One

OVERVIEW

Autism spectrum disorders (ASD) are among the most common childhood disorders, with prevalence rates reaching near 1% of the population (CDC, 2007a, 2007b). Defined as a lifelong neurodevelopmental disorder with a complex genetic etiology, ASD's symptoms tend to unfold over the course of early development. Research indicates that 80% to 90% of parents report their first concerns about their child's development by the second birthday and often earlier. However, the mean age of diagnosis continues to be well over the age of three despite these concerns (Chawarska et al., 2007). Moreover, when experienced clinicians make a diagnosis of ASD at 18 to 24 months, the stability of diagnosis is quite strong, also around 80% to 90% (Chawarska et al., 2009). This highlights an extremely concerning gap between when first concerns are raised and when something is actually done to help the child; often because of a limited awareness of the early markers of ASD by professionals on the front line. These facts underscore the necessity for clinicians of all disciplines to learn about and

CAUTION

Most parents of children who develop ASD express concerns regarding their child's development prior to the second birthday, well over a year before diagnostic evaluations take place, on average. Professionals need to be extra vigilant in not only validating concerns, but also in taking immediate action to assess and identify potential risk for ASD.

be vigilant for the early signs of ASD, so that children can be effectively evaluated and efficiently diagnosed. Only then can these children subsequently receive the critical early and intensive intervention that is associated with optimal outcome (National Research Council, 2001).

DIAGNOSTIC CRITERIA

Although the causes of ASD are likely neurobiological in nature, the spectrum of disorders still requires diagnosis based on behavioral symptomatology. The current diagnostic criteria put forth in the *Diagnostic and Statistical Manual, Fourth Edition, Text Revision* (DSM-IV-TR; APA, 2000) fall under the category of Pervasive Developmental Disorders (PDD), which includes Autistic Disorder, or autism; Asperger's Disorder, or Asperger syndrome, Rett's Disorder, Childhood Disintegrative Disorder (CDD), and Pervasive Developmental Disorder, Not Otherwise Specified (PDD-NOS). The behavioral features of all five PDDs fall within the following subcategories: (1) impairments in social interaction; (2) impairments in communication; and (3) restricted, repetitive, and stereotyped patterns of behavior, interests, and activities. A diagnosis of Autistic Disorder, the most prototypical of the PDDs, requires onset of delays or deviance in development prior to the age of three and a total of at least six impairments in all subcategories, with at least two falling within the area of social interaction.

Rapid Reference 1.1

Pervasive Developmental Disorders in the DSM-IV-TR

Autistic Disorder

Asperger's Disorder

Rett's Disorder

Childhood Disintegrative Disorder

Pervasive Developmental Disorder, Not Otherwise Specified

ASPERGER SYNDROME

The description of Asperger syndrome is more complicated. The *DSM-IV-TR* defines the disorder as having impairment in at least two areas of social interaction and one restricted, repetitive, and stereotyped pattern of behavior, but not meeting full criteria for Autistic Disorder. The criteria further stipulate that there can be no clinically significant delays in the development of language, cognition, and self-help adaptive skills *during the first three years of life* (APA, 2000). However, many clinicians overlook the text of the *DSM-IV-TR* and resort only to the charts. In this case, the text signifying "during the first three years of life" would be missed, resulting in misdiagnoses of older individuals with Asperger syndrome who most certainly do present with areas of deficit in cognitive, adaptive, and language abilities (e.g., Klin et al., 2007; Saulnier & Klin, 2007).

Furthermore, if one refers only to the charts and not the text, the description of circumscribed interests—the all-encompassing preoccupations with topics of interest that tend to be more specific to Asperger syndrome than the other PDDs—would be similarly overlooked. These criteria have generated a great deal of controversy, resulting in tremendous variation in the diagnosis of Asperger syndrome, both clinically and in research. This variability and lack of consistency in defining the disorder has ultimately resulted in removal of the subtype from the forthcoming *DSM-5* (APA, 2010), which is not without controversy (e.g., Wing, Gould, & Gillberg, 2011).

