity to the environment. Within child psychiatry, broader developments in classification were also important, for example, the use of multiaxial approaches to diagnosis with greater specification of levels of functioning and the advent of the Research Diagnostic Criteria approach at Washington University in St. Louis (Spitzer et al. 1978).

These convergent lines of research led to a decision to include “infantile autism” as an officially recognized diagnosis in DSM-III in 1980. To emphasize its uniqueness, a new term for the class of disorder to which autism was assigned was coined: pervasive developmental disorder. As suggested by the name infantile autism, autism in DSM-III was indeed focused on early development. Given developmental gains over time, a category was included for “residual infantile autism” to classify cases that had met criteria for infantile autism earlier in development. Another category was included for children who developed a condition similar to autism after some period of normal development.

The recognition of autism was clearly a major advance, as were other more general changes (e.g., the use of multiple axes for coding additional information). However, other aspects were problematic. For example, the notion of “residual” autism failed to acknowledge the persistence of difficulties over time. The overall approach was monothetic (every criterion had to be met) and therefore lacked flexibility. There was inadequate attention to developmental change. A number of changes were therefore contemplated for the revision of DSM-III.

**DSM-III TO DSM-IV**

A major revision of the definition of autism was undertaken in DSM-III-R (Am. Psychiatr. Assoc. 1987). Conceptually, these changes were conveyed in the name change of the disorder—from infantile autism to autistic disorder. This reflected an emphasis on a developmental approach not limited to the youngest children with autism (Siegel et al. 1989, Waterhouse et al. 1993).

Sixteen detailed criteria were provided, grouped into the three major domains of dysfunction observed in autism: qualitative impairment in reciprocal social interaction and in communication, as well as restricted interests. For a diagnosis of autism, eight criteria had to be endorsed, with at least two from the social category and at least one each from the other two categories. This polythetic approach gave greater diagnostic flexibility. The final version of DSM-III-R was based on the results of a field trial (Spitzer & Siegel 1990). Unfortunately, the field trial was problematic in some respects. For example, cases could be included on the basis of a review of materials rather than direct examination, and inappropriate comparison subjects (individuals with conduct disorder) were included in the comparison group.

Positive and negative aspects of DSM-III-R were quickly noted. Positive aspects included the greater attention to developmental change and developmental level (Volkmar et al. 1992b). Unfortunately the improved system significantly increased false positive rates (Factor et al. 1989, Hertzig et al. 1990, Volkmar et al. 1992a). Including examples of specific behaviors within criteria tended to reify the example (rather than the underlying concept). In contrast to DSM-III, the DSM-III-R approach emphasized current examination without the necessity of including early history (i.e., early onset). It was problematic as well that there seemed to be a growing divergence in the DSM-III-R with the approach taken in the draft of the tenth edition of the World Health Organization’s International Statistical Classification of Diseases and Related Health Problems (ICD-10; World Health Organ. 1990) (Volkmar 1992).

The ICD-10 adopted a rather different approach to autism and, more conceptually, to all disorders. Although the ICD and the DSM are related in important ways to issues of diagnostic coding, these systems also developed in distinct ways. For example, the DSM-III and subsequent
editions have used a single book for both research and clinical purposes, whereas the ICD-10 adopted a two-volume approach—one providing a set of criteria for research purposes and another focusing on clinical description. Additional differences exist, such as approaches to comorbidity. For research, the ICD-10 offered the advantage of much more detailed and copious diagnostic criteria. The interpretation of studies across countries presented one potential difficulty resulting from the major differences in the ICD and the DSM-III-R approaches to diagnosis (Volkmar et al. 1992a). These issues were given serious consideration in the major revision of diagnostic approach undertaken in the DSM-IV (Am. Psychiatr. Assoc. 1994).

