Practice Guidelines

Autism Spectrum Disorder: Updated Guidelines from the American Academy of Pediatrics

Key Points for Practice

- AAP, in contrast to the USPSTF, recommends screening for ASD at 18 and 24 months of age.
- Chromosomal microarray and fragile X testing can provide prognostic and genetic counseling information to parents of children with ASD.
- Applied behavior analysis is an intensive behavior therapy for ASD that can improve outcomes, especially at younger ages.
- Wandering and suicide are major safety threats in children and adolescents with ASD.

From the *AFP* Editors

Autism spectrum disorder (ASD) is a neurodevelopmental disorder affecting one in 59 children in the United States and other industrialized nations. In addition to behavior issues, ASD is associated with seizures, gastrointestinal concerns, sleep difficulties, and mental health issues that can profoundly impact the quality of life of children and their families. The American Academy of Pediatrics (AAP) has published updated guidelines for diagnosis and treatment of ASD.

Screening and Diagnosis

Although the U.S. Preventive Services Task Force (USPSTF) found insufficient evidence for screening, the AAP recommends a combination of developmental surveillance and standardized autism screening tests at 18 and 24 months of age based on better outcomes with earlier intervention. Early symptoms that may be identified via surveillance include not responding to name by 12 months of age, not pointing to items of interest by 14 months of age, avoiding eye contact, repetitive movements, unusual reactions to sensations, and echolalia. The AAP recommends the Modified Checklist for Autism in Toddlers (M-CHAT) for initial screening in children up to 30 months of age. Children with persistent deficits on M-CHAT Revised with Follow-Up have a 47% risk of ASD and a 95% risk of any developmental disorder. No screening tools are valid for children older than 30 months.

According to criteria from the *Diagnostic and Statistical Manual of Mental Disorders*, 5th ed., ASD is diagnosed by the presence of three social communication issues and two of four restrictive or repetitive behaviors (https://www.aafp.org/afp/2016/1215/p972.html#afp20161215p972-t1). Nearly one-third of children with ASD have intellectual disability or minimal verbal ability.

After ASD is diagnosed, severity of deficits should be characterized by a multidisciplinary team; all evaluations require formal cognitive and language assessments. Occupational therapy, physical therapy, audiology, and visual assessments also should be included. Children with developmental delays benefit from referral to early intervention school services.

One in 10 children with ASD will no longer meet diagnostic criteria by adulthood. Patients who lose their ASD diagnosis in young adulthood tend to have higher verbal and intellectual capacity in early childhood and earlier intervention.

Testing for Causes of ASD

Chromosomal microarray and fragile X testing are recommended for all children with ASD to predict prognosis. Chromosomal microarray will reveal genetic abnormalities in up to 42% of children with ASD. Fragile X testing is positive in less than 1% of patients with ASD, but it is important for genetic counseling. Targeted testing for disorders

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This series is coordinated by Michael J. Arnold, MD, contributing editor.

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such as tuberous sclerosis and Rett syndrome is useful only if presentation suggests these disorders. Whole exome sequencing shows an abnormality in up to one-fifth of patients with ASD and can be considered if other testing is negative.

Screening for metabolic abnormalities outside of required state newborn screening is typically not useful. Children with motor delays should have creatine kinase and thyroid-stimulating hormone testing. Although children with ASD have increased risk of seizures, findings on screening electroencephalography are often abnormal in children without seizures. Electroencephalography is used only to evaluate neurologic abnormalities and suspected seizures.

Interventions

Early intervention usually includes applied behavior analysis, which focuses on learning and reinforcing acceptable behaviors while extinguishing problematic behaviors. Applied behavior analysis improves cognitive skills and behavior but is an intensive process that typically requires many hours per week of patient participation. Early intervention that includes applied behavior analysis improves cognitive functioning and language skills, with better results from more intense therapy.

Nearly all children with ASD require school-based assistance through an individualized education program, which often includes educational interventions, speech therapy, and occupational therapy. Up to 30% of children with ASD never acquire verbal speech; some will use augmented and alternative communication that includes a picture exchange communication system or speech-generating devices. About two-thirds of preschool children with ASD require occupational therapy for motor, strength, and adaptive skills.

Other treatments proposed for ASD, including antifungal agents, immunotherapy, or hyperbaric oxygen, do not have evidence of benefit. There is caution against using chelation therapy because of ineffectiveness and a risk of harm. Discussing treatments with little or negative evidence often requires shared decision-making with parents or caregivers.

Associated Psychiatric Conditions

Most children with ASD have mood or attention disorders. Cognitive behavior therapy is effective for anxiety in school-aged children with ASD, although medications are often necessary. Depression and suicide occur more often in patients with ASD, making screening important. Suicide risk is elevated in racial minorities and males and those with peer victimization, behavior problems, lower socioeconomic status, and lower education level.

New-onset aggression or self-injury should trigger a medical examination, review of home safety, and potentially emergency stabilization with medication. Antidepressants have inconsistent results in children with ASD and do not effectively treat aggression. Risperidone (Risperdal) and aripiprazole (Abilify) are approved by the U.S. Food and Drug Administration for irritation or aggression in ASD, but they can cause extrapyramidal symptoms (e.g., tardive dyskinesia) and excessive weight gain.

Disrupted sleep is common and can exacerbate behavior issues and limit the effectiveness of behavior therapies. Parent education and behavior interventions can be effective, and melatonin improves sleep in ASD. Other therapies are unproven.

Wandering is a dangerous behavior common in ASD. Nearly one-half of children younger than 10 years have wandered from home, leading to police calls, drownings, and accidents. Strict supervision with boundaries, including locks, is often required.

Associated Medical Conditions

Seizures affect one in 16 children with ASD, and risk increases with intellectual disability. In 20% of patients, the first seizure occurs in adulthood.

Gastrointestinal symptoms are common in children with ASD and often manifest as poor behavior or sleep disruption. Acute abdominal pain may suggest bowel obstruction or perforation. Selective eating is common and can be dangerous if associated with pica. Food refusal often requires assistance from a dietitian and the applied behavior analysis professional. Blood iron and lead levels should be monitored in those with persistent pica.

Poor dentition is a common source of pain, and children with ASD have fewer dental visits. Dentists who use behavioral strategies can prevent the need for sedation with dental treatments.

Transition to Adulthood

Guardianship should be discussed as the patient approaches adulthood. Full or limited guardianship will depend on function and cognitive

ability. Young adults with ASD may qualify for selective security income for care of people with developmental disabilities.

Editor's Note: This updated AAP guideline continues to recommend universal screening for ASD at the 18- and 24-month visits in contrast to the USPSTF findings of insufficient evidence to recommend for or against screening. The American Academy of Family Physicians continues to endorse the USPSTF recommendation. A recent intervention that achieved 91% screening in a medical system with 26,000 children showed the limitations and advantages of ASD screening (https://pediatrics.aappublications.org/content/144/4/e20183963). Only 39% of children with autism were identified by screening, yet those who were identified by screening were diagnosed an average of seven months earlier.—Michael J. Arnold, MD, Contributing Editor

The views expressed in this article are those of the author and do not necessarily reflect the official policy or position of the Department of the Navy, the Department of Defense, or the U.S. government.

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