

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, find problems related to an ASD that may not be obvious, and guide the doctor in treating your child most effectively. Almost 90% of kids with ASDs have no known cause of the ASD.

What tests might the doctor order?

The tests your child's doctor orders will depend on your child's situation. Often, children with ASDs have blood and hearing tests. Your child may also be sent for genetic testing. If your child has gastrointestinal (GI) problems, your doctor may order special x-rays of the intestines. If your child has seizures, the doctor may order neurologic tests.

How will the doctor decide which tests are needed?

The doctor 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 recent hearing test to rule out a hearing loss that could account for some of the speech delay. Most infants have a hearing screen near the time of birth. Another test is needed for any child with speech delay, even if the one at birth was normal. 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 can make the symptoms worse. Rarely, children with severe hearing loss show behaviors similar to children with ASDs. The doctor should order a hearing test as soon as she thinks a child might have autism.

Why are blood tests needed?

The doctor may order blood tests to help find 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. When delays are felt to be global, blood tests are usually ordered to test the number and structure of your child's chromosomes and to test for fragile X syndrome. Fragile X syndrome is the most common known cause of ASDs, but it accounts for only about 5% of autism cases. If there is a family history of fragile X syndrome, the doctor will likely order this test even if your child's delays are not global.

Some children with ASDs, mainly those with global delays in all developmental areas, may have abnormal physical features on their face, hands, feet, and skin. If your doctor notices any of these features, he may order more blood tests. This is to see if these findings are really clues to possible rare genetic causes of syndromes associated with ASDs. A syndrome is when a group of conditions 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. If you live in a large city, the doctor may refer you to a clinical geneticist or neurologist who can decide if more tests are needed.

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. It may be hard to know if they are seizures or just movements that are common in children with ASDs. If your child's doctor thinks your child has seizures, she will likely refer your child to a *neurologist*—a doctor who checks the brain. The neurologist may order an electroencephalogram (EEG) to study how your child's brain works. An EEG may be done during an office visit, but it will more likely be done while your child sleeps in a lab or hospital.

Your child's head may be too small or too large. When the only finding is a head that is a little larger than normal, often no more tests are needed. About one third of children have "big" heads during the first few years of life, and no treatment is needed. The neurologist may also order special x-ray tests called computed tomography (CT) scans or magnetic resonance imaging (MRI) to look for any unusual findings inside the head.

Neurologic tests may also be ordered to find the cause of regression (going backwards in development). About one third of children who are diagnosed with an ASD appear to develop normally and then regress. Autism regression often happens between 15 and 24 months, 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 (unless the regression was earlier or later than normal or in other areas of your child's development).

What other tests might our child need?

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

Many children with ASDs tend to put nonfood items 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 the autistic symptoms worse.

The information contained in this publication should not be used as a substitute for the medical care and advice of your pediatrician. There may be variations in treatment that your pediatrician may recommend based on individual facts and circumstances.

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