The revision process for the DSM-IV included reviews of existing research by a series of work groups to identify, to the extent possible, areas where consensus had been reached as well as areas of controversy. Changes from the DSM-III-R were made only when well justified by empirical evidence and in light of the pending changes with the ICD-10. The process included a series of commissioned literature reviews published in peer-reviewed journals. One major issue was the inclusion of other disorders along with autism in the overarching category. These conditions included Asperger’s disorder (Szatmari 1991), Rett’s disorder (Gilberg 1994, Rutter 1994) and an apparently rare condition variously termed disintegrative psychosis, Heller’s syndrome, or childhood disintegrative disorder (Volkmar 1992). In general, the consensus was that, to the extent possible, comparability of the DSM-IV and the ICD-10 was desired (Rutter & Schopler 1992). A series of papers was commissioned relative to issues of differences in diagnostic approaches to autism. These suggested that the DSM-III-R was more developmentally oriented than the ICD-10 but was overly broad (Volkmar et al. 1992a,b). A major issue was to balance the sensitivity and specificity of the diagnostic approach while maintaining a flexible and developmentally oriented definition of the condition.

A field trial (Vokmar et al. 1994) was undertaken to address these issues. This field trial was international in scope (and conducted in conjunction with the ICD-10 revision); it included more than 20 sites and over 100 raters who provided information on nearly 1,000 cases. In contrast to the DSM-III-R, cases were included only when the clinician considered autism a potential factor in differential diagnosis. Cases were evaluated by both more- and less-experienced raters generally on the basis of current examination, although for some of the rare conditions potentially included in the ICD-10, cases were specifically solicited on the basis of available records. In most instances, multiple sources of information (history, past examinations, current assessment) were available, and raters judged the quality of information available to them to be good to excellent in about 75% of the cases. The field trial used a standard coding system to obtain information on old (DSM-III, DSM-III-R) and possible new (DSM-IV, ICD-10) criteria for autism and related disorders. Raters also provided other information on the case and on themselves (e.g., level of their own previous experience with autism, the quality of information on the case).

The results of the field trial suggested that the DSM-III-R differed significantly from the DSM-III (in the “lifetime” diagnostic sense), the ICD-10, and clinician judgment. DSM-III-R criteria resulted in a high rate of false positive cases, particularly if significant intellectual disability was present (in such cases, the false positive rate was about 60%). The detailed draft ICD-10 research definition worked well but was, as expected, more detailed than was felt desirable for DSM-IV (which was intended for both clinical use and research). Interrater reliability and internal consistency were calculated, along with other properties of the diagnostic classifications, using signal detection analysis and factor analysis.

Agreement among experienced raters on clinical diagnosis was excellent, with the relatively rare disagreements focused on fine-grained distinctions between autism and other disorders in the pervasive developmental disorder (PDD) class. Less-experienced raters had the same pattern but with lower overall levels of reliability. Factor analysis produced several potential solutions,
including the traditional triad of domains of difficulty (social, communication, and restricted interests). A two-factor structure (social communication and restricted interests) and a five-factor structure (in which restricted interests criteria were sorted into three groups) were also identified. Issues of diagnostic stability and sensitivity and specificity by age and IQ were also assessed. The final versions of the DSM-IV and ICD-10 proved reasonably robust, with a good balance of clinical and research utility; differences between the two systems were relatively minor (e.g., the ICD-10 provided various codes for ways in which cases could be subthreshold). Another aspect of the joint field trial was the inclusion of other disorders within the PDD class.

