

## Chapter

# Diagnosis and Definition of Autism and Other Pervasive Developmental Disorders

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## Introduction

Approaches to classification of medical, mental health, and developmental problems have many different uses, origins, and orientations (Rutter & Uher, 2012). Ideally, they facilitate communication for research and clinical work and can help inform public health, educational, and other needs, and social policy (Volkmar et al., 2017b). In this chapter, we provide a relatively concise overview of the different approaches used in the diagnosis of autism/autism spectrum disorder (ASD). We emphasize that even though we discuss various diagnostics systems, it is the *process* of diagnosis that is usually most important in helping the particular individual, i.e., a diagnosis or set of diagnoses cannot be viewed outside the context of a fuller description based on clinical assessment.

A range of competing considerations arise relative to official diagnostic systems. These systems should neither be too *broad* nor too *narrow*. There are some intrinsic tensions in striking a balance between clinical and research needs. For some purposes, such as educational interventions, broader diagnostic approaches may be more useful. For other purposes, such as the manifestations of a single gene disorder, a narrower diagnostic view may be more appropriate. Even in the latter case a range of clinical phenotypes may, of course, be observed. Additional considerations arise relative to the issue of the degree of impairment. For example, if an individual has the manifestations of Tourette's disorder but is *not* impaired do they have a disorder – even if she or he might pass a genetic risk on to his children? Such problems arise in autism with respect to the broader autism phenotype (those who have subclinical expression of the traits associated with autism; discussed in more detail later) and boundaries of the condition in relation to the broad range of normalcy (Ingersoll & Wainer, 2014).

As discussed later in this chapter, dimensional diagnostic considerations and approaches also have important uses in that they may provide more nuanced information. Historically tests of intelligence and adaptive skills have routinely been used in assessment of individuals with autism but a number of diagnostic instruments have now been developed as well. Presently, reliance on theoretical orientation (once the guiding light of DSM I and II) has lost favor in preference to a more statistically and empirically oriented approach that sticks to clinical phenomenology (Volkmar et al., 2017b). It is now recognized that while theory has important uses, it can be a handicap in that theoretical

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orientations are often not shared, and classification systems based on theory can quickly result in major complications. Indeed since 1980, diagnostic systems have been trending away from theoretical orientations and toward the research diagnostic criteria approach (Spitzer et al., 1978).

It should be noted that like all conditions, but perhaps even more so in autism, developmental correlates of the condition are important. Indeed, as we discuss subsequently, viewing autism as a “social learning disability” places an increased focus on understanding the implications of the diagnostic label for learning as well as for treatment. Discussed in detail later in this volume, issues of diagnoses also have important implications for genetic and epidemiological work. Further, the profound changes in outcome (i.e., with earlier diagnosis and treatment) have impacted what is considered the “face” of autism over time. For example, it is becoming more and more common for individuals with ASD to graduate secondary schools, move to college, and join the workforce (Volkmar et al., 2017a). Diagnostic considerations must encompass the ability of the individuals to change while, if appropriate, retaining their diagnosis. Indeed many “optimal outcome” (Fein et al., 2013) individuals exhibit continued problems with associated conditions like anxiety and depression. Moreover, diagnostic complexities tend to increase when gender- and culture-specific manifestations of autism are considered (Freeth et al., 2014; Øien et al., 2017).

## History of Diagnostic Concepts

The description of the syndrome of Early Infantile Autism by Leo Kanner (Kanner, 1943) remains a “classic,” if not “the” classic, example of a clinician researcher “discovering” a previously unknown condition. His original description of 11 cases emphasized two essential features: (i) “autism” (a term borrowed from Eugen Bleuler’s 1911 work in the field of schizophrenia, used to describe a pattern of social disinterest and self-isolating behaviors) and (ii) something he termed “insistence on sameness/resistance to change.” This latter notion ended up covering several somewhat different concepts – trouble with change, a preference for the familiar and repetitive, and some of the unusual mannerisms and motor stereotypies he noted (Volkmar et al., 2014c). Kanner also mentioned many of the aspects of autism that remain commonly observed today, e.g., major problems with communication, including a failure to talk at all or echolalia speech. He took pains to note that the children did not have an unusual appearance (i.e., unlike children with trisomy 21) and sometimes had isolated areas of ability. The year after Kanner’s report, the Viennese medical student Hans Asperger (Asperger, 1944) also used the term autism in describing an unusual personality type in a small group of boys who had difficulties in joining groups. These boys were socially very isolated but had strong circumscribed interests. These two clinical descriptions (of Kanner and Asperger) differed in many respects but did have an important point of connection: the emphasis of problems of social engagement (Klin et al., 2014).