CAUTION

Clinicians are cautioned against merely relying on the *DSM-IV-TR* charts for determining diagnostic criteria for the PDDs, as the descriptions of the most differentiating features of Asperger syndrome are included within the text of the *DSM-IV-TR* but not within the charts.

Clinicians and researchers who have closely studied and worked with individuals with Asperger syndrome invariably view this subtype as qualitatively distinct from the other

CAUTION

Common misconceptions of Asperger Syndrome include the following characteristics interpreted in isolation:

- Individuals with ASD without cognitive impairment
- Individuals with ASD who have higher verbal than nonverbal IQ scores
- Individuals with ASD who have social intent and motivation to interact with others
- Asperger Syndrome is "mild autism"

PDDs, given the verbosity, social motivation, and fixation on topics of interest in these individuals. Ironically, these same symptoms can cause the most confusion in differential diagnosis. Common misconceptions propose those with Asperger's to be individuals without cognitive impairment; individuals with higher verbal than nonverbal IQ scores; individuals who have social intent; individuals who have mild or subtle social impairments; or individuals with perseverative interests, such as *Thomas the Tank Engine*—confusing getting "stuck" on a character or video rather than wanting to obsessively collect details about the topic of, for instance, trains. These misconceptions can have negative implications on outcome for individuals with Asperger syndrome because they are assumed to be less impaired and more able to navigate the world without supports—which is certainly not the case for many individuals.

It is not one of these behaviors, in isolation, that defines Asperger syndrome, but the overall profile of behavior, including developmental history. In early childhood, the social vulnerabilities of toddlers with Asperger syndrome are often masked by their relative strengths in other areas—such as their often precocious language; fixation on numbers and letters to the point of self-reading; and bourgeoning circumscribed interests. It is typically not until these children are immersed in social settings, where the social demands far outweigh their capacity to engage, that red flags are raised.

During the school-age years, individuals with Asperger syndrome tend to have more social motivation to interact with their peers, often inserting themselves into interactions inappropriately and/or lacking

CAUTION

Unlike autism, Asperger syndrome is often not detected in the first few years of life because in early childhood, the social vulnerabilities of toddlers with Asperger syndrome are often masked by their precocious language, affinity for numbers and letters, and regurgitation of facts on topics of interest. It is not until these children are immersed in social settings, such as preschool, that their true social impairments are recognized. For this reason, clinicians need to be extra vigilant in screening for social impairments in young children who have strong language and cognitive skills.

the appropriate social awareness to effectively navigate an interaction. Yet, they can have just enough awareness to understand the failed nature of their attempts, placing them at great risk for anxiety, depression, and isolation. In autism, individuals tend to be more socially passive; they certainly may respond to direct interaction, often even appropriately, but they are less likely to initiate interactions with their peers. Furthermore, self-awareness in autism can be more impaired, acting as a buffer in that individuals might not be as cognizant of their failed social experiences. Nevertheless, as stressed previously, social motivation should not be interpreted in isolation when distinguishing Asperger syndrome from other PDDs.

DON'T FORGET

Individuals with ASD, particularly those with Asperger Syndrome who tend to have a modicum of social awareness, are at great risk for mood disorders such as anxiety and depression. These symptoms can emerge as early as school age, but are most prominent in adolescents and adults and, therefore, should be monitored and treated accordingly.

PERVASIVE DEVELOPMENTAL DISORDER, NOT OTHERWISE SPECIFIED

A diagnosis of PDD-NOS requires impairment in reciprocal social interaction (i.e., symptoms in subcategory 1) with associated

Rapid Reference 1.2

Distinctions Between Asperger Syndrome and Other PDDs

Asperger Syndrome	Autism, PDD-NOS
• Early history marked by intact or precocious speech development	• Early history marked by significant language delays/impairments
• Extreme verbosity and one-sided conversations	• Limited speech and/or stereotyped language (e.g., echolalia, scripting)
• Social motivation in the absence of ability to effectively navigate social interactions	• Social passivity—more apt to monitor peers rather than initiate interaction
• May have stronger rote verbal than nonverbal cognitive scores—*though not diagnostic!*	• Tend to have stronger rote nonverbal than verbal cognitive scores
• Circumscribed interests—all-absorbing interest on a topic, including collecting facts on the topic, and this interest pervades and dominates conversations	• Perseverative interests—fixations on objects/movies/activities that become overly repetitive, and the individual has difficulty disengaging from the interest