Nonautistic PDDs in DSM-IV

As part of the field trial, data were collected on a range of conditions related to autism; these conditions were potential candidates for inclusion in ICD-10. Asperger’s disorder had initially been described the year after Kanner’s (1943) report on autism, but this condition was minimally studied until its inclusion in the DSM-IV and the ICD-10. Asperger (1944) originally used the word autism in the name of the condition (autistic personality disorder) for a group of cases in which boys had severe social impairment and motor problems but apparently good verbal skills; because of the international political climate, he was unaware of Kanner’s (1943) report the preceding year. Despite the apparent verbal precocity of his subjects (they were described as “little professors”), Asperger emphasized that these children were extremely socially isolated and could not join groups, and their lives and that of their families were dominated by unusual and often esoteric special interests. Asperger also mentioned other features that differed significantly from Kanner’s early description, including verbal precocity, motor clumsiness, and positive family history. Asperger’s report received rather little interest in the English-language literature until Lorna Wing’s report and provision of a series of case studies (Wing 1981). A lack of consistency in the definition of Asperger’s syndrome complicated research studies, although some areas of potential differentiation from autism were observed, such as distinct neuropsychological profiles. Another complication was the plethora of terms, from a range of disciplines, used in describing the difficulties of verbally able but socially impaired individuals; these terms included right hemisphere learning difficulty, semantic pragmatic language disorder, and nonverbal learning disability (Volkmar et al. 2013a).

A tentative decision had been made to include Asperger’s disorder in ICD-10, and the DSM-IV field trial provided some data supporting this inclusion. The relatively large (nearly 50) number of cases with clinician-assigned diagnoses of Asperger’s disorder differed in important ways both from similarly cognitively able cases of autism and cases of pervasive developmental disorder—not otherwise specified (PDD-NOS) (Volkmar et al. 1994). The original definition provided in the DSM-IV proved somewhat controversial, differing in several ways from that originally suggested by Asperger. As a result, several different approaches to diagnosis remained in active use. However, studies using more stringent approaches continued to suggest distinctions from higher-functioning autism, and a body of literature on the important potential differences for clinical intervention began to develop (Volkmar et al. 2013a). A number of studies indicated a frequent association between Asperger’s syndrome and the neuropsychological profile of nonverbal learning disability (Klin et al. 1995, Lincoln et al. 1998, Volkmar et al. 2013a). Interest in the condition increased dramatically (from fewer than 100 scientific papers from 1944 to 1993 to approximately 1,700 after 1994, when the DSM-IV appeared). The decision to eliminate Asperger’s from the DSM-5 has been controversial (Fitzgerald 2012, Gibbs et al. 2012, McPartland et al. 2012, Tanguay 2011, Tsai 2012).

Two other disorders characterized by developmental regression were also included in the DSM-IV within the PDD category. Loss of skills in autism was initially reported by Kanner (1943). Many
years before Kanner’s report, Heller, an Austrian educator and researcher, described a group of children who developed normally for some years before experiencing a marked loss of skills and presenting with autistic-like symptoms (Heller 1908, Westphal et al. 2013). Although childhood disintegrative disorder (also sometimes referred to as Heller’s syndrome or disintegrative psychosis) is rare, a distinct course with a dramatic loss of skills and an apparently worse outcome than in autism is evident (Volkmar 1992). Typically, the age of regression is between three and four years (although sometimes earlier or later), but early development is normal and the loss of skills unequivocal. A variety of clinical features are noted during the relatively rapid period of skill loss, and there is some suggestion of potential differences from autism in neuropsychological functioning (Westphal et al. 2013). The ability to identify these rare cases for study may be complicated by the loss of the diagnostic category in the DSM-5.

Rett’s disorder was also included in the PDD class in the DSM-IV. This progressive disorder, initially apparently confined to girls, was characterized by a very specific form of deterioration and clinical course and features. In his original report, Rett (1966) speculated that the disorder might be a form of autism, partly because of the apparent loss of social skills in the preschool period. However, the subsequent course was quite distinctive. At the time the DSM-IV was developed, Rett’s disorder was not considered a form of autism, but the PDD group seemed the most logical place for its inclusion (Rutter 1994). Subsequent to its inclusion, a specific genetic etiology was found, relating to a single gene defect (MECP2—a regulator gene on the X chromosome; Zoghbi 1988). In the DSM-5, the category is eliminated, but Rett’s disorder can be designated as a specifier for genetic etiology. The rationale for this decision was in part the concern about listing specific genetic conditions, as such, in the DSM-5; this approach suggests that as additional genetic etiologies of autism are identified, they may be removed from the DSM.

