One

OVERVIEW

DIAGNOSTIC CRITERIA

Autism spectrum disorder (ASD) is among the most common childhood disorders, with prevalence rates currently estimating 1 in 36 school-aged children (Maenner, Warren, Williams et al., 2023). ASD is four times more prevalent in males than females, but as will be highlighted later in this book, the diagnostic presentation of females assigned at birth is complex and, consequently, could suggest an underrepresentation of true female prevalence rates. Although the cause(s) of ASD is likely neurobiological/genetic in nature, the exact etiology remains unknown. As such, the diagnosis of ASD has historically been determined based on behavioral symptomatology that, through the lens of a medical model, are generally described as delays, deficits, impairments, or similar terminology that implies the need for remediation or a "cure" (Pellicano & den Houting, 2022). As the spectrum has broadened over the 21st century to include an increasing number of individuals with intact language and cognition, many autistic individuals identify as being neurodiverse or neurodivergent and do not seek to be cured or viewed as disordered, but rather wish to be accepted and supported for their unique differences (den Houting, 2019). Nevertheless, the label still implies that the individual is experiencing challenges, vulnerabilities, and in some cases delays/deficits that negatively affect multiple areas in life. The authors of this book have tremendous respect for neurodiversity, as we do for every individual across the extremely diverse and complex spectrum, including those more severely affected. Thus, in our aim to help educate and train clinicians on how to effectively identify ASD based on behavioral "symptoms," we strive to focus on the diagnostic label serving to enhance the quality of life and self-determination rather than to imply curing or fixing anybody. We will use terms

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such as symptoms, disorder, condition, and disability throughout the text given that this terminology is still part of the diagnostic criteria and medical nomenclature to which clinicians are exposed. We will also use identity-first language (autistic individual) opposed to person-first language (individual with autism) to respect that not all individuals view ASD as something separate from who they are.

The current diagnostic criteria for ASD put forth in the *Diagnostic and Statistical Manual, Fifth Edition, Text Revision* (DSM-5-TR; APA, 2022) require persistent symptoms in two broad areas: (1) *the presence of delays/deficits/challenges in social communication and interaction* in all three of the following areas either currently or by developmental history: (a) social-emotional reciprocity (e.g., shared interests, shared affect, and conversations), (b) nonverbal communication (e.g., integrated eye contact, and directed facial expressions, (c) use and understanding of gestures and body posture), and (d) in developing, maintaining, and understanding relationships (e.g., interactive imaginative play, interest in peers, and making friends); and (2) *the presence of restricted, repetitive, or stereotyped behaviors* that include at least two of the following four areas: (a) stereotypical motor movements or use of objects (e.g., hand flapping, spinning, lining up toys, and repetitive or stereotyped speech), (b) insistence on sameness or routine (e.g., difficulty with transitions, changes in routine, and rigidity in thinking), (c) highly restricted interests that become overly intense or all-consuming (e.g., intense interests on topics and fixation on objects), and (d) sensory processing impairments (e.g., hyper- or hypo-reactivity to sensory input, or seeking out sensory input). This combination of social vulnerabilities and stereotyped behaviors must be present in early development (even if symptoms go undetected until adolescence or adulthood) and clinically impairing across multiple contexts in life.

Rapid Reference 1.1

Social communication and interaction criteria (must include all three either currently or by history):

1. *Challenges in social-emotional reciprocity*
2. *Challenges in nonverbal communication*
3. *Challenges in developing, maintaining, and understanding relationships*

Restricted, repetitive, and stereotyped behaviors (must include at least two either currently or by history)

1. *Stereotypical motor movements or use of objects*
2. *Insistence on sameness or routine*
3. *Highly restricted interests*
4. *Sensory processing impairments*

In addition to listing the diagnostic criteria for the two categories of social communication/interaction symptoms and restricted, repetitive, and stereotyped behaviors, the DSM-5 has three severity levels that clinicians need to apply to both categories. Level 1 is specified for individuals who require support at the lowest level (e.g., symptoms interfere with functioning in one or more contexts, but they are mild); Level 2 is specified for those who require substantial support (e.g., there are marked impairments that interfere across multiple contexts); and Level 3 is specified for those who require very substantial support (e.g., delays or deficits are severe and entail comprehensive supports across contexts). Clinicians can specify different levels based on each category. For instance, an individual with intact language and cognition may only require Level 1 support for their social communication and interaction skills, but same individual's stereotypical behaviors might be so intrusive to life that Level 2 is required for this category.