With the wisdom of hindsight, it is, of course, possible to identify descriptions of children prior to the release of the Kanner paper who likely would have had these conditions. For example, reports of “wild” or feral children (Favazza, 1977; Candland, 1995) may have exhibited autism and either run away from their parents [a problematic behavior in autism that remains common today (Anderson et al., 2012)] or were abandoned by them (a practice still common in some developing countries). In their historical review

of Kanner's work and the history of autism Donvanm and Zuker (2016) note that well documented cases may have been noted in the 1800s without an awareness of the distinctiveness of their disorder.

Following Kanner's initial report subsequent research only slowly began to accumulate. His use of the term autism became a source of confusion between autism and childhood schizophrenia, i.e., Bleuler's earlier use of the term to describe self-centered thinking in schizophrenia suggested to many that infantile autism was potentially the first manifestation of schizophrenia. Additional sources of confusion had to do with the early impression that children with autism did not exhibit intellectual deficits when, despite their occasional isolated areas of nonverbal ability (Hermelin, 2001), most had full scale scores in the intellectual disability range (Tsatsanis, 2005). The common psychodynamic theorizing of the time also led some to speculate environmental factors might play a role in pathogenesis (Despert, 1971). Of particular note was the psychodynamic theory of the "Refrigerator Mother" championed by Bettelheim (1967) which suggested that autism was a mental state of children withdrawing from the external world and into themselves as a result of the extreme circumstances created by the emotional abandonment and rejection of their mothers. Accordingly, early treatment techniques often emphasized dynamic treatments and sometimes separation from parents (Bettelheim, 1974).

As research began to increase several findings emerged. Kolvin and associates (Kolvin, 1971; Kolvin et al., 1971) demonstrated that autism is *not* a manifestation of schizophrenia with the production of comparative studies clearly distinguishing childhood schizophrenia and autism by their associated symptoms, phenomenology, age of onset, and family history. Secondly, evidence was mounting to support that autism was clearly a brain-based disorder – children with autism exhibited a number of neurological problems including a high risk for development of recurrent seizures in adolescence (Rutter, 1972). Thirdly, studies of fraternal and identical twins made it clear that autism was a strongly genetic disorder with much higher concordance rates among identical twins (Folstein & Rutter, 1977). Finally, studies that compared unstructured psychotherapy to structured teaching suggested that the latter was much more effective in producing developmental gains (Bartak & Rutter, 1973).

These advances in research led to an awareness of the need for better definitions of autism and related disorders. This awareness occurred within the context of a broader movement in psychiatry to recognize basic features of disorders with minimal theoretical "baggage" so as to increase reliability of diagnoses (Spitzer et al., 1978) as well as an awareness of the need for the consideration of broader contextual factors in how disorders were expressed (Rutter et al., 1975). Several noteworthy attempts were made to provide more truly operational definitions of autism. Ritvo and Freeman (1978) proposed a definition that incorporated many of the features mentioned by Kanner but also included other aspects (e.g., unusual sensory responses, unusual rates or sequences of development). This attempt was important in that it represented a consensus of a group of experts in the field (National Society for Autistic Children; NSAC). At the same time, Rutter (1978) provided a synthesis of Kanner's (1943) definition and subsequent research, emphasizing difficulties that were distinctive to this condition (e.g., not just due to lowered mental age). Of note, both of these proposed definition updates were associated with unusual behaviors emerging during the first 30 months of life. These new more rigorous diagnostic approaches had a significant impact on the landmark –, 3rd edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-III; APA, 1980).

