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Autism Spectrum Disorders

Autism Spectrum Disorder (ASD) is a developmental disorder characterized by impairment in social interaction, communication skills and behavior. It is one of the most common neurodevelopmental disorders, with the most recent estimates indicating that 1 of every 68 children in the United States may meet criteria for diagnosis of an ASD. Boys are more commonly affected than girls (5:1), and there is evidence that White children are diagnosed more frequently than Black or Latino children.¹

Prevalence estimates have been growing over the past two decades, likely due, at least in part, to changes in the diagnostic criteria for ASD over that same time period. The diagnostic criteria for ASD have changed again with the most recent revision of the Diagnostic and Statistical Manual-5, in 2013 and it is important for families, stakeholders and providers to understand why these changes were made, and the potential impact on diagnosis and access to services. Though the behaviors associated with autism had been described nearly 40 years prior, it was not recognized as a clinical diagnosis until the publication of DSM-III in 1980. Over time, additional Autism Spectrum Disorders, or as they were previously called Pervasive Developmental Disorders, were broken into distinct diagnoses (Autism, Asperger Syndrome and Pervasive Developmental Disorder NOS). Unfortunately, use of these distinct categories proved to be problematic. The rates at which each of the categories within the PDD umbrella were diagnosed varied across different demographic features like race or region of the country where diagnosis was made.¹ The rate at which different categories were diagnosed varied over time, with different diagnoses seeming to rise and fall in popularity over time², and also varied by the location and experience of the provider making the diagnosis³. Diagnoses also showed poor stability, with up to 25% of children diagnosed with an Autism Spectrum Disorder carrying at least two distinct PDD diagnoses by the time they turned 8⁴. With diagnosis representing the means by which individuals access care and services, the variability in use of diagnostic labels presented a barrier to care. With these concerns in mind, the DSM-5 workgroup sought to update the diagnostic criteria based on the existing empirical evidence, to provide more sensitive and specific diagnosis but to also allow for diagnosis across a wide range of intellectual ability, and also provide a way to allow for changes in presentation due to development across the lifespan. Criteria were designed to provide a cohesive behavioral presentation, while recognizing that there may be a wide range of causes or origins, and to allow diagnosis of comorbid disorders.

The changes in DSM-5 criteria for ASD include:⁵

- Creating one single continuous diagnostic category rather than distinct categories (Autism Spectrum Disorder vs. PDD, Asperger Syndrome and Autism).
- Use of two symptom categories, Socialization/Communication impairment and Repetitive Behaviors/Restricted/Repetitive Patterns of Behavior rather than 3 categories which had proven to be difficult to separate from one another. The first category represents a joining or link of previously separated social and communication deficits.

- Inclusion of symptoms of sensory processing differences, a domain reported by many individuals with ASD, but which had not been previously included as a diagnostic feature.
- Inclusion of more specific, concrete descriptions of symptoms to promote greater accuracy in diagnosis.
- Age of onset criteria have been extended from early childhood to later in development when "social demands exceed limited capacities" to reflect the fact that many individuals may not experience more significant concerns until school age or later.⁶
- The introduction of severity specifiers (i.e. Level 1: "Requiring Support," Level 2: "Requiring Substantial Support," and Level 3: "Requiring Very Substantial Support") to describe symptoms of Social/Communication impairment and Restricted/Repetitive Patterns of behavior. These specifiers are provided to assist in linking individuals on the Autism Spectrum to specific supports in education, the community, etc. The specific behaviors included in the diagnostic criteria are anchored to severity range by language of diagnostic features (e.g. "ranging, for example, from difficulties adjusting behavior to suit various social contexts; to difficulties in sharing imaginative play or in making friends; to absence of interest in peers")
- For the first time, the diagnostic criteria allows for diagnosis of a full range of co-morbid difficulties and associated features. For example, in previous iterations of the DSM, ADHD was specifically listed as a rule out diagnosis for ASD, and could not be diagnosed at the same time, despite clear evidence that this is a common co-occurring area of impairment.⁷

DSM-5 also introduces a new disorder: Pragmatic/Social Communication Disorder to describe individuals who have impairment in socialization and social communication in the absence of repetitive behaviors and restricted interests. Research suggests that this diagnostic presentation may be distinct from ASD, with different neurological underpinnings.^{8,9}

