



### Why would my child need genetic testing?

We recommend genetic testing for all children with Autism Spectrum Disorder. Testing may help explain why your child has autism and can give your family helpful information about:

- The best kind of medical care for your child
- Other possible health problems that can happen with certain genetic conditions
- Autism risk in other family members, including future pregnancies

### What is a genetic test?

A genetic test is usually a blood test. It looks at chromosomes and genes. Chromosomes contain genes, and genes tell our bodies how to function. There are many types of genetic tests, and your child's doctor will work with you to decide on which are best for your child. The most common tests are **chromosomal microarray (CMA)** and **Fragile X** testing.

### Chromosomal microarray (CMA)

#### How it works:

- CMA looks for missing or extra pieces of chromosomes.
- Having missing or extra pieces of chromosome can cause autism, especially if that piece has important genes.
- Everyone has some tiny missing or extra pieces of chromosomes. The missing or extra pieces do not always cause problems.
- Chromosomes come in pairs. One chromosome from each pair comes from each parent (mother and father). For example, a person has two copies of chromosome 1. One copy comes from the mother, and the other copy comes from the father.
- Sometimes a person may inherit both copies of a chromosome from the same parent. For example, both copies of chromosome 1 come from the mother with no copy coming from the father. This can happen with just a portion of the chromosome. For instance, one chromosome 1 comes from the mother, while the other chromosome 1 has portions from the mother *and* the father. CMA can tell if this is happening in your child. It could indicate that the mother and father are potentially closely or distantly related. In this situation, your child may be at higher risk for certain genetic disorders.

#### Possible results:

Your child's test can have 3 different results.

1. Normal
  - No missing or extra pieces of chromosomes were seen.
  - Your child inherited one copy of each chromosome from each parent.
2. Variant of uncertain significance
  - A missing or extra piece of a chromosome was seen.
  - A pair of chromosomes (or part of a pair) was found to be from the same parent, and it's not clear if this caused autism or other health issues.
  - Sometimes we test the parents to see if the change runs in the family, and to see if the chromosome change is the cause of your child's autism.
3. Abnormal
  - A missing or extra piece of chromosome causes autism, or another condition was seen.
  - A pair (or part of a pair) of chromosomes was found to be from the same parent and that caused autism or a particular condition.
  - Sometimes the abnormal result is not related to autism at all but might show another genetic condition.

### Fragile X syndrome testing

#### How it works:

- Fragile X testing looks for changes at a gene located on the X chromosome, called "FMR1."
- Males are usually more affected than females. But both may show some symptoms, like behavioral and learning problems.

#### Possible results:

Your child's test results may be normal or abnormal.

- Abnormal
  - It could show that your child has Fragile X syndrome or that your child is a carrier for Fragile X syndrome. Children with this condition have intellectual disabilities, behavior and learning challenges, and certain physical features.
  - Your doctor will go over any abnormal results with you.

## When will I find out the results?

- It usually takes 4-8 weeks. Your child's doctor may recommend a visit with a genetic specialist for more information about what the results mean for your family.

## How much does it cost?

We recommend talking with your insurance company to understand the costs of genetic tests. They may decide what genetic testing can be done and how much you need to pay out-of-pocket.

Please ask your child's doctor if you need insurance authorization and what the cost to you may be.

### Contact us

Genetics Department (857) 218-4637