Consistent with the DSM-III and III-R, the DSM-IV included a subthreshold condition for cases with problems suggestive of autism but failure to meet full diagnostic criteria. The DSM-IV’s PDD-NOS and the ICD-10’s equivalent “atypical autism” gave clinicians (and researchers) the potential to identify cases with some features of autism in the absence of full criteria. This category had important implications for service access or, in research, for family studies of the broader autism phenotype and autism spectrum. For PDD-NOS, there was an unintentional overlap with an older diagnostic concept, “atypical personality development” (Rank 1949, Rank & MacNaughton 1949). But as a practical matter, such “subthreshold” or PDD-NOS cases clearly greatly outnumbered those with more “classic” autism (Fombonne 2005). Given the nature of the obviously very loose definition of PDD-NOS, there has been considerable diagnostic heterogeneity, and attempts have been made to identify specific subgroups/subtypes (Towbin 2005). PDD-NOS was removed from the DSM-5, emphasizing the requirement to fulfill a full breadth of diagnostic criteria to meet the threshold for ASD. A novel DSM-5 diagnosis, social communication disorder, reflects symptoms in only the social and communicative domains (absent repetitive behaviors) and corresponds more directly to one form of DSM-IV PDD-NOS.

DIMENSIONAL APPROACHES TO DIAGNOSIS

Although dimensional instruments and assessments are not the primary focus of this review, we should note that they are important and, particularly with the DSM-5, have had a major role in the development of categorical criteria. Since the publication of Rimland’s first diagnostic checklist (Rimland 1968), many such instruments have been developed—some for screening and others for diagnosis (Lord 2013, Stone & Ibanez 2013). Some of these focus on infants and younger children, others on older individuals or the more cognitively able. Some instruments are based on parent or teacher report, others on direct observation. Most focus on autism, but a few concentrate on
Asperger’s disorder (Campbell 2005). In some cases, instruments have been developed specifically to assess the broader range of issues relevant to the broader autism phenotype (Constantino et al. 2003, Constantino & Todd 2003). For the most psychometrically robust instruments, extensive training and demonstration of reliability are needed. Other dimensional approaches—for example, the use of tests of intelligence, executive functioning, and adaptive skills—are, of course, also highly relevant to the assessment of persons with ASD. It is important to note that unlike these normative approaches, those designed for use in ASD face special challenges.

Challenges for dimensional assessment in ASD include the broad range of syndrome expression, age- and IQ-related issues in syndrome expression, the relevance of historical information versus current examination, and the degree to which sometimes highly infrequent (but important) behaviors are sampled. There is also the standard challenge of ensuring adequate reliability among clinicians (Lord 2013). Aspects of item administration or scoring can present difficulties. The degree to which clinical judgment is important also varies across instruments, as does flexibility in administration and intended age range or developmental level. In general, these instruments appear to work best in the mid range of age (school age) and level of ability (mild to moderate intellectual disability), with greater challenges beyond the mid range. There can also be a cost of balancing clinical sensibility with reliability of coding in terms of the need for concrete operationalization of symptom manifestation.

The potential for quantifying symptoms with dimensional measures has important research implications. These approaches hold promise to provide measures of severity that can be assessed during treatment and to inform potential new approaches to identifying subtypes. The balance of strengths and weaknesses may vary with context, as research and clinical settings favor distinct characteristics of instruments. Screening instruments may focus on parent report or direct observation (or both), and they have a different set of issues (Stone & Ibanez 2013). As noted above, available diagnostic instruments also probably work best in school-age children with ASD who have some language disability and mild to moderate cognitive disability. Their use becomes more complex in other age and IQ ranges. These issues are relevant to the implementation of the DSM-5 because the field has become increasingly reliant on the diagnostic application of standardized dimensional instruments. Although the desire to make use of a considerable body of research on these instruments is commendable (Regier et al. 2010), challenges will likely arise in translating them into real-world clinical practice.