impairments in at least one of the remaining two subcategories. Therefore, under the current taxonomy, an individual does not necessarily have to present with stereotypical behaviors (i.e., symptoms falling under subcategory 3) to carry a diagnosis of PDD-NOS. The proposed diagnostic criteria for a *DSM-5* diagnosis of ASD, however, require *at least two* stereotyped behaviors (see Table 1.1). This will most certainly impact many individuals who currently hold the label of PDD-NOS, as it raises the question as to what label, if any, will be appropriate to merit the same degree of services for these individuals.

Table 1.1. Comparison Between *DSM-IV* and Proposed *DSM-5* Diagnostic Criteria for Autism Spectrum Disorders

	DSM-IV	DSM-5
Category	*Pervasive Developmental Disorders*	*Autism Spectrum Disorder*
Category Subtypes	1. Autistic Disorder 2. Asperger's Disorder 3. Pervasive Developmental Disorder, Not Otherwise Specified (PDD-NOS) 4. Rett's Disorder 5. Childhood Disintegrative Disorder	None
Symptom Subcategories	1. Impairments in Social Interaction 2. Impairments in Communication 3. Restricted, Repetitive, and Stereotyped Patterns of Behavior, Interests, and Activities	1. Deficits in Social Communication and Social Interaction 2. Restricted, Repetitive Patterns of Behavior, Interests, or Activities
Diagnostic Criteria	1. *Autistic Disorder* = at least six total symptoms across all three subcategories, at least two of which are in social interaction 2. *Asperger's Disorder* = symptoms in social interaction and restricted behaviors, with no delays in the development of language, cognition, or adaptive self-help skills in first three years of life; but not to full criteria for Autistic Disorder 3. *PDD-NOS* = social impairments *and* symptoms in either communication and/or restricted behaviors; but not to full criteria for Autistic Disorder	1. *ASD* = three required criteria in social communication and social interaction *and* at least two out of four restricted and repetitive patterns of behavior 2. Symptoms must be present in early childhood (even if not fully manifested until social demands exceed the child's level of social functioning)

DON'T FORGET

Rett's syndrome is differentiated by ASD in that it is more prevalent in girls than boys; it is associated with a mutation in the MECP2 gene; there is early regression of psychomotor development in the first year of life; and there is a deceleration of head circumference.

RETT'S DISORDER AND CHILDHOOD DISINTEGRATIVE DISORDER

Rett's Disorder and CDD are rare, regressive-type disorders where at the outset of the respective regressions in development, the individual's behavioral presentation is similar to autism. In Rett's Disorder, pre- and perinatal development are apparently normal, followed by a regression in psychomotor development and social engagement between the ages of 5 and 48 months. There is also a deceleration of head circumference and progression of hand washing/hand wringing mannerisms. What differentiates Rett's from all other PDDs is that Rett's, to date, is predominantly prevalent in females, whereas the remaining PDDs are, overall, four to five times more prevalent in males. A genetic mutation on the MECP2 gene has also been identified in the majority of Rett's cases (Van Acker, Loncola, & Van Acker, 2005).

In CDD, early development is spared for the first two to three years, after which there is a clinically significant loss of previously acquired skills in at least two of the following areas: receptive or expressive language, social skills, adaptive skills, toileting skills, play skills, or motor development. The regression must take place before age 10, but in most cases, the regression occurs between ages 2 and 3 (Volkmar, Koenig, & State, 2005). At the outset of the regression, individuals with CDD often are afflicted with severe or profound intellectual disability in addition to the autism symptomatology. Very little is known about the etiology of CDD or the triggers of the regression, although research suggests that triggers can be associated with (but not caused by) psychosocial stressors, such as those that are common to preschool-age children. These could include birth of a sibling, death of a family member, or a significant hospitalization

(Volkmar, Koenig, & State, 2005). CDD should not be confused with regressive autism, which occurs in about 10% of cases of ASD and where there is a reported loss of or plateau in development of skills prior to the age of 2. Children with regressive autism do not appear to be as impaired as children with CDD at the outset of their regression; yet, just as little is known about regressive autism and how it is differentiated from autism without regression. To reiterate, ASD is a neurodevelopmental disorder and, as such, the symptoms unfold over the first few years of life. Thus, deviance in the developmental course of social communication and behavioral skills around 18 to 24 months of age is anticipated in ASD but can often be misconstrued as regression.

