



Lab Tests

Why would our child need lab tests?

All children have some lab tests at birth and as part of normal checkups. Children with autism spectrum disorders (ASDs) often need more tests. These tests can help find the cause of the disability or problems related to an ASD that may not be obvious and guide the doctor in treating your child most effectively. New genetic tests now make it possible to find an underlying cause for ASDs in many more children than was previously possible. Genetic testing can also provide information about the likelihood of an ASD in future children or for children of other family members.

What tests might our child's pediatrician order?

The tests your child's pediatrician orders will depend on your child's situation. Often, children with ASDs have vision, hearing, and blood tests including genetic testing. If your child has seizures, the doctor may order neurologic tests. Your pediatrician may send you to a specialist for tests related to intestinal or sleep symptoms.

How will our child's pediatrician decide which tests are needed?

The pediatrician will look at your child's medical and developmental history, current development, physical appearance, behavior, and family history to decide which tests to order. Only tests that provide useful information should be ordered. In some children, very few tests will be needed. Some of the tests that may be ordered are discussed later on.

Why are hearing tests needed?

All children with ASDs should have a hearing test to rule out hearing loss that could account for speech delay. Most infants have a hearing screen near the time of birth, but another test is needed for any child with speech delay, even if the one at birth was normal. The doctor should order a hearing test as soon as she thinks a child might have an ASD.

There are many reasons why a child can lose some hearing during the first few years of life. Although hearing loss does not cause ASDs, it affects development of communication. Rarely, children with severe hearing loss show behaviors similar to children with ASDs.

Why are blood tests needed?

The pediatrician may order blood tests to help understand the cause of your child's ASD, especially if your child has global delays (delays in all or most areas of development). Sometimes it is hard to tell if children with ASDs have additional delays because they often have a hard time following directions and cooperating with testing. Blood tests are usually ordered to look for an abnormality in your child's DNA that might cause ASDs. Presently, abnormalities in DNA can be found in up to 10% to 20% of children with ASDs. Tests usually include a chromosomal microarray to find small areas with extra or missing pieces of DNA and testing for fragile X syndrome. Fragile X syndrome is a common known cause of ASDs, but it accounts for only about 1% to 2% of autism cases.

Some children with ASDs, mainly those with global delays, may have differences in the development of their facial features, hands, feet, and skin. If your child's pediatrician notices any of these features, more blood tests may be ordered. This is to see if these findings are really clues to possible rare genetic syndromes associated with ASDs. A *syndrome* is when a group of symptoms or signs occurs together.

The doctor may also look at your child's birth records to make sure all of the newborn screening tests were normal. If these results cannot be found, new tests may be ordered.

Why are neurologic tests needed?

Some children with ASDs may have seizures. Others may have movements of their face, hands, or arms that they cannot control that are not seizures. If your child's pediatrician thinks your child has seizures, he will likely refer your child to a *neurologist*—a specialist in medical problems affecting the brain. The neurologist may order an electroencephalogram (EEG) to study how your child's brain works.

Your child's head may be larger or smaller than average. Young children with ASDs tend to have larger than average head size. Your doctor or the developmental specialist or neurologist will decide whether a special radiology test called magnetic resonance imaging (MRI) should be obtained to look for any differences in the way your child's brain developed. This is more likely to be ordered if your child has a very small head or an abnormality on neurologic examination.

Neurologic tests may also be ordered to find the cause of regression (loss) or slowing of developmental milestones. About one-third of children who are diagnosed with an ASD appear to develop normally and then lose skills or stop developing new skills for a while. Autism regression often happens between 15 and 24 months of age, and researchers do not know why. If your child is tested during the time of regression, the doctor may order an EEG or MRI. If your child has stopped regressing and is now making progress in development, these tests may not be ordered.

What other tests might our child need?

If your child gets repeatedly sick with vomiting and unusual weakness during flu-like illnesses, your child's pediatrician may suspect a biochemical disturbance and refer you to a metabolic specialist or clinical geneticist for further testing.

Many children with ASDs tend to put things other than food in their mouth even when they are older. If your child does this, the doctor may order a lead level test, especially if your family lives in a high-risk area. Lead does not cause ASDs, but toxic levels can harm learning and make ASD symptoms worse. Iron deficiency may also make children put things other than food in their mouths. Your doctor might look for iron deficiency with blood counts and measures of iron stores. Low iron might also be related to poor sleep.

Glossary

Chromosome testing or karyotype. Chromosome testing is an older test than microarray but still is an important way to find larger rearrangements and abnormalities in DNA structure. DNA is stained with a special dye and looked at under a microscope. If the DNA packages are arranged in a special order and placed on a piece of paper from the biggest to the smallest pairs, this is called a karyotype. There are normally 23 pairs of DNA packages, or chromosomes. This test is still the test of choice for finding non-subtle changes such as an extra chromosome 21, the cause of Down syndrome.

Comparative genomic hybridization microarray. *Microarray* is a highly sensitive test using computer technology that can detect tiny DNA changes, such as an extra or missing piece of DNA. This test is also so sensitive that it can detect variation in the DNA pattern, which is often referred to as *copy number variation* (CNV). In the future, CNV may give important clues about gene changes that can lead to ASD.

Electroencephalogram (EEG). An EEG is a brain-wave test performed to look for seizures, which result from abnormal electrical activity in the brain. Special wires are placed on the child's head and hooked to a machine that picks up electrical waves from the brain and displays them on a monitor. A neurologist is the medical expert who interprets brain-wave patterns.

Fragile X syndrome. Fragile X syndrome is a genetic disorder that causes developmental delay and may be associated with ASDs. It has more severe symptoms in boys than girls because the gene is located on the X chromosome. The diagnosis of fragile X syndrome is made by a special DNA test that looks for certain changes that occur in a specific gene.

Magnetic resonance imaging (MRI). Magnetic resonance imaging of the brain is a sensitive way to look at how the brain has been formed. It does not involve x-ray exposure but does take a while to perform and may require sedation in children with ASDs who are unable to stay still.

The recommendations in this publication do not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account individual circumstances, may be appropriate. Original document included as part of *Autism: Caring for Children With Autism Spectrum Disorders: A Resource Toolkit for Clinicians*, 2nd Edition. Copyright © 2013 American Academy of Pediatrics. All Rights Reserved. The American Academy of Pediatrics does not review or endorse any modifications made to this document and in no event shall the AAP be liable for any such changes.

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