MOVING TOWARD DSM-5

In creating the DSM-5 criteria, the Neurodevelopmental Disorders Work Group sought to preserve the strengths of the DSM-IV approach while improving upon its limitations. The criteria provided in DSM-IV were highly effective in supporting the development of standardized assessment methods and in facilitating research, with scientific publications on autism increasing dramatically. These advances were aided by the consistency between the DSM-IV and ICD-10 criteria. Comparable American and international systems fostered multinational collaborations and the emergence of similar autism programs around the world. Increased awareness of autism has brought greater support for earlier intervention and improved outcomes (Comm. Educ. Interv. Child. Autism Natl. Res. Counc. 2001, Howlin 2013). Advances in the genetics of autism have been dramatic since the DSM-IV and DSM-IV-TR were published, with a number of potential genes contributing to autism now identified (Rutter & Thupar 2013). The focus of the DSM-IV and ICD-10 on consistent application across functional levels has increased awareness of severe social disabilities in more cognitively able individuals and correspondingly increased access to services for this population.
Despite this growth during the tenure of the DSM-IV, criticisms have been raised since its inception. The DSM-IV field trial was large and international in nature and included a wide range of individuals (varying in age, level of cognitive ability, and other factors), but it was not an epidemiological sample. Cases did include young children and adults, but infants with autism were not adequately represented (Chawarska et al. 2008), and work on DSM-IV criteria suggests reduced diagnostic stability before age three (Lord 1996). Children diagnosed with autism often displayed the social and communicative features of the disorder prior to age three, with the “restricted interests” criterion emerging after age three and preceded by sensory difficulties not included as diagnostic criteria in DSM-IV (Chawarska et al. 2008).

Substantial concerns have been raised about the validity of DSM-IV PDD subtypes (Mayes et al. 2001, Ozonoff & Griffith 2000). The validity of these clinically defined subtypes has been questioned (Howlin 2003), and to varying degrees, the validity of all subtypes, barring autistic disorder, has been challenged—although, at least for Rett’s disorder, a specific genetic etiology has been found. For Asperger’s disorder, concerns have been raised about the stringency (or lack thereof) of the DSM-IV diagnostic criteria (Miller & Ozonoff 1997, Woodbury-Smith et al. 2005). In response to dissatisfaction with the original description, the entire written narrative for Asperger’s disorder was replaced in the DSM-IV-TR, but no changes were made to the wording of actual diagnostic criteria (Am. Psychiatr. Assoc. 2000). Several different approaches to the diagnosis of this condition remain in use, with more stringent criteria sets apparently more likely to yield differences in neuropsychological profiles, family history, and comorbid psychiatric conditions in probands (Klin et al. 1995, 2005a,b, Lincoln et al. 1998). Lord and colleagues (2011) reported in a large multisite study that assessment location was more predictive of a diagnosis of Asperger’s syndrome than were characteristics of the individual child, suggesting that the diagnosis was applied inconsistently across clinics. For childhood disintegrative disorder, concerns have related to the reliability of parental report of regression and whether the condition is sufficiently distinctive to merit inclusion as a separate disorder (Hansen et al. 2008, Jones & Campbell 2010, Kurita et al. 2004, Luyster et al. 2005, Rogers 2004). However, a major source of this disagreement relates to the nature of methods used to assess regression. In one study, approximately 20% of a large sample of parents of children with autism reported regression, but only a small fraction of these could clearly be shown to have regressed significantly (Siperstein & Volkmar 2004). For the broader PDD-NOS group, recognition of many genetic etiologies has led to the conceptualization of PDD-NOS as a milder end of a continuum or spectrum (the broader autism phenotype; Piven 2001). This subgroup is undoubtedly the most heterogeneous (Towbin 2005) and the least well studied.

