Autism Spectrum Disorders and Psychiatry: Update on Diagnostic and Treatment Considerations

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What are the most effective assessment practices for ASD during the developmental stages of early childhood, middle childhood, adolescence, and adulthood?

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This activity offers CE credits for:
1. Physicians (CME)
2. Other

ACTIVITY GOAL
The focus of this article is on diagnostic and treatment considerations in autism spectrum disorder (ASD).

LEARNING OBJECTIVES
At the end of this CE activity, participants should be able to:
• Determine best assessment practices for ASD during the developmental stages of early childhood, middle childhood, adolescence, and adulthood
• Identify the differential diagnoses for each developmental stage
• Decide which interventions are best suited for each particular patient

TARGET AUDIENCE
This continuing medical education activity is intended for psychiatrists, psychologists, primary care physicians, physician assistants, nurse practitioners, and other health care professionals who seek to improve their care for patients with mental health disorders.

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Jennifer Foss-Feig, PhD, has no disclosures to report.
James McPartland, PhD, reports that he was a co-investigator on a Janssen clinical trial.
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DSM-5 criteria for ASD require the presence of symptoms during early development, although a caveat is provided that problems may at times not manifest until social demands exceed limited capacities, which allows later diagnoses to be made. Criteria also include a clause that prequalifies individuals with a DSM-IV diagnosis for a maintained DSM-5 diagnosis. Previously a diagnosis of ADHD could not be made along with ASD; this exclusionary criterion is no longer present, which allows for better capture of comorbid conditions.

Assessment and diagnosis

A comprehensive assessment for ASD includes direct evaluation of the child and collection of collateral information from parents and other caregivers. This includes information about the child’s birth and his or her functioning at home, at school, and in the community as well as a family history and developmental, medical, and intervention history. Psychological testing should be conducted to assess the child’s developmental/cognitive and adaptive (ie, “real world”) functioning to provide a baseline for understanding the child’s learning capacity and current abilities. Other tests, such as evaluations of academic achievement, motor abilities, neuropsychological functioning, and psychological well-being, may be administered to rule out alternative or comorbid diagnoses (eg, learning, attention, or mood problems) and to inform educational and intervention planning. A speech-language pathologist and/or an occupational or physical therapist may conduct additional assessments. Hearing, vision, and/or genetic testing may be required for specific concerns or to rule out alternative explanations for presenting problems.

Multiple screening and diagnostic tools are available for evaluating children and adults for ASD. The Autism Diagnostic Observation Schedule (ADOS), now in its second edition, is a semi-structured, clinician-administered, play- and conversation-based instrument that is widely considered to be the gold standard. While the ADOS is the most psychometrically robust assessment tool available, it also requires extensive training and demonstration of reliability to be used with fidelity. The Autism Diagnostic Interview-Revised (ADI-R) is a structured clinical interview. It has good reliability; however, because of its length, it is used infrequently in clinical practice.

New tools are being developed to assist with expedient ASD-specific history taking and assessment in psychiatric practices. In the interim, a brief clinical interview with direct inquiry about current and historical presentations of DSM-5 symptom domains can often suffice. Because of enormous heterogeneity in the clinical presentation of ASD, the diagnosis is almost always made by an expert with specific training and experience. In many cases, consultation among psychologists, psychiatrists, developmental pediatricians, speech-language pathologists, and educators is critical to tease apart the most likely diagnostic explanation for a child’s presenting problems.

Challenges and differential diagnoses in early childhood

The most reliable early markers for ASD are failure to respond to one’s name; lack of social smiling and shared attention; delays in babbling and lack of words by 16 to 18 months or phrases by 24 months; and failure to use gestures such as pointing, showing, or reaching. Any loss of language or social skills is cause for concern.
The most common reason a toddler is referred for an ASD evaluation is a delay in spoken language. **Table 1** provides a list of questions that need to be answered when considering a diagnosis of ASD in early childhood. There are several competing reasons for which children can present with language delays. As a result, in young children, assessment for ASD is best done by a team of professionals, including a psychologist with extensive knowledge of early childhood, a speech-language pathologist, and often an occupational or physical therapist or pediatrician who can rule out other causes of delay. For children who have ASD or who show early signs of ASD, early detection and consequent intervention are key predictors of positive outcome.