Implications for changes in diagnostic criteria

Early studies evaluating the impact of changes in the diagnostic criteria for ASD indicate that most individuals who received a diagnosis of one of the autism spectrum disorders under DSM-IV criteria would retain an ASD diagnosis using DSM-5 criteria, with estimates ranging from 75-90% retaining their diagnosis.^{10,11,12} Children with more severe symptoms, lower IQ and more severe language impairment were more likely to retain their ASD label. PDD NOS and Asperger Syndrome had proven to be less stable diagnostic categories in previous research, given that individuals with these diagnoses present with fewer symptoms that are more likely to diminish with intervention and with development.¹³ For individuals in these groups, most will retain an ASD designation and a large proportion of those who do not meet criteria for continued classification under the diagnostic umbrella of ASD will meet criteria for diagnosis of Social Pragmatic Language disorder. Furthermore, a child's ASD diagnostic status appears to remain stable across development, though structural language impairment or cognitive performance may improve in response to intervention and time.¹⁴ Nonetheless, analyses do indicate that a proportion of individuals who were diagnosed under DSM-IV criteria will not qualify for diagnosis under DSM-5, leaving them at risk for loss of access to educational and other services.¹⁵ In addition to the potential to lose access to services, members of the Autistic Community are also concerned that the change to a new diagnostic classification, and loss of Asperger Syndrome as a diagnostic category also represents a loss of a cultural identity that is central to many individuals.¹⁷ Additional concerns include lack of sensitivity in the current diagnostic criteria for ways in which ASD may display differently in girls/women compared to boys/men.¹⁶ Like many behaviorally-defined conditions, however, concerns remain that ASD is likely due to many different underlying causes, and presents with many subtle important differences. Treating it as a single disorder may therefore compromise research and may make delivery of appropriate care very challenging. Continued emphasis on developing a better understanding of the genetics and bio-medical markers of ASD is recommended by many in the medical and research communities.¹⁸

Screening Guidelines

Given the high prevalence of ASD in the population, and the evidence that early intervention has significant benefits for socialization and communication skills, the American Academy of Pediatrics has established a set of guidelines for early screening and diagnosis of Autism Spectrum Disorders. The algorithm provides a special emphasis on the role of the primary care pediatrician as these are the people who parents are most likely to first express concerns to, and who are uniquely situated to monitor development in sequence in the first years of life. Primary Care Physicians (PCPs) are urged to act with the acronym ALARM in mind (Autism is prevalent, Listen to parents, Act early, Refer, and Monitor). More specific guidelines include:¹⁹

- **Level 1:**
 - Monitor developmental milestones in routine well visits and provide additional visits for closer watch of children with additional risk factors. Score 1 for each of the following factors and move to next steps for children who score 2 or higher:
 - Sibling with ASD
 - Parental concern
 - Other caregiver concern (e.g., teacher, daycare provider, relative)
 - Pediatrician concern
 - Perform routine ASD screening using an empirically supported tool like the Modified Checklist for Autism in Toddlers-Revised (M-CHAT-R) (<https://www.m-chat.org/>) for children with increased risk factors, and again at 18 and 24 month visits regardless of other risk factors.
 - Red flags for immediate referral include:
 - No babbling or pointing or other gesture by 12 months
 - No single words by 16 months
 - No 2-word spontaneous phrases by 24 months
 - Loss of language or social skills at any age
 - If screening measures are positive or concerning, provide parental education and refer immediately for additional evaluations, and schedule a follow up visit to continue to monitor development and outcome of referrals.
 - Audiology evaluation
 - Early Intervention/Early Childhood Education evaluation
 - Evaluation through specialized autism centers
- **Level 2 (after referral from PCP):**
 - Multidisciplinary evaluations are considered the gold-standard for ASD diagnosis.
 - Early Intervention/Birth to 3/County Early Education Evaluation
 - Specialty multidisciplinary autism evaluation
 - Autism assessment with measures like Autism Diagnostic Observation Schedule (ADOS) to be administered by a trained professional.
 - Medical evaluation including assessment of sleep, neurological status and genetic and metabolic screening
 - Speech/Language Evaluation
 - Neuropsychological/Psychoeducational Evaluation for cognitive ability, attention and educational concerns
 - Behavioral evaluation for concerns with anxiety; repetitive, self-injurious or other interfering behaviors

There are a number of steps parents and caregivers can take if they have concerns about their child's development. The first step is often discussing these concerns with the child's pediatrician who can help to direct the family to local resources and assist in coordinating care. Families can also seek evaluation for autism or other developmental concerns at no cost through their local Birth to Three, Early Intervention (preschool aged) or Child Find (school aged) program, and do not need to be referred by their physician to start this process. For children who are diagnosed with a qualifying developmental delay, these programs will also provide intervention at no cost to the family. Depending on the specific needs of the child, families may also pursue additional speech/language, occupational, or behavioral therapies through community providers and may also consult with developmental pediatricians or other medical specialists who can assist in addressing other medical concerns. The website firstsigns.org is an excellent resource for families and caregivers, providing more specific information about typical development as well as examples of developmental differences, links to local resources, and guidance about what steps to take for further evaluation.

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Additional resources:

<http://www.cdc.gov/ncbddd/actearly/index.html>

<http://www.aafp.org/afp/2008/1201/p1301.html>

<http://pediatrics.aappublications.org/content/120/5/1183.full>

ABOUT THE TECHNICAL ASSISTANCE NETWORK FOR CHILDREN'S BEHAVIORAL HEALTH

The [Technical Assistance Network for Children's Behavioral Health](#) (TA Network), funded by the Substance Abuse and Mental Health Services Administration, Child, Adolescent and Family Branch, partners with states and communities to develop the most effective and sustainable systems of care possible for the benefit of children and youth with behavioral health needs and their families. We provide technical assistance and support across the nation to state and local agencies, including youth and family leadership and organizations.

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