Genetic Testing for Autism Spectrum Disorder: What to Expect

Genetic testing may help find a cause of your child’s autism spectrum disorder (ASD) in his/her genetic makeup, or DNA. In this handout, you will learn the types of genetic testing we do for children with ASD and how we can best care for your child based on the test results.

WHAT IS AUTISM SPECTRUM DISORDER?
Autism spectrum disorder (ASD) is a disorder that affects the way your child’s brain develops.

Children who have ASD have a harder time with speech and language and interacting with others. They can also have behaviors they repeat, like flapping their hands, jumping, repeating words or arranging toys in a certain order.

Your child’s ASD diagnosis should be made by a developmental specialist or neurologist (doctor who cares for the brain and nerves).

IS THERE A GENETIC CAUSE OF ASD?
Yes. About 1 of every 10 children with ASD has a genetic cause that can be found by genetic testing. Although we believe genetics play a role in ASD, this doesn’t appear to be the only factor. We also believe that many other factors play a role in whether a child develops ASD.

Many times, a child with ASD is the first one to be affected in the family. In some cases, there is a clear genetic cause of the ASD. Some of these genetic changes are passed down within a family and some start new in the child with ASD. Changes that start new are caused by chance. They are not the parents’ fault.

In other cases, there are genetic factors that increase the risk for a child to develop ASD. This means that there are many factors coming together to cause ASD. Genetics is just 1 piece of the puzzle. In other cases, there might be many outside factors that cause ASD and genetics does not play a role.

HOW CAN GENETIC TESTING HELP FIND A CAUSE OF MY CHILD’S ASD?
Genetic testing is used to try and better understand the cause of your child’s ASD. Genetic testing alone does not diagnose ASD.

If current testing doesn’t find a genetic cause for your child’s ASD, it doesn’t mean the cause isn’t genetic. It means our current testing hasn’t found the cause yet. Genetic testing is changing all the time with research, so you can ask if your child should have more genetic testing in about 2-5 years.
WHAT TYPES OF GENETIC TESTING WILL MY CHILD HAVE?

Your child may have 2 or 3 types of genetic tests, based on his/her gender and certain symptoms. The tests might include:

- **Chromosomal microarray.** This test looks at all of the pieces of your child’s DNA and checks for pieces that are missing (called microdeletions) or pieces that are extra (called microduplications).

- **Fragile X testing.** This is a test done to find Fragile X syndrome in boys. Boys who have Fragile X syndrome have intellectual disabilities (below average intelligence and set of life skills before a certain age) and sometimes ASD. In rare cases, girls can have a mild (less serious) form of Fragile X syndrome.

- **Metabolic testing.** This testing checks for inborn errors of metabolism (rare disorders in which the body can’t turn food into energy) that rarely cause ASD.

WHAT HAPPENS IF MY CHILD’S TEST RESULT IS POSITIVE OR NEGATIVE?

- **If your child’s test result is positive for a genetic cause of ASD,** we can try to help personalize your child’s care. Sometimes, we can learn more about what your family can expect in the future. We can also connect you with other families who have children with the same genetic cause.

- **If your child’s test result is negative for a genetic cause of ASD,** we will talk with you about the best choices for your child’s care plan. Your geneticist (genetics doctor) might also test for other conditions that might cause your child’s ASD. Genetic testing is changing all the time with research, so your doctor might suggest more genetic testing in about 2-5 years.

How do genetic traits get passed along?

Everyone has 2 copies of just about every gene in their body. Our genes are the instructions that tell our body how to build and develop. A person’s genes determine his/her eye color, hair color, different disorders, height, weight and much more. Our genes are packaged into chromosomes. Everyone gets 1 copy of each chromosome from each parent. When you combine both sets of chromosomes from the parents, the child has a total of 46 chromosomes (23 from the mother and 23 from the father).

Chromosomes live in every cell in the body. When the body makes new cells, the DNA must copy itself over and over again. Sometimes, pieces of the DNA are left out or extra pieces are added in. These copy mistakes can cause different disorders, including ASD. These mistakes are random. They are not anyone’s fault and are not caused by something a parent did or didn’t do.