Laboratory Tests

Why would my child need laboratory tests?

All children have some laboratory tests at birth and as part of regular checkups. Children with autism spectrum disorder (ASD) often need more tests. These tests can help find the cause of the condition or problems related to it that may not be obvious. This helps guide your child's doctor in treating your child best.

New genetic tests now make it possible to find a cause for ASD in many more children than before. Genetic testing can also give information about the chance of ASD in future children or for children of other family members.

What tests might my child's pediatrician request?

The tests your child's doctor asks for will depend on your child's situation. Often, children with ASD have vision, hearing, and blood tests including genetic testing. If your child has seizures, your child's doctor may ask for neurological tests, which are tests of the brain and nervous system. Your child's pediatrician may send your child to a specialist for tests for gastrointestinal or sleep symptoms.

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

Your child's pediatrician will look at how your child is developing and acting now and in the past. Also, she will look at whether your child's body or growth shows any differences. This will help her decide which tests to ask for. Only tests that provide useful information should be requested. For some children, very few tests will be needed. Some of the tests that may be asked for are discussed later on.

Why are hearing tests needed?

All children with ASD should have a hearing test to rule out hearing loss that could explain speech delays. Most infants have a hearing test near the time of birth, but another test is needed for any child with speech delays, even if the one at birth didn't show any hearing loss. The doctor should ask for a hearing test for any child with language delays or possible ASD.

There are many reasons why a child can lose some hearing during the first few years after birth. While hearing loss does not cause ASD, it affects how your child communicates. Rarely, children with severe hearing loss show behaviors like those of children with ASD.

Why are blood tests needed?

Your child's pediatrician may ask for blood tests to help understand the cause of your child's ASD, especially if your child has delays in many areas. Sometimes it is hard to tell whether children with ASD have some kinds of delays because they often have a hard time cooperating with testing. Blood tests are usually requested to look for a difference in your child's DNA that might cause ASD. As of now, DNA changes can be found in up to 10% to 20% of children with ASD. Tests could include a chromosomal microarray analysis to find small areas with extra or missing pieces of DNA and testing for fragile X syndrome.

Fragile X syndrome is found in about 1 out of 100 children with ASD.

Some children with ASD, mainly those with many delays, may have differences in the development of their facial features, hands, feet, and skin. If your child's pediatrician notices any of these things, more blood tests may be requested. This is to see if these findings are really clues for rare genetic syndromes associated with ASD. A *syndrome* is when a group of physical differences occur together.

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

Why are neurological tests needed?

Some children with ASD 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 of medical problems that affect the brain. The neurologist may request *electroencephalography* (EEG) to study how your child's brain works.

Your child's head may be larger or smaller than average. Young children with ASD tend to have a larger than average sized head.

Your child's doctor or the specialist will decide whether a special radiology test called *magnetic resonance imaging* (MRI) should be done to look for any differences in the way your child's brain

Laboratory Tests

Autism

Why are neurological tests needed? (*continued*)

formed. This is more likely to be done if your child has a very small head or a difference found during examination.

The tests MRI and EEG may not be useful for every child with ASD.

Neurological tests may also be done to find the cause of loss or slowing of developmental milestones. About 1 in 4 children who has ASD appears to develop normally and then lose skills or stop developing new skills for a while. This is called *regression.* Autism regression often happens between 15 months of age and 24 months of age, and researchers do not know why. If your child is tested during one of these periods, your child's doctor may request EEG or MRI. If your child has stopped regressing and is now making progress in development, these tests may not be requested.

What other tests might my child need?

If your child is often sick with vomiting and unusual weakness during flulike illnesses, your child's pediatrician may suspect a biochemical disturbance and refer your child to a metabolic specialist or clinical geneticist for more testing.

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

Glossary

Chromosome testing or karyotype. Chromosome testing is a form of testing older than chromosomal microarray analysis. It is still 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. When the DNA packages are arranged in a special order and placed onto a piece of paper from the biggest pairs to the smallest pairs, it is called a *karyotype.* There are normally 23 pairs of DNA packages, or chromosomes. This form of testing is still the choice for finding obvious changes such as an extra chromosome 21, the cause of Down syndrome.

Comparative genomic hybridization (also known as chromosomal microarray analysis). Chromosomal microarray analysis 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.*

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

Fragile X syndrome. Fragile X syndrome is a genetic disorder that causes developmental delays and may be related to autism spectrum disorder. The signs are more severe in boys than in girls because the gene is located on the X chromosome. The diagnosis is made by a special DNA test that helps look for certain changes that occur in a specific gene. The changes caused by fragile X syndrome do not show up on a microarray, so a separate test is needed.

Magnetic resonance imaging (MRI). This type of 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 takes a while to perform and may require sedation of children with ASD who are unable to stay still.

Whole-exome sequencing. This is a test that observes the sequence or order of nucleotides, that is, the building blocks of DNA. This test is even more sensitive than chromosomal microarray analysis and may become the standard test in the future.

Insurance coverage for diagnostic testing varies by plan. Make sure to check with your plan if the test will be covered and if you have any co-pays or must meet a deductible first.

Test results often take days to weeks to come back. If a genetic test result is abnormal, your doctor might also test you or your partner to help clarify the results.

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN®