

Understanding Your Positive AutismNext Genetic Test Result

INFORMATION FOR PATIENTS WITH A **PATHOGENIC MUTATION** OR **VARIANT THAT IS LIKELY PATHOGENIC**

Result	MUTATION	The testing done for you/your family member shows a pathogenic mutation (a disease-causing change in the gene, like a spelling mistake), or a variant that is likely disease-causing, in a gene that causes an autism spectrum disorder (ASD).
Gene	DEFINITION	Genes are instructions for how our