**DSM-5 Criteria**

The fifth edition of the DSM was published in May 2013 (Am. Psychiatr. Assoc. 2013). The PDD class was merged into a single class of ASD, and a related disorder, social communication disorder, was added. The umbrella category of ASD encompasses the previously distinct PDDs of autistic disorder, Asperger’s disorder, and PDD-NOS. The triad of impairments spanning social interaction, communicative behavior, and repetitive and restricted behaviors was collapsed into two domains, preserving restricted and repetitive behaviors but merging social and communicative difficulties into a single domain. This social-communication category became monothetic, i.e., it required that a person demonstrate symptoms across all three clusters to meet criteria for ASD. The restricted and repetitive behaviors domain remained polythetic, requiring evidence of symptoms in two of four symptom groupings; the repetitive and restricted behavior domain also added a symptom cluster reflecting sensory difficulties. A specification that symptoms must be
present in early development enacted a universal onset criterion, with the caveat that problems may not manifest until social demands exceed limited capacities. The new diagnostic category, social communication disorder, defined by pragmatic difficulties and problems in the use of verbal and nonverbal communication in social contexts, was introduced as a distinct disorder from ASD; although similar to PDD-NOS in many regards, it is grouped as a communication disorder and with a corresponding ICD-10 code mapping onto specific developmental disorders of speech and language. Symptoms must result in clinically significant impairment across contexts and cannot be explained by intellectual disability or global developmental delay alone, i.e., difficulties exceed those predicted based on overall developmental level. The DSM-5 stipulates that a diagnosis of DSM-5 ASD should be applied to individuals with well-established DSM-IV diagnoses on the autism spectrum; addressing concerns about increased diagnostic stringency, this exception rule allows individuals with Asperger’s disorder and PDD-NOS. This effectively maintains the DSM-IV system, but it also effectively creates two systems that remain in use.

DSM-5 criteria for ASD introduce a series of specifiers, reflecting a volume-wide effort to include themes and descriptors that apply transdiagnostically. These addenda provide related information about the current presentation of a person meeting criteria for ASD. A first specifier describes whether a known etiological factor (i.e., medical condition, genetic syndrome, or environmental exposure) is present. The second, a severity specifier common across DSM-5 diagnostic categories, describes required level of support and impact on a person’s levels of functioning separately for each domain of symptoms (i.e., social communicative and repetitive behaviors). Severity specifiers range from levels 1 to 3, indicating a need for support, substantial support, or very substantial support, respectively. It is made explicit that severity specifiers do not correspond directly to qualification for or need for services because they do not account for a person’s profile of abilities or an individual hierarchy of intervention objectives. The third specifier indicates whether intellectual impairment is present. The fourth specifier indicates whether language impairment is present and is to be provided separately for receptive and expressive language along with a concise description of the actual language skills possessed by the individual. The final specifier indicates whether catatonia is present. As noted previously, the use of these specifiers is meant, in some ways, to replace the previous multiaxial system.