DON'T FORGET

The DSM-5 has three levels of severity that need to be applied to each category of symptoms (social communication/interaction and restricted, repetitive, and stereotyped behaviors) as follows:

- *Level 1*: the individual requires support
- *Level 2*: the individual requires substantial support
- *Level 3*: the individual requires very substantial support

The DSM-5 also offers clinical specifiers for ASD that allow for descriptions of common co-occurring conditions. These include specifying whether ASD is accompanied by co-occurring intellectual impairment or language impairment (e.g., nonverbal, minimally verbal, phrase speech, and sentence speech), whether ASD is associated with a known genetic or other medical condition (e.g., Fragile X syndrome and Down syndrome), environmental factor, other neurodevelopmental (e.g., ADHD), mental (e.g., anxiety and depression), or behavioral problem, or whether co-occurring catatonia is present. Catatonic behaviors can include posturing, limited response to external stimuli, stupor, or mutism. Catatonia can be present in as many as 10% of autistic individuals, with features

DON'T FORGET

The DSM-5 has clinical specifiers that allow for indicating all co-occurring conditions. This helps inform levels of support needed as well as types of interventions.

- With or without accompanying intellectual impairment
- With or without accompanying language impairment
- Associated with a known genetic or medical condition, or environmental factor
- Associated with another neurodevelopmental, mental, or behavioral problem
- With catatonia

generally developing in late adolescence (Vaquerizo-Serrano, Salazar De Pablo, Singh, & Santosh, 2021).

There is vast heterogeneity in symptom expression across the spectrum and throughout development, with some individuals experiencing only mild challenges in certain areas and other individuals experiencing quite debilitating challenges. Symptoms that impact autistic individuals are not just limited to the criteria for ASD, but also include varying levels of speech and language, cognition, emotional and behavioral regulation, and attention and executive functioning. Thus, clinicians need to be knowledgeable of these broader areas. When autism was first defined in the 1900s, the vast majority of individuals had co-occurring language and cognitive deficits that significantly affected development. However, current epidemiological data show that only about 38% of autistic children in the United States have co-occurring intellectual disability (ID; Maenner et al., 2023). This shift in levels of cognitive and language functioning has sparked a great deal of controversy in the field. Where historically there used to be a clear delineation of impairment to meet diagnostic criteria; currently, the diagnostic line between what is considered neurotypical and neurodiverse is quite blurred (Rodgaard, Jensen, Vergnes, Soulieres, & Mottron, 2019). This raises concerns about whether autism is a single condition or perhaps multiple disabilities, and whether the spectrum has become too broad to be accurately defined. Subtypes of autism were developed in the DSM-IV and DSM-IV-TR (APA, 1994, 2000) with autistic disorder, Asperger's disorder, pervasive developmental disorder, not otherwise specified, Rett's disorder, and childhood disintegrative disorder. However, research failed to show that these subtypes indeed had distinct phenotypes. Rather, research indicated that the main factor differentiating subtypes was the geographical site or clinicians providing the diagnoses (Lord, Petkova, Hus et al., 2012). Even expert clinical teams using gold-standard measures were not reliable in determining the categorical subtypes, of autism which led to the transition to the more dimensional definition of ASD.

Certainly, individuals with significant cognitive and language impairments and affiliated behavioral challenges have drastically different needs than individuals with intact cognition and language. In 2021, the Lancet Commission published a report that formally recognized the term "profound autism" to distinguish autistic individuals who are so severely affected that they will likely require 24-hour support throughout their lives (Lord, Charman, Havdahl et al., 2022). In contrast, individuals with intact language and cognition can often function quite independently across aspects of life. Many of these individuals identify as being neurodiverse and do not view themselves as having a disability but

DON'T FORGET

There is significant variability in symptom expression throughout the spectrum in autism symptomatology, speech and language skills, cognition, emotional and behavioral regulation, and attention and executive functioning. Thus, clinicians need to be knowledgeable of these broad areas.