**Challenges and differential diagnosis in middle childhood**

As social, behavioral, and academic demands increase in elementary school, a second wave of children is referred for ASD evaluation. Many children present for diagnostic evaluation who had previously received various, less comprehensive diagnoses (eg, speech delay, ADHD, sensory integration disorder). A primary consideration is whether the social deficit is primary (ie, likely to be due to ASD) or secondary to other challenges. Primary ASD features in middle childhood often relate to difficulty engaging in age-appropriate peer relationships, collaborative and imaginative play, back-and-forth conversation on a flexible range of topics, and age-appropriate interests. Whereas repetitive motor mannerisms (eg, toe walking, hand flapping) are common in typically developing younger children, when these behaviors persist into later years, they are increasingly indicative of ASD.

Differential diagnostic concerns also involve other disorders with overlapping symptoms. For example, children with ADHD and oppositional defiant disorder can have difficulties maintaining friendships because of impulsivity and acting out behaviors. Children with anxiety can have difficulty approaching and engaging peers or show difficulty with transitions and change in routine. Rigid and repetitive behaviors can characterize children with OCD, and some tics can be mistaken as stereotyped motor mannerisms.

Children with continued language delays or communication disorders can also be suspected of having ASD, as can children with global developmental delays or intellectual disability. Differential diagnosis in middle childhood often requires a more comprehensive psychological and/or neuropsychological evaluation to appropriately assess for ASD and rule out other possible explanations for social deficits.

**Challenges and differential diagnosis in adolescents and adults**

For adolescents and adults who present for a first-time evaluation for ASD, it is important to determine whether social withdrawal and difficulties with peer interactions are truly due to a long-standing ASD or to more recent-onset mood or anxiety disorder. Schizophrenia and other psychotic disorders should also be considered. Clinicians must evaluate whether social and other skills have plateaued versus declined more precipitously. In addition, a developmental and family history is needed with particular attention to symptoms of ASD that best differentiate autism from other psychiatric disorders.

Among patients with ASD, comorbid anxiety and depression commonly emerge in adolescence and may require an additional diagnosis to inform targeted treatment. Self-report of anxiety and depression symptoms can be difficult to elicit in individuals with ASD (eg, due to weaknesses in introspection or to expressive language vulnerabilities); thus, increased reliance on caregiver and teacher reports and on behavioral manifestations is necessary.

With increasing attention paid to ASD, a growing number of adults are presenting to clinics to find out whether they might have ASD that had not been identified previously. Differentials prominent in adolescence continue to be important to consider in adulthood. When adults present because of work-related difficulties, consideration of undetected learning disabilities can be worthwhile. First-time diagnosis in adulthood is often complicated by the fact that a good developmental history is difficult to obtain, often requiring clinicians to rely on assessment of current presentation alone.

**Treatment**

Treatment should follow from and be based on a thorough assessment. In addition to characterizing ASD symptomatology, to best inform treatment, an initial assessment should provide information about comorbidities, cognition, academic performance, social behavior, communication skills, and independent living skills. There is no single treatment appropriate for every individual with ASD, and it remains difficult to predict which patients will benefit most from treatment and in which ability domains. The best course of action is to account for age, symptoms, and functioning level when planning treatment. Across all age ranges, interventions must intensively address the individual's most pressing needs.
At the same time, progress should be monitored so that interventions can be started, adjusted, or discontinued in a way that is developmentally sensitive, supports the patient, and allows for emerging independence.

