Pediatric Patient Education™
Expert advice from the American Academy of Pediatrics

ASD—How Is Autism Spectrum Disorder Diagnosed?

Autism spectrum disorder (ASD) is complex, and symptoms are different for each child. Here is information from the American Academy of Pediatrics about how ASD is diagnosed.

Diagnosis of ASD can be complicated for a number of reasons.

- There are no specific medical tests, for example, a blood test, to diagnose ASD, so doctors must rely on information from families about their child's development and behavior and on what can be observed during well-child (health supervision) checkups.
- The condition is complex, and symptoms are different for each child. This is why the AAP recommends there be screening for ASD at specific well-child checkups (18- and 24-month visits) as well as ongoing monitoring of a child's development in the course of well-child care.

Talk with your child's doctor if you feel your child needs to be screened or rescreened and share your concerns—you know your child the best.

Evaluation

When ASD is suspected as a cause of language and social delays, the child should be referred for early intervention (EI) services and an evaluation to determine whether ASD is the proper diagnosis. Although diagnostic evaluations may take some time to arrange, some EI services can start without a diagnosis. To start services through EI programs (younger than 3 years) or the school system (3 years or older), a child must qualify for these services. This qualification will be determined by one of these programs. Meanwhile, a diagnosis of autism may be determined by a doctor or psychologist who has expertise in the diagnosis of ASD or by a team of specialists that might include developmental pediatricians, child neurologists, child psychiatrists, psychologists, speech-language pathologists, occupational or physical therapists, educators, and social workers.

Typically, a diagnostic evaluation includes a

- Careful observation of play and child-caregiver interactions.
- Detailed history and physical examination.
- Review of records of previous EI, school, or other evaluations.
- Developmental assessment of all skills (motor, language, social, self-help, and cognitive). ASD is suspected when the child's social and language functioning is significantly more impaired than their overall level of motor, adaptive, and cognitive skills.
- Hearing test. All children with any speech delays or those suspected of having ASD should have their hearing formally tested.
- Language evaluation that provides standardized scores of expressive language (including speech/gestures) and receptive language (understanding of others speech/gestures), as well as an evaluation of pragmatic language (social use of language) and articulation (pronunciation).

A diagnosis of ASD is made by using all the information collected, such as a child's history, observations, and tests. All these components are important, but they do not have to be completed at the same time. There is value in receiving a diagnosis and continuing to add information to a supports and services plan.

Medical Tests

As mentioned earlier, ASD may be associated with a known syndrome or medical condition. Newer, more sensitive tests may be able to determine an underlying cause of ASD in more children. Laboratory tests may be appropriate to rule out other possible medical conditions that could cause ASD symptoms depending on findings from the child's history and physical examination. If these tests are appropriate, the child may be referred to another specialist, such as a geneticist or a pediatric neurologist, to help diagnose medical conditions that might cause or be associated with symptoms of ASD.

- **Genetic tests.** It is recommended that families be offered genetic testing, such as chromosomal microarray testing, to help identify potential reasons a child might have autism. At present, up to 42% of children with ASD have genetic abnormalities that can be identified through chromosomal microarray testing. Depending on findings from the child's history and physical examination, other types of genetic testing looking for specific disorders like fragile X syndrome, Rett syndrome, or tuberous sclerosis complex may be recommended. Genetic testing should be strongly considered if a child has either unusual physical features or developmental delays or if there is a family history of fragile X syndrome, intellectual disability of an unknown cause, or other genetic disorders. Recommendations for genetic testing may change as new tests are developed.
- **Lead test.** Lead screening is an important component of well-child checkups. A child's blood lead level should be tested when they live in a high-risk environment, such as an older building, or continue to put things into their mouth.
- **Other tests.** Depending on findings from the child's medical history and physical examination, electroencephalography (EEG), magnetic resonance imaging (MRI), or tests for metabolic disorders may be requested. Children with ASD may be picky eaters, so your child's doctor may recommend looking for evidence of iron or vitamin deficiencies, especially vitamin D deficiency.

NOTE: There is not enough clinical evidence to recommend any of the following tests specifically for ASD: hair analysis, the routine measurement of multiple vitamin or nutrient levels, intestinal permeability studies, stool analysis, urinary peptide analysis, or the measurement of mercury or other heavy metals.

Visit HealthyChildren.org for more information.

Adapted from the American Academy of Pediatrics patient education booklet, *Understanding Autism Spectrum Disorder (ASD)*.