## Difference in Approaches to and Challenges for Diagnosis

As noted previously, several important developments and conceptual shifts regarding autism occurred in the decades after Kanner's initial description of the condition. These included the growing awareness that autism was a brain-based and strongly genetic disorder, and that it responded best to structured educational treatment approaches. In addition, several attempts were made to provide great diagnostic specification to Kanner's original "definition" (actually a clinical description). Of particular note were the NSAC definition proposed by Ritvo and Freeman (1978) and Rutter's synthesis (1978) of Kanner's definition with subsequent research. Additionally, as mentioned above, several important moves were afoot more broadly relative to psychiatric diagnoses in general, and for childhood disorders, like autism, in particular. Among these were growing movements away from more theoretically based classification schemes to more "phenomenologically" based and clinically descriptive ones, as well as attempts to provide a broader context to disorders by noting associated medical conditions, developmental problems, and so forth. Much of this work grew out of the attempts by Guze and others in the Washington University Psychiatry Department (Woodruff et al., 1974).

## Categorical Diagnostic Approaches

The first two iterations of the Diagnostic and Statistical Manual of Mental Disorders (DSM) produced by the American Psychiatric Association (APA) were released in 1952 and 1968. In both of these early editions, the term "autistic" was solely used as a descriptor for social withdrawal behaviors in individuals with schizophrenia, and most children who expressed the symptoms described by Kanner were diagnosed with what was referred to as childhood schizophrenia; a label designed to describe the early manifestations of symptoms that would eventually develop into adult schizophrenia (APA, 1952, 1968). With awareness of the marked limitations of prior editions of the DSM and of the need for major changes, including in the area of childhood onset psychiatric disorders, the decision was made to radically change the approach employed in DSM-III (APA, 1980). This ended up being a critical turning point in the evolution of what has been considered the diagnostic features of autism in the over 70 years since Kanner's initial clinical descriptions (see Table 1.1).

### DSM-III

The landmark 3rd edition of the DSM represented a major shift in approaches to psychiatric diagnoses, incorporating an emphasis on clinical description (rather than theoretical formulation) with the goal of attaining increased reliability and refinement of diagnostic constructs. In recognition of the large body of work showing its distinctiveness as a disorder, for the first time autism (precisely, "infantile autism") was officially included in the DSM-III as a unique disorder (Spitzer et al., 1979). It was included in a new "class" (super-category) of disorders termed the pervasive developmental disorders (PDD). This term had (by design) no previous history and was meant to be theoretical. In retrospect, another term like ASD or autism and related conditions might have been a more appropriate choice. In any case, official recognition of autism as a unique disorder was a major step forward.

The actual definition of infantile autism in DSM-III was in general just that, i.e., one most consistent and descriptive of the earliest form of the disorder. A set of monothetic

**Table 1.1** The evolution of autism as a diagnostic concept from Kanner to DSM-5

Source	Summary of approach
Kanner (1943)	Onset thought to be congenital includes two essential features: <ul style="list-style-type: none"> <li>• Autism (lack of social engagement)</li> <li>• Resistance to change/insistence on sameness</li> </ul>
Rutter (1978)	Synthesis of Kanner's work within subsequent research: <ul style="list-style-type: none"> <li>• Social deviance (not just due to intellectual delay)</li> <li>• Language problems or absent language (also not just due to intellectual delay)</li> <li>• Resistance to change/insistence on sameness</li> <li>• Early onset</li> </ul>
NSAC (Rivto & Freeman, 1978)	Included unusual sensitivities and patterns of development (differences in rates and sequence of development)
DSM-III (APA, 1980)	First official recognition of "infantile autism": <ul style="list-style-type: none"> <li>• Monothetic definition of autism – more in keeping with Rutter (1978) but focused on more classic presentation in early childhood</li> <li>• 4 other conditions in new category of PDD</li> <li>• "Residual infantile autism" category for adults (once met criteria but no longer did so)</li> <li>• Atypical PDD (sub-threshold category)</li> </ul>
DSM-III-R (APA, 1987)	More developmentally oriented and flexible approach: <ul style="list-style-type: none"> <li>• 16 criteria in 3 categories (social, communication/play, odd behaviors)</li> <li>• Polythetic (at least 8 criteria must be met, 2 of which were social)</li> <li>• Field trial used but this was limited in important respects; problems with false positive rates</li> <li>• Sub-threshold category retitled to PDD-NOS</li> </ul>
DSM-IV (APA, 1994) and ICD-10 (WHO, 1993)	Convergence of DSM-IV and ICD-10 represents a major breakthrough: <ul style="list-style-type: none"> <li>• Polythetic and flexible definition</li> <li>• Similar in organization to DSM-III-R but with new/revisited criteria</li> <li>• New categories recognized included Asperger's disorder; PDD-NOS retained</li> </ul>
DSM-5 (APA, 2013)	Major revision <ul style="list-style-type: none"> <li>• Made extensive use of data collected on standardized diagnostic instruments, but limited field trial data</li> <li>• Social and communication items now grouped together (polytheism)</li> <li>• ASD replaces disorders within DSM-IV PDD category</li> <li>• Addition of new category (social communication disorder) with unclear relationship to ASD</li> <li>• Previously "well-established" DMS-IV diagnoses (e.g., Asperger's disorder) could be "grandfathered" in</li> </ul>