rather advocate for acceptance of neurodivergence. Yet, it is important to highlight that even some higher-cognitive autistic individuals struggle tremendously in life despite their strong intellectual capacity. For this reason, the historically misused phrase "high functioning autism" has resulted in an underestimate of services and supports that autistic individuals require because of the erroneous assumption that intact language and cognition equate to functional independence (Alvares, Bebbington, Cleary et al., 2019). Similarly, the phrase "low functioning autism" misrepresents individuals with cognitive and language impairments who may be functioning quite well despite their delays. Clinicians are therefore advised to keep terminology descriptive of specific skill areas, and use the severity levels and clinical specifiers to advocate for appropriate supports.

CAUTION

Avoid using terminology such as "high functioning or low functioning autism," as these labels make erroneous assumptions about an individual's true profile of strengths and/or vulnerabilities. Preferred language describes specific profiles:

- Autism without cognitive impairment
- Autism without language impairment
- Minimally verbal or nonverbal
- Profound autism

ASD has a complex genetic etiology with symptoms unfolding over the first few years of life. Infants who will go on to develop ASD are not born exhibiting full autism symptom expression; rather, as symptoms emerge, developmental milestones are derailed resulting in a diverging trajectory away from neurotypical development. Thus, some infants appear to be developing within age expectations in the first few months or even the first year of life, but they then fail to keep pace with chronological age expectations during the second year. As developmental divergence occurs, typically developing milestones can fade, while the presence of atypical behaviors can emerge. It is therefore essential that clinicians be knowledgeable of normative development in addition to merely observing for the presence of ASD symptoms because it is often the case that infants at risk for ASD have words, eye contact, gestures, etc., but the limited frequency and range of these skills are what causes concern.

Research indicates that 80–90% 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 over the age of four despite these concerns (Maenner et al., 2023; Chawarska et al., 2007). When experienced clinicians make a diagnosis of ASD at 18 to 24 months, the stability of diagnosis is

CAUTION

Clinicians involved in early detection of ASD need to be knowledgeable of normative developmental milestones in addition to ASD symptoms because as ASD unfolds in infancy and toddlerhood, it is often the limited frequency and range of expected skills that causes concern rather than the emergence of atypical behaviors.

CAUTION

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

CAUTION

Social motivation and reciprocity should not be interpreted in isolation when distinguishing autism from non-autism conditions.

DON'T FORGET

Autistic individuals with intact language and cognition may have awareness of their challenges and therefore are at great risk for mood disorders such as anxiety and depression. These symptoms can emerge at any point in development and should be monitored and treated accordingly.

quite strong, also around 80–90% (Chawarska et al., 2009). This highlights the extremely concerning gap between when first concerns are raised and when something is actually done to help the child, often because of limited awareness of the early markers of ASD by professionals on the front line as well as barriers to access to services. These facts underscore the necessity for clinicians of all disciplines to learn about and be vigilant of the early signs of ASD so that children can be effectively evaluated and efficiently diagnosed. These include (but are not limited to) delays or deficits in joint attention, shared affect, social smiling, response to name, frequency and range of gestures, communicative intent, and functional speech.

In autistic toddlers and preschoolers who have intact cognition and language, social vulnerabilities can be masked by strengths in other areas, such as 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 outweigh the capacity to engage, that social concerns are raised. As autistic children with intact language and cognition move through the school-age years, they can be motivated to interact with their peers, but they might insert themselves into interactions inappropriately and/or lack the appropriate social awareness to effectively navigate interactions. Yet, they can have just enough awareness to understand the failed nature of their attempts, placing them at great risk for experiencing symptoms of anxiety, depression, and isolation. This contrasts with more profound forms of autism, where individuals can be socially passive, withdrawn, or less likely to initiate interactions with others. Nonetheless, social motivation and reciprocity should not be interpreted in isolation when distinguishing autism from non-autism conditions.

