



## **What is autism?**

Autism spectrum disorder (ASD) is a complex developmental condition that can cause difficulties with communication, social interaction, and behaviors. Autism is referred to as a “spectrum” because of the variability in the effects and severity of symptoms that impact a person. The diagnosis of ASD is often made in early childhood, and is a lifelong condition. According to the [Centers for Disease Control \(CDC\)](#), about 1 in 59 children (1-2%) are affected by ASD.

Of note, in 2013, updates to the classification and diagnostic criteria for ASD were released ([Diagnostic and Statistical Manual of Mental Disorders \(DSM-5\)](#)). Those previously diagnosed with autistic disorder, Asperger’s syndrome, or pervasive developmental disorder not otherwise specified (PDD-NOS), are now all referred to as “autism spectrum disorder” or ASD.

## **What causes autism?**

In general, we do not fully understand the underlying cause for ASD. For the majority, it is likely caused by a combination of multiple genetic and environmental (nongenetic) factors (called [multifactorial](#)). Researchers have shown that about 20-40% of those with ASD will have an underlying genetic disorder, which means that they have a specific change in their genetic material that has contributed to their ASD. The remaining cases are likely due to a combination of factors. Examples of nongenetic risk factors include advanced parental age (when a man is 40 years old or older at the time of conception), short periods between pregnancies (less than 12-24 months), and pregnancy or delivery complications. In addition, the chances a child will have ASD increases if they have a family member with ASD.

For those who have an underlying genetic condition as the cause of their ASD, they may also have a chromosome abnormality or single gene disorder which can involve other health issues (such as [PTEN Hamartoma Tumor syndrome](#)). The likelihood of an underlying genetic condition may be impacted by whether a child has other features or health problems, such as distinctive physical features, [birth defects](#), seizures, etc. This is often referred to as “syndromic” or “complex” ASD, and is more likely to have a genetic cause.

## **What type of genetic testing may be recommended?**

There are multiple types of tests that may be recommended for those who have ASD. The most common include [chromosome microarray](#) and [fragile X](#) testing. The [chromosome microarray](#) is used to identify small missing or extra pieces of chromosome material, and



can identify or rule out several hundred known genetic syndromes. This testing has a detection rate (the chance for the testing to come back positive) of about 5-15% in those with ASD. [Fragile X](#) is a specific condition that can cause ASD, and accounts for less than 5% of those with ASD.

Further genetic testing that may be considered includes single gene testing, [multigene panels](#), or [whole exome sequencing](#). About 5-15% of individuals with ASD are found to have a single gene disorder that can be found with these types of tests.

Other testing that may be recommended includes biochemical screening, which involves measuring the levels of different substances in the blood or urine. These tests are used to look for [metabolic conditions](#) that can also cause ASD. These type of conditions account for less than 1% of those with ASD.

### **What is the chance to have another child with autism?**

The chance to have another child with ASD (called the recurrence risk) can be dependent on whether an underlying genetic cause was identified. For instance, if the ASD in the family is determined to be caused by a genetic condition, the recurrence risk could be as high as 50%. In general, for a full sibling of an individual with ASD (regardless of other health conditions or if an underlying genetic cause was identified) the chance ranges from 7% to 20%. For families where there are two children with ASD the recurrence risk for a full sibling to be affected is estimated to be 25-35%. For other family members, such as half siblings, the chance is increased over the general population chance of 1-2%. It is also important to note that siblings of those with ASD are more likely to have language delays or autistic-like speech patterns, but may not have ASD.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

### **Additional Resources**

[Autism Speaks](#)

[Center for Disease Control \(CDC\)](#)

[National Institute of Health\(NIH\)](#)