Abbreviations: ASD = autism spectrum disorder; APA = American Psychiatric Association; DSM = Diagnostic and Statistical Manual of Mental Disorders; ICD = international classification of diseases; NOS = not otherwise specified; NSAC = National Society for Autistic Children; PDD = pervasive developmental disorder; WHO = World Health Organization.

criteria (i.e., all criteria had to be met) were provided, including pervasive deficits in social interaction of the type seen in the youngest and most seriously impaired young children. This was eventually recognized as a problem in that it became clear that social skills would continue to develop over time in children with infantile autism – albeit in rather odd ways (Wing & Gould, 1979; Davis & Carter, 2014). The criteria proposed were, in some respects, consistent with Rutter's earlier (1978) approach but the lack of

a developmental orientation was problematic. Instead the problem of changes in symptomatic presentation was addressed in a new way – the inclusion of a category called “residual infantile autism” for persons who once met criteria but no longer did so. The terminology selected for this category [in ways harkening to the recent concept of “optimal outcome” (Suh et al., 2014)] was unfortunate, as for most individuals, problems were far from “residual.” Consistent with Rutter’s (1978) paper, an onset of no later than 30 months was also proposed as an essential feature.

Other complications that arose over “new” concepts were included in the overarching atypical PDD group. For all categories in DSM-III an “atypical” code was included (i.e., for “sub-threshold cases”) where precise criteria were not met but there was still a need for service. In this case (of atypical PDD) the diagnostic term had an unanticipated historical connection, in that the term “atypical personality development” was previously used by Rank (Rank, 1949; Putnam & Rank, 1953) to refer to children with some of the features of what would now be seen as characteristic of autism. The DSM-III also included a category for “Childhood Onset PDD” (COPDD) for cases with an apparent autistic-like condition that developed after 30 months of age. This category probably had to do with an awareness of Kolvin’s work reporting the existence of a few such cases in his large case series (Kolvin, 1971). As a practical matter, these children likely had the condition described much earlier by Heller (Heller, 1908, 1930). Somewhat paradoxically the definition for COPDD was rather more flexible than that of infantile autism and again a category of “residual” COPDD was included to deal with the issue of developmental changes.

Issues with the DSM-III approach were quickly apparent. The lack of developmental orientation was a major concern. The criteria emphasized gross deficits in language development rather than the broader notion of communication (i.e., including nonverbal communication). The decision to automatically exclude children from having both autism and schizophrenia was understandable given the history of confusion of these two diagnostic concepts, but it was unclear why an adult person with autism might not be just as much at risk for schizophrenia as other adults in the population (Volkmar & Cohen, 1991). These concerns were addressed in a rather speedy revision of DSM-III which appeared less than a decade later – DSM-III-R (APA, 1987).

## DSM-III-R

While the release of the DSM-III marked a major advance in its official recognition of autism, an awareness of its lack of developmental orientation was noteworthy. As a result of the overall revision of DSM-III, an attempt was made to address this deficiency in the DSM-III-R (APA, 1987; Waterhouse et al., 1993).

As part of the revisions within the DSM-III-R, the name of the disorder was changed to autistic disorder from the DSM-III name of infantile autism. The very detailed and developmentally oriented diagnostic criteria was much more consistent with views of Lorna Wing and her colleagues (e.g., see Wing, 1981) who advocated for a somewhat broader diagnostic approach. By intention the removal of “infantile” from the name meant that the detailed criteria could be used to apply to the entire range or spectrum of autism, i.e., from young children to adults and from lower to higher cognitive functioning individuals. This approach also put much greater weight on developmental considerations. Finally, the DSM-III concept of “residual” infantile autism was lost with the release of the DSM-III-R.