Based on our collective and extensive experience in conducting multidisciplinary diagnostic evaluations, we present a comprehensive developmental approach to the assessment and diagnosis of ASD, including how to assess for the common differential and co-occurring condition throughout the lifespan, how to interpret assessment results and convey the findings in a comprehensive written report, and how to use individual profiles of strengths and vulnerabilities to inform educational and intervention programming. Beginning with Chapter 2, we discuss the necessity for obtaining a baseline of developmental and cognitive functioning to identify individual profiles of strengths and vulnerabilities. Chapter 3 then outlines how the speech, language, and communication assessment informs the diagnostic process. Given the significance of adaptive behavior to functional outcome in ASD, Chapter 4 describes how assessing for adaptive behavior, executive functioning, and social-emotional skills helps inform both diagnosis and intervention.

Chapters 5 and 6 collectively outline the assessment of autism symptomatology, 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 co-occurring conditions that arise through referrals for diagnostic evaluations in ASD throughout the lifespan. As the spectrum continues to broaden, Chapter 8 focuses on the many challenges in correctly identifying ASD when symptom expression is obscured by racial/ethnic/cultural disparities, gender and sexual identity, and other complexities that can result in misdiagnoses or missed diagnoses. Chapter 9 covers the extremely important transitional period from school-age to adulthood, where evaluations are often necessary to inform postsecondary education, college, vocations, and adult life. Chapter 10 ties the process together with three samples of integrated reports from model comprehensive diagnostic evaluations—one of a toddler, one of a school-aged child, and one of a female adult receiving an ASD diagnosis for the first time. Our hope is that this model will be useful in informing both burgeoning clinicians just starting out in the field and 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 for evaluations.

REFERENCES

Alvares, G. A., Bebbington, K., Cleary, D., Evans, K., Glasson, E. J., Maybery, M. T., Pillar, S., Uljarević, M., Varcin, K., Wray, J., & Whitehouse, A. J. (2019). The misnomer of 'high functioning autism': Intelligence is an imprecise predictor of functional abilities at diagnosis. *Autism*, *24*(1), 221–232.

American Psychiatric Association. (1994). *Diagnostic and Statistical Manual of Mental Disorders* (4th ed.). Washington, DC: Author.

American Psychiatric Association. (2000). *Diagnostic and Statistical Manual of Mental Disorders* (4th ed., text rev.). Washington, DC: Author.

American Psychiatric Association. (2022). *Diagnostic and Statistical Manual of Mental Disorders* (5th ed., text rev.). Washington, DC: Author.

Chawarska, K., Klin, A., Paul, R., Macari, S., & Volkmar, F. (2009). A prospective study of toddlers with ASD: Short-term diagnostic and cognitive outcomes. *Journal of Child Psychology and Psychiatry*, *50*(10), 1235–1245.

Chawarska, K., Paul, R., Klin, A., Hannigen, S., Dichtel, L. E., & Volkmar, F. (2007). Parental recognition of developmental problems in toddlers with autism spectrum disorders. *Journal of Autism and Developmental Disorders*, *37*(1), 62–73.

den Houting, J. (2019). Neurodiversity: An insider's perspective. *Autism*, *23*(2), 271–273.

Lord, C., Charman, T., Havdahl, A., et al. (2022). The Lancet Commission on the future of care and clinical research in autism. *Lancet*, *399*(10321), 271–334.

Lord, C., Petkova, E., Hus, V., et al. (2012). A multisite study of the clinical diagnosis of different autism spectrum disorders. *Archives of General Psychiatry*, *69*(3):306–313.

Maenner, M. J., Warren, Z., Williams, A. R., et al. (2023). Prevalence and characteristics of autism spectrum disorder among children aged 8 years: Autism and developmental disabilities monitoring network, 11 sites, United States, 2020. *MMWR Surveillance Summaries*, *72*(2):1–14.

Pellicano, E., & den Houting, J. (2022). Annual research review: Shifting from 'normal science' to neurodiversity in autism science. *Journal of Child Psychology and Psychiatry*, *63*(4), 381–396.

Rodgaard, E., Jensen, K., Vergnes, J., Soulieres, I., & Mottron, L. (2019). Temporal changes in effect sizes of studies comparing individuals with and without autism. A meta-analysis. *JAMA*, *76*(11):1124–1132.

Vaquerizo-Serrano J., Salazar De Pablo G., Singh J., & Santosh P.2021). Catatonia in autism spectrum disorders: A systematic review and meta-analysis. *Eur Psychiatry*, *65*(1):e4.