

## **Autism Spectrum Disorder: An Overview**

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Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder characterized by pervasive deficits in social communication and interaction, alongside patterns of restricted, repetitive, stereotyped behaviors and interests (American Psychiatric Association, 2013). The causes, as well as neurobiological and genetic bases for ASD is still unknown, yet there is developing research in these areas. Although diagnosis does not necessarily need to be made in early childhood, it is important to establish a developmental history consistent with such symptoms, in order to distinguish such deficits from those associated with trauma or other adverse impacts later in life. Beyond the main diagnostic criteria, there is significant diversity in the symptom presentations that are demonstrated by people with ASD, including overall symptom severity, levels of receptive and expressive language skills, intellectual ability, and related deficits (Evans et al, 2018). ASD may occur with or without either intellectual impairment, and/or functional language deficits. ASD also commonly co-occurs with other psychiatric diagnoses, including anxiety and depression. Further, ASD can also present with challenging behaviors such as aggression and self-injury, especially in those with intellectual impairment and language deficits. It has been well established that ASD is diagnosed more often in males than in females, with recent estimates suggesting that ASD is 4.3 times as prevalent among boys as among girls (Maenner et al., 2016). Despite the fact that this is well known, there is considerable uncertainty about the nature of this sex/gender discrepancy, although it has become widely accepted that males and females with ASD present differently.

In terms of assessment for ASD, there are a plethora of screening instruments and diagnostic tools available. The primary goal of ASD screening tools is to facilitate the early identification and treatment for ASD. The Modified Checklist for Autism in Toddlers (M-CHAT) is the most widely used screening instrument for ASD in young children (Robins et al, 2001; Ibanez et al, 2014). The M-CHAT has been documented as high in sensitivity, although lower in specificity and positive predictive value (PPV) (Oien et al, 2018). The M-CHAT is more useful in predicting ASD in children at risk for ASD (Robins et al, 2001; Kleinman et al, 2014; Robins et al, 2014), with a definitive lack of adequate follow-up studies on the children who screen negative for ASD (McPheeters, 2016). With regard to the lack of follow-up, only one published study to date (Oien, 2018) has looked at the developmental and temperamental characteristics of children who screen negative on the M-CHAT but later receive an ASD diagnosis, and the findings are preliminary, at best.

Beyond preliminary screening, it is also important to examine differences in diagnostic instrumentation for ASD. The most commonly relied upon diagnostic instrument for ASD (what is considered to be the gold-standard in ASD assessment) is the Autism Diagnostic Observation Schedule- Second Edition (ADOS-2) (Lord et al, 2000; 2012). The ADOS-2 is a semi-structured observational assessment designed to evaluate all aspects of ASD diagnosis, including communication, social interaction, and stereotyped behaviors and restricted interests (Lord et al, 2000; 2012). Additionally, the Autism Diagnostic Interview - Revised (ADI-R) is also considered a gold-standard diagnostic tool in the field, and relies primarily on parent or caregiver report through a structured interview, rather than direct observation (Rutter, et al, 2003).

With regard to diversity, ASD occurs in all racial and ethnic groups, all socio-economic groups, and in all parts of the world. Still, it is challenging to estimate the prevalence in various

groups. This is because minorities and members of lower socio-economic (SES) groups are more difficult to recruit and retain in autism-focused research (Zamora, Williams, Higareda, Wheeler, & Levitt, 2016) and in pediatric research more broadly (Kelly, Ackerman, & Friedman, 2005). Beyond that, there is a second and perhaps more critical finding related to ASD and ethnic minorities and low SES groups: age of diagnosis. Recent research shows that children of ethnic minorities are often diagnosed later in childhood, resulting in them missing out on opportunities for early and intensive intervention which, when combined with functional language at a young age, is associated with optimal outcomes.

People with ASD in general are at risk of a wide range of difficulties, including emotional, behavioral, social, occupational, and economic (e.g. Howlin and Moss, 2012). Studies have also highlighted the complexities involved in ASD females who do not have the opportunity to understand themselves in the context of neurodiversity - due to late or lack-of diagnosis - and the resultant tendency to waste time and efforts on camouflaging behavior (Bargiela et al, 2016). ASD females are at far greater risk of bullying, as well as being taken advantage of socially, due mostly in part to subtle difficulties in perceiving and responding appropriately to social cues. Many of these females have missed out on the benefits of early-intervention, most often in the social realm, and can be plagued with identity issues later in life as they try to play catch-up in light of a new diagnosis. The timely identification of ASD can mitigate some of these risks for ASD females, thus improving the quality of life, by increasing access to services, reducing their self-criticism, and possibly helping to foster a positive sense of identity (Hurlbutt and Chalmers, 2002; Portway and Johnson, 2005; Ruiz Calzada et al, 2012; Russell and Norwich, 2012; Wong et al, 2015). ASD has also been found to be more common in lesbian, gay, bisexual, transgender, or queer (LGBTQ+) populations, who tend to present with

more co-occurring mental health concerns and physical health issues as compared with their cisgender peers with ASD (Hall, Bataza, Streed, Boyd, & Kurth, 2020).

Treatment options for people with ASD are vast and there is no common and agreed upon treatment for ASD. While Applied Behavior Analysis (ABA) therapy is arguably the most common therapy for ASD, there are a range of other options, depending on the individual symptoms. In particular, ABA-based early and intensive intervention has been found to be highly effective (Peters-Scheffer, Didden, Korzilius, & Sturmey, 2011). Due to the social deficits that are hallmark to ASD, speech and language therapy is commonly utilized as well for developing early learning skills as well as social pragmatics and other necessary communication skills. Occupational therapy is frequently prescribed due to the sensory symptoms often associated with ASD and also to support the development of daily living skills. As previously noted, ASD can commonly present with mental health challenges for which more traditional outpatient therapies (e.g. cognitive-behavioral therapy) may be utilized, should the patient have in-tact cognition and language skills. Finally, medication is commonly used to treat various symptoms. It is important to note overall that due to the highly idiosyncratic nature of ASD, the prescribed treatments may look different for each individual depending on areas of strength and deficit. Although ASD is a lifelong disorder, there is some research to suggest that an individual may gain enough adaptive and social skills via effective and intensive intervention where they no longer meet criteria for ASD on standardized measures. It appears at this time that more follow up data and replication may be necessary, as the results remain controversial (Broderick, 2009).

ASD is a complex disorder of which there are still many unknowns. The directions for future research in the field of ASD are vast. Regarding assessment, a need has certainly been demonstrated in the area of furthering our understanding of early markers of ASD in order to

delineate and improve the sensitivity of screening and diagnostic instruments for ASD.

Ultimately, improved interpretation of screening measures may lead to earlier identification for children, who need services in early childhood. Treatment studies of all kinds (ABA, speech, medication trials, etc.) should also be continued to determine effective treatments for ASD.

Finally, biomedical and neurobiological research needs to continue in its pursuits to understand the genetic and neural basis of ASD.

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