As in the previous version, DSM-III-R criteria were grouped in three major domains of dysfunction (social, communicative, and restricted interests/behaviors). However, this time the diagnostic criteria were polythetic rather than monothetic. Early age of onset was no longer required (but could be specified). The final diagnostic algorithm (based on a small field trial; Spitzer & Siegel, 1990) included 16 criteria and for the diagnosis to be met at least 8 of these had to apply (with a specified distribution over the three areas). This approach meant that the earlier DSM-III concept of COPDD could be dropped; however, the DSM-III-R continued to include an “atypical” category – the latter replaced with a “not otherwise specified” label (Volkmar & Klin, 2005).

The greater developmental orientation of the DSM-III-R approach was clearly needed. Unfortunately, this more flexible approach also seemed to come at a price with a number of studies suggesting a high false positive rate – this was particularly true with more severe mental retardation. The inclusion of specific examples in criteria was also seen as somewhat problematic as it tended to reify these. An additional complexity (to some extent) was the potential for changes in samples, e.g., in longitudinal and epidemiological studies (Volkmar & Klin, 2005; Volkmar et al., 1992a,b). With the wisdom of hindsight, the field trial was also somewhat problematic in that the comparison group included cases of conduct disorder (where criteria were also being evaluated), i.e., the comparison group was probably highly inappropriate and might have impacted the determination of final scoring rules. All these issues were considered as preparations for DSM-IV – a major revision of DSM – began to be made.

## DSM-IV and ICD-10

For autism and related disorders, the DSM-IV (APA, 1994) and ICD-10 (WHO, 1993) approaches are very appropriately considered together given their very close correspondence. A brief history of the overall similarities and differences in these systems is helpful (for a more detailed summary see Volkmar et al., 2017). The origins of the International Classification of Disease (ICD) can be traced to efforts in the mid-1800s to provide comprehensive information on morbidity and mortality. This was part of a larger effort to improve hygiene and medical care practices by introducing a standardized system for tracking illnesses. A similar effort began in the United States in the late 1800s and these efforts converged in 1900 in the International Classification of Causes of Death. Revisions were made to this system periodically with the World Health Organization (WHO) assuming control of revisions in 1948. As revisions of the ICD took place, updates were made given advances in understanding of pathophysiology and the book itself expanded to cover all illness – including psychiatric disorders. The DSM-III marked a major change in psychiatric diagnoses and around the world this system became the most commonly used. At the same time diagnostic codes had to correspond in the DSM and ICD approaches.

The 10th edition update of the World Health Organization’s International Classification of Diseases (ICD-10) was scheduled to appear in the early 1990s and issues of similarity and difference in the upcoming releases of the DSM and ICD systems were a concern (Volkmar et al., 1994). One important difference was that while the DSM existed in only one version (used for both clinical and research purposes) the ICD adopted a different approach with a detailed set of research diagnostics guidelines and a more general clinical guide to diagnosis (for two rather separate audiences). Other differences existed as well, such as in approaches to co-morbidity, with DSM encouraging multiple diagnoses while

ICD discouraged this practice (see Volkmar et al., 2017b for a discussion). Given the significant growth in research on autism occurring during this time, major differences between the two official diagnostic systems were a significant concern and eventually led to coordination of the two systems (Volkmar et al., 1992).

The fourth edition update of the DSM (DSM-IV) was undertaken several years after the publication of the DSM-III-R (APA, 1987). The process for DSM-IV was different than that employed previously in several ways. Specifically, this process included a series of steps including commissioned literature reviews, data reanalysis, consideration of differences from the draft of ICD-10, as well as a major field trial (Volkmar et al., 1994). This process quickly revealed several important issues relevant to similarities and differences between ICD-10 and DSM-IV. For example, the draft ICD-10 recognized Asperger's disorder (Szatmari, 1991), Rett's disorder (Tsai, 1992; Gillberg, 1994; Rutter, 1994a, 1994b), and the condition termed Childhood Disintegrative Disorder (Volkmar, 1992) for the rare cases in which autism had an onset during later childhood (i.e., after age 3). There appeared to be a consensus in the field that comparability of DSM-IV and ICD-10 would facilitate both clinical work and research (Rutter & Schopler, 1992). The series of papers commissioned for DSM-IV found that while the DSM-III-R was more developmentally oriented than its predecessor, it also seemed to be overly broad (Volkmar et al., 1992a, 1992b). Resultantly, one of the objectives for the DSM-IV was to design a diagnostic system that was both flexible and developmentally oriented, but did not over (or under) diagnose autism based on considerations of age or developmental level.