**Behavioral and therapeutic interventions**

Early, intensive, individualized behavioral intervention is highly recommended for toddlers and young children with ASD. Traditional guidelines recommend 25 or more hours per week of direct intervention between a child and a trained clinician; newer models increasingly utilize parents in the role of interventionist to maximize the number of engaged, face-to-face hours a child receives. Applied Behavior Analysis (ABA), derived from early studies done at UCLA by Ivar Lovaas, has been shown to be effective. More recently, ABA approaches have evolved to include increasingly developmentally based and naturalistic models (Table 2). Although more randomized clinical trials are needed—including direct comparisons between active interventions—given the benefits of early intensive interventions for young children with ASD, the ABA approach remains a best-practice recommendation.

As children age, a variety of behavioral, therapeutic, academic, and often psychopharmacological interventions may be necessary. Most children will need a combination of supports, which may change across development and settings. Many children will require speech and language therapy to help them develop basic expressive and receptive language. Some will need intensive speech and language services and behavioral interventions that target the development of alternative communication systems (eg, sign language, picture exchange) when language does not emerge. Even among children with ASD and higher cognitive and language skills, speech and language therapy is often recommended to hone pragmatic skills. Supplemental use of visual communication strategies (eg, picture schedules) is often helpful in conjunction with verbal communication interventions and supports.

Teaching and rehearsing social rules and scripts—first in therapeutic settings and then in more naturalistic settings—are effective for many children. Several manualized social skills programs, such as the Program for Education and Enrichment of Relational Skills (PEERS), have been developed to structure social skills interventions.

Behavioral interventions for children with ASD—both clinician- and parent-mediated—help build skills for the classroom, increase on-task behavior, ease difficulties with transitions and changes in routine, build social and communication skills, improve executive functioning, and decrease challenging behaviors. Functional behavioral analyses can be helpful for identifying behavioral treatment goals and appropriate ways to address them across settings. Overall, intervention plans are best designed when they use strengths (eg, cognitive skills) to compensate for weaknesses, while simultaneously employing additional external supports to facilitate learning and socialization.

For adolescents and adults, support is usually necessary for transitioning from school to community and vocational settings. Many families require assistance to connect with disability services and to consider housing and support options, such as group home settings. Mental health clinicians can be helpful in supporting discussions of long-term planning, legal guardianship, and reasonable expectations for an individual’s work and day-to-day living plans.

For cognitively able individuals, college is often possible, but social and academic support is frequently required to help with navigating the challenges of a more self-directed academic program and dorm life. An increasing number of universities now offer ASD-specific programs. However, these can be costly, and there is little indication of their effectiveness. For many families, it may be necessary to select a college and work with administrators to develop a custom plan to support the student in the college transition.

Persons with ASD often have significant difficulty with obtaining and maintaining employment because of their social vulnerabilities. Psychotherapy and job coaching may be necessary to build interview, interpersonal, and on-the-job vocational skills and to identify employment settings where an individual can be most successful.

Finally, many individuals with ASD desire romantic relationships, and issues of sex and sexuality are common. Guiding exploration and navigation of sexuality and relationship issues can be an important role for mental health clinicians.

**Pharmacotherapy**

There are no medications that effectively target the core social symptoms of ASD. However, many individuals with ASD benefit from drug treatment to address a subset of their presenting symptoms or associated psychiatric problems. Risperidone and aripiprazole are atypical antipsychotics that have demonstrated efficacy and are FDA approved for reducing irritability, challenging behaviors, and repetitive behaviors in children with ASD. There is, however, strong evidence for adverse effects, such as weight gain, sedation, and extrapyramidal effects.
All other medications are used off-label for ASD, although several are FDA approved for children with other conditions. Stimulants can be effective for children with ASD and comorbid ADHD or attention problems. One randomized controlled study showed improvement in hyperactivity among children with ASD. SSRIs are often used to address comorbid mood and anxiety symptoms. Studies have examined whether they are also effective in reducing ASD-specific repetitive behaviors, but support for their effectiveness in this capacity is insufficient.