DON'T FORGET

Childhood Disintegrative Disorder is distinct from regressive autism in that the regression of skills in CDD occurs after the age of 2, and substantial delays are evident at the outset of the regression in CDD in many areas of development, including language, social functioning, self-help skills, motor skills, and play skills.

DSM-5

Recent nomenclature has moved in the direction of considering the PDDs a spectrum of disorders; that is, taking more of a dimensional rather than categorical approach to diagnostic conceptualization. Thus, more common terminology refers to Autism Spectrum Disorders (ASD), typically signifying Autistic Disorder, Asperger syndrome, and PDD-NOS (given the rarity and relatively limited public awareness of Rett's

CAUTION

Autism is a neurodevelopmental disorder where symptoms (i.e., deviance in behavioral development) tend to manifest in the second year of life. This unfolding of symptomatology can be misinterpreted as a regression in the development of skills.

DON'T FORGET

Despite the vast heterogeneity observed between individuals across the autism spectrum, the common thread among the five Pervasive Developmental Disorders is that they are all *social disabilities*.

Disorder and CDD). The *DSM-5*, slated to be published in 2013, is proposing to change the diagnostic category to *Autism Spectrum Disorder*, eliminating the subtypes altogether (see Table 1.1; APA, 2010). Given these forthcoming changes, this book will focus on the broad spectrum of autism rather than on specific subtypes. Nevertheless, the authors will highlight when specific features that are more relevant to one subtype than another merit analysis (e.g., in Asperger syndrome).

No two individuals under the umbrella of the autism spectrum have identical presentations. There is more heterogeneity than similarity of symptom expression, which has resulted in a host of theories as to what the causes might be. Nonetheless, the common thread of all five PDDs, or ASDs, is the resulting social disability and the limited capacity to independently navigate the social world, whether expressed as substantial deficits in rudimentary social skills, more subtle vulnerabilities in interpreting the nuances of social interactions, or any variation in between. These social impairments are qualitatively different from and more severe than the vulnerabilities in social development that can be observed in other developmental disorders.

With the following book, we present a model process for identifying the symptoms of ASD while accurately differentiating the nature of social disabilities from mere delays in social development. As such, the focus is more on the differentials between ASD and other neurodevelopmental disorders rather than on distinguishing among the ASD subtypes, especially given the aforementioned *DSM-5* changes. Having obtained extensive experience in multidisciplinary diagnostic evaluations, we adopt the comprehensive developmental approach to assessment, diagnosis, interpretation, and report writing. Beginning with Chapter 2, the necessity for obtaining a baseline of cognitive and developmental functioning is discussed. Because the essence of social

interaction is communication, Chapter 3 then outlines how the speech, language, and communication assessment informs the diagnostic process. This naturally transitions to Chapter 4, where aberrant or problematic behaviors are often the result of impaired communicative functioning, thus highlighting the need to functionally assess these behaviors and then replace them with more adaptive means of communicating.

Chapters 5 and 6 collectively outline the diagnostic assessment, which entails the gathering of historic information, observing the individual in natural contexts, and directly assessing behaviors through interaction and play. Chapter 7 focuses on the common differentials and comorbidities that arise through referrals for diagnostic evaluations in ASD throughout the life span. Finally, Chapter 8 ties the process together with two samples of integrated reports from model comprehensive diagnostic evaluations—one of a toddler and one of a school-aged child. Our hope is that this model will be useful in informing both burgeoning clinicians just starting out in the field, as well as seasoned professionals who are experiencing an increased exposure to ASDs and, subsequently, are seeking knowledge of how to effectively identify, diagnose, and/or refer patients at risk.

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