Endeavoring to clarify and address these issues, a larger international field trial was undertaken in conjunction with the ICD-10 revision (Volkmar et al., 1994). The field trial was a major undertaking with data collected over the course of a year at over 20 sites by more than 100 raters who provided information on nearly 1000 cases. Unlike the DSM-III-R field trial, comparisons of cases were taken only if the rater believed autism was a reasonable consideration in their differential diagnosis. Levels of rater experience and basic information on cases rated was collected along with reliability ratings records. Typically, multiple sources of information (history, past examinations, current assessment) were available and raters judged the quality of information available to them from good to excellent in about 75 percent of cases. In this field trial, inter-rater reliability was generally good and for less experienced raters it improved if explicit diagnostic criteria were employed.

Data from this field trial confirmed that relative to either DSM-III (in its "Lifetime" sense) and the draft ICD-10 the DSM-III-R criteria resulted in a high rate of false positives (in cases with associated severe intellectual disability, the false positive rate was about 60 percent). 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 was employed. The rather more detailed draft ICD-10 research definition worked well but its level of detail posed some issues for use in DSM-IV (which, in contrast to the ICD-10 research definition, was intended for use for both clinical and research purposes). A factor analysis produced several potential solutions including the traditional triad of domains of difficulty (social, communication, and restricted interests), a two-factor solution (social communication and restricted interests), and a five-factor solution (in which the restricted interests criteria sorted into three groups) were also identified. By eliminating some of the ICD-10 criteria a system was produced that yielded a good balance of diagnostic stability and sensitivity and specificity over both age and IQ levels.

Another aspect of the field trial was the issue including other disorders along with autism in the PDD category. Asperger's disorder had been relatively little studied until

Wing's clinical account (Wing, 1981). The tentative decision to include Asperger's disorder in ICD-10 was supported by the field trial data. Nearly 50 cases with clinician-assigned diagnoses of Asperger's were included in the field trial and these cases differed in several ways from similarly cognitively able cases of both autism and the DSM-III sub-threshold category of atypical PDD (Volkmar et al., 1994). The original definition provided in 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 differences from higher functioning autism, and a body of literature on the important potential differences for clinical intervention began to develop (Chiang et al., 2014; Volkmar et al., 2014b). Several studies also noted an association of the condition with the neuropsychological profile of nonverbal learning disability (Klin et al., 1996; Lincoln et al., 1998). As described subsequently, the decision to eliminate Asperger's in DSM-5 has been controversial (Tanguay, 2011; Fitzgerald, 2012; Tsai, 2012; Gibbs et al., 2012; McPartland et al., 2012).

Two additional disorders, characterized by developmental regression, were also included in DSM-IV within the PDD category. At the start of the twentieth century, Heller (Heller, 1908), an Austrian educator and researcher, had described cases in which children grew-up normally for some years before having a marked loss of skills and presentation with what would now be seen as autistic-like symptoms. Although originally termed Heller's syndrome or disintegrative psychosis, ICD-10 adopted the Childhood Disintegrative Disorder for this rare condition – cases which had (apparently) inspired the inclusion of the COPDD category in DSM-III. The second condition had been identified by Rett (1966) in his report of the onset of regression and development of multiple symptoms suggestive of autism in a group of girls. While the clinical features of the condition differed in many ways from autism and the course was quite distinctive (Van Acker et al., 2014), some speculated that it might be a form of autism, partly because of the apparent loss of social skills in the preschool period. The decision to include the condition in DSM-IV and ICD-10 in the PDD class was made given the distinctiveness of the condition and indeed a gene associated with the condition has now been identified (Jellinger et al., 1988).