Newer drugs are in development. The GABA agonist arbaclofen targets challenging behavioral and social withdrawal, and intranasal oxytocin targets social communication deficits. However, while several potential pharmacological agents have shown promise in pre-clinical animal models, clinical trials have demonstrated more tempered results. More research remains to determine both the efficacy and the safety of these drugs. Overall, it is important to consider whether particular medications are helpful in alleviating symptoms and to remain vigilant to avoid polypharmacy, while chasing elusive symptoms whose biological origin and responsiveness to medication are not yet understood.

Complementary and alternative medicine

There is an enormous range of complementary and alternative medicine (CAM) approaches for ASD. Many families use one or more CAM approaches in an attempt to treat core or comorbid symptoms (eg, sleep or gastrointestinal problems, hyperactivity) or to address adverse effects of conventional treatments. CAMs include dietary supplements (eg, vitamins, amino acids, probiotics); alternative interventions (eg, acupuncture, equine therapy, auditory integration training); and biologically based approaches (eg, antibiotics, antifungal medications, digestive enzymes, chelation, hyperbaric oxygen chambers). Gluten- and/or casein-free diets, secretin, and melatonin are also common CAM strategies.

Empirical support for most CAM interventions is limited. Nevertheless, many families are eager to pursue CAM, in part because of anecdotal support for these approaches and in part out of an understandable desire to pursue all options when symptoms persist despite conventional treatment. Many families, however, obtain their information about CAM approaches from other families, the Internet, or non-medical professionals. Thus, it is critical that mental health professionals inquire about and maintain an ongoing dialogue with families about their use of CAM and provide information about evidence-based adverse effects.

As a general heuristic, the principle of “do no harm” applies to CAM as in all other aspects of medicine. If (1) the child is not being harmed physically or mentally and (2) the resource burden CAM interventions place on the family is not detracting from other, evidence-based interventions, then clinicians can support families in trying various treatments. Maintaining a dialogue about what treatments are being pursued and ensuring decisions are made in consultation with a physician are of utmost importance.

Future directions

Precise ASD diagnosis and routinely effective treatment are hampered by the heterogeneity captured under the ASD umbrella and by the as-yet poorly understood etiology of associated deficits. By now, it is widely accepted that ASD is not one disorder, but rather a group of “autisms” that share similar clinical phenotypes but result from diverse genetic and developmental pathways. A promising avenue for characterizing heterogeneity of ASD has grown from the National Institute of Mental Health Research Domain Criteria (RDoC) initiative. Under the RDoC framework, DSM categories are eschewed in favor of cross-cutting, dimensional features that can map biological processes and neurological pathways transdiagnostically. Symptom clusters are conceptualized individually in a manner that allows researchers and clinicians to better understand their respective biological origins and affected brain-based processes, which in turn may lead to more informed and targeted treatment.

At present, the RDoC frame is primarily used in research. However, it may lead to development of more precise subtyping, diagnostic approaches, and treatment strategies that are more congruent with biology and can be more flexibly adapted to heterogeneity in clinical presentation. Another important avenue within ASD research involves identification of biomarkers as predictors and indicators of treatment response and outcome. Researchers and clinicians currently rely primarily on subjective clinician- and parent-report measures to choose treatments and measure outcomes. However, a recently launched project, the Autism Biomarkers Consortium for Clinical Trials (www.asdbiomarkers.org), aims to develop objective biomarkers to evaluate the effectiveness of ASD interventions for social and communicative function. Identification of useful biomarkers and targeted outcome measures has the potential to improve ASD...
treatment on many fronts—leading to more targeted treatment selection, efficient and sensitive measurement of treatment response, and efficient evaluation of novel behavioral and pharmacological interventions. These goals have significant public health benefits and can stand to improve ASD best-practice approaches in the future.

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Table 1 – Questions to consider before making a diagnosis of ASD in to...

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References:


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