Studies Evaluating DSM-5 Criteria

A number of studies have explored the possibility that the population of individuals meeting criteria for ASD could change using the new criteria. A particular focus has been on whether the DSM-5 revisions could affect the proportion of individuals meeting criteria for ASD compared to DSM-IV-TR. To investigate this possibility, our own research group reexamined the large data set collected as part of the DSM-IV field trial (McPartland et al. 2012). Sensitivity and specificity were examined by mapping symptom checklists collected during the field trial onto proposed DSM-5 diagnostic criteria. The algorithm suggested that 60.6% of individuals clinically diagnosed with an ASD met revised diagnostic criteria; 94.9% of individuals without a clinical diagnosis were accurately excluded from the spectrum. Sensitivity varied by diagnostic subgroup such that individuals with milder forms of autism were less likely to meet criteria (Asperger’s disorder = 0.25; PDD-NOS = 0.28) than individuals with classic autism (autistic disorder = 0.76). Individuals with cognitive impairment were less likely to meet criteria (IQ < 70 = 0.70) than were individuals with normative intellectual abilities (IQ ≥ 70 = 0.46). Another study of cognitively able adults compared DSM-IV, ICD-10, and DSM-5 criteria (Wilson et al. 2013) and indicated that over half of cases with an ICD-10 PDD diagnosis also met DSM-5 criteria for ASD, with nearly 20% of those not meeting criteria for DSM-5 ASD meeting criteria for social communication.
disorder. Mattila and colleagues (2011) obtained similar results in a smaller study that relied on a previous version of the proposed criteria. This study indicated that, overall, 46% of individuals with ASD met DSM-5 diagnostic threshold. Smaller numbers of individuals with average IQ (36%) or Asperger’s syndrome (0%) met revised diagnostic criteria. Worley & Matson (2012) demonstrated that individuals meeting proposed DSM-5 criteria tended to have more severe impairments than individuals meeting DSM-IV-TR criteria, a pattern replicated by Matson et al. (2012a), who also found that 36.5% of individuals in a sample of developmentally disabled adults with ASD failed to meet proposed criteria. Matson and colleagues (2012b) reported that 47.8% of toddlers meeting DSM-IV-TR ASD criteria did not meet DSM-5 criteria. A large study using a national registry found that up to 12% of individuals with DSM-IV ASD may not meet DSM-5 criteria, and the effect is most pronounced among females (Frazier et al. 2012). A study of an existing dataset of adults indicated higher sensitivity with parent report than observational assessment, highlighting the influence of assessment method on ascertainment (Mazefsky et al. 2013). A recent study with a large sample size evaluated with standardized instruments demonstrated approximately 91% sensitivity, suggesting that individuals currently meeting criteria for ASD will continue to do so according to the DSM-5 (Huerta et al. 2012). Specificity was reduced when only one standardized instrument was available (parent report or observational measure).

Several studies have specifically focused on the DSM-5’s applicability to specific subgroups potentially affected by the increased emphasis on symptom endorsement in all three DSM-IV symptom domains. Toddlers have been a focus because of the DSM-5’s increased emphasis on the presence of repetitive and restricted behaviors, which may not manifest in early development. In an initial study, Worley & Matson (2012) reported on a large sample of cases and found fewer young children met DSM-5 criteria relative to DSM-IV criteria. A subsequent study focused on toddlers found similar results (Matson et al. 2012), as has work by Barton et al. (2013). Other studies have suggested that individuals meeting DSM-IV criteria for PDD-NOS may fail to exhibit the breadth of symptoms required by DSM-5 (Gibbs et al. 2012, Taheri & Perry 2012).

Most published studies suggest that DSM-5 criteria offer greater specificity but may result in reduced sensitivity, especially for specific subgroups, including very young children, cognitively able individuals, and individuals meeting criteria for DSM-IV PDD-NOS (Frances & Widiger 2012, Jones 2012, Regier et al. 2013). All of these studies all have limitations, including the use of historical data, reliance on outdated versions of proposed DSM-5 criteria, or exclusive reliance on specific assessment methods (i.e., clinician observation versus parent report). Most importantly, none of these studies compared criteria sets prospectively by concurrently evaluating patients on multiple criteria sets using the actual DSM-5 criteria set. There has also been significant methodological variability among these studies in terms of data collection (e.g., reanalysis of data versus collecting data) and in terms of symptom endorsement (e.g., clinical assessment versus standardized assessment), and these factors are demonstrated to influence whether individuals meet criteria (Mazefsky et al. 2013). Most of the aforementioned studies were obtained in research settings, so the sensitivity and specificity that will be evident in purely clinical settings remains unclear (Tsai 2012). A key short-term objective will be to understand the application of new criteria in smaller, purely clinical settings and in contexts in which the diagnostic criteria set itself will be employed as a checklist without reliance on standardized assessment measures.

Studies have also addressed the move from a three-factor to a two-factor conceptualization of symptom domains. Although the large research data set analyzed for DSM-5 indicated that a two-factor solution was most parsimonious, other studies (e.g., Sipes & Mattson 2013) suggest that several different factor solutions are possible. The DSM-IV field trial factor analysis yielded reasonable two-, three-, or five-factor solutions (Volkmar et al. 1994). The three-factor, polythetic solution adopted by the DSM-IV yielded more flexible application (i.e., more than 2,000 possible
combinations of criteria in DSM-IV could result in an autism diagnosis compared to less than 20 in DSM-5).

UNANSWERED QUESTIONS

As of composition of this review, the DSM-IV exception rule had not yet been examined in research studies or extensively discussed in popular media. Initial impressions suggest that the exception rule will resolve several ongoing concerns and present interesting questions for the implementation and carrying forward of DSM-5 criteria. By prequalifying individuals with a DSM-IV diagnosis, this clause temporarily assuages concerns about changes in the prevalence of ASD wrought by the change in diagnostic criteria. However, this approach creates, de facto, two concurrent systems of diagnosis. Thus the existing population of individuals with ASD will continue to meet criteria and, consequently, remain entitled to extant clinical, medical, and educational services. New cases must meet DSM-5 criteria and therefore will be subject to potentially different thresholds for accessing services. Many stakeholders had also questioned whether publication of DSM-5 criteria could lead to mandated diagnostic updates; the exception rule is intended to prevent this. Potential changes attributable to the revised symptom set would emerge more gradually as children are born or age into a diagnostic system based on DSM-5.

The approach adopted raises several interesting questions for clinicians to consider in the coming years. How will it apply to children with well-established DSM-IV diagnoses who, after treatment, no longer meet the threshold for either criteria set? Guidelines for the regularity of reevaluation and transition to DSM-5 criteria within the life span of individuals need to be established. The DSM-IV exception rule will create a period in which individuals concurrently in the service system may have diagnoses based on different systems of classification. Long-term research studies will need to devise strategies to manage individuals with similar phenotypic characteristics who may meet or fail to meet the DSM-5 diagnostic threshold based on whether they had previously been evaluated according to DSM-IV criteria.

The issue of sample comparability in research studies has been a matter of debate. Several studies suggest that the new criteria may affect the compatibility of future research with prior work. This is a known risk in development of any new diagnostic criteria set. It is highly relevant in ASD, given the volume of research conducted during the tenure of DSM-IV. If DSM-5 changes the composition of the spectrum, comparisons of current and historical samples would be unreliable. The removal of subdiagnoses also complicates mapping future and historical samples onto one another. On the basis of these observations, Ghaziuddin (2010) has suggested the potential for a loss of information building from research conducted using the current diagnostic system. A related challenge will be compatibility with ICD-10 criteria (and possibly criteria for ICD-11, currently in preparation), which may make it difficult in future work to compare studies conducted inside versus outside of the United States.

The sociological impact of these changes is not yet known. Elimination of subdiagnoses may impact those who have found a diagnosis important in terms of service access or insight into one’s difficulties (Kaland 2011). Removal of subtypes also precludes their possible validation (Baron-Cohen 2009, Ghanizadeh 2011, Mandy et al. 2011). Wing and colleagues (2011) raised concerns about the nonspecificity of sensory behavior, omission of a criterion describing lack of imagination, challenges in verifying onset criteria in adults, and problems applying social-communicative criteria in infancy.

Concerns about the impact of DSM changes should be considered in the context of sweeping changes occurring in psychiatric research. The director of the National Institute of Mental Health has expressed the organization’s intention to move away from research based on diagnostic
categories toward emphasis on biological processes that can be applied transdiagnostically (Insel 2013). It is possible that DSM-based groupings may become decreasingly relevant in federally sponsored research.

**DISCLOSURE STATEMENT**

The authors are not aware of any affiliations, memberships, funding, or financial holdings that might be perceived as affecting the objectivity of this review.

**LITERATURE CITED**


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Autism Science Foundation: http://www.autismsciencefoundation.org
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