Consistent with both DSM-III and DMS-III-R, DSM-IV included a sub-threshold condition for cases with problems suggestive of autism or a similar condition but with a failure to meet full diagnostic criteria. In DSM-IV, the term Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS) was used and essentially corresponded to the ICD-10's equivalent term of atypical PDD. This category had important implications for service access and for family studies of the "broader autism phenotype" and autism spectrum (Ingersoll & Wainer, 2014). Unintentionally, for PDD-NOS there was some overlap with an older diagnostic concept "atypical personality development" (Rank, 1949; Rank & MacNaughton, 1949). Several changes in proposal for other categories (notably Asperger's disorder and PDD-NOS) were made in the final stages of the DSM process; these have raised some problems addressed, in part, in the text-revision edition of the DSM-IV-TR (Buitelaar et al., 1999; Miller & Ozonoff, 1997). In order to address some of these concerns for Asperger's disorder, the text was very extensively revised although no changes in the formal criteria were made.

## DSM-5

One of the most dramatic shifts to the diagnostic criteria for autism and related disorders occurred in the most recent iteration of the APA diagnostic classification manual, the

DSM-5 (APA, 2013). For the DSM-5, the PDD category of conditions was disregarded, and was replaced with the singular ASD, which combined the previously distinct conditions from the DSM-IV of autistic disorder (equivalent to Childhood Autism in the ICD-10), Asperger's syndrome, Childhood Disintegrative Disorder, and PDD-NOS. The hope being that a single "spectrum disorder" would better encompass our current understanding about the clinical presentation and course of the PDDs, and would ideally facilitate more effective treatment and identification going forward. In the process of accomplishing this reclassification of four previously distinct conditions into the newly defined ASD, the DSM-5 collapsed the triad of impairments from the DSM-IV (and the ICD-10) into the following duo of core areas of impairment (APA, 2013):

- (i) Persistent deficits in social interaction and social communication (impaired social-emotional reciprocity; deficits in non-verbal social communicative behaviors; and impaired ability to develop, maintain, and understand social relationships).
- (ii) Restricted and repetitive patterns of activities, behaviors, or interests (repetitive or stereotyped motor movements, speech, or use of objects; inflexibility with regard to routine adherence, insistence on consistency, or ritualized patterns of behavior; highly restricted interests that are abnormal in focus or intensity; hypo- or hyper-reactivity to sensory input or atypical interest in sensory aspects of their environment).

To adjust for inclusion of the less severe manifestations of ASD (i.e., Asperger's syndrome, PDD-NOS), the DSM-5 replaced the previous criteria that impairments must be evident before the age of 3, to the more flexible criteria that symptoms must be present during the "early developmental period," and includes the caveat that they may not become fully apparent until later stages of life when social demands surpass the individual's limited abilities (APA, 2013). Additionally, the expression of these symptoms must result in clinically significant impairment in some critical area of functioning (e.g., occupational, social, academic).

This collapse of conditions into the unified ASD category has been met with some resistance. Firstly, there have been concerns raised regarding a decreased sensitivity in the diagnostic criteria as a result of the attempts to increase the diagnostic specificity (Hazen et al., 2013). In particular, the concern is that individuals who would have qualified for a PDD diagnosis under DSM-IV criteria may not under the criteria established in the DSM-5, and as a result would lose access to needed and beneficial services. More specifically, the DSM-5 requirement that both the social/communication and the repetitive behavior impairments must be present could exclude those who previously would have been classified with PDD-NOS, as that diagnosis did not require the repetitive behavior symptoms (Ozonoff, 2012). Studies exploring this concern have produced conflicting results, with some suggesting that 24 percent to 39 percent of individuals who would have previously met criteria for a PDD would be excluded based on the criteria for ASD in the DSM-5, and others reporting that 91 percent of individuals who would meet criteria for a PDD under the DSM-IV would also meet criteria for ASD under the DSM-5 (Hazen et al., 2013). Of note, these studies were consistent in their findings that the updated DSM-5 criteria produced significant improvements in diagnostic specificity.

In an effort to address the concerns over diagnostic sensitivity, the DSM-5 introduced a new condition called Social Communication Disorder, intended to provide a label for children without the repetitive behavior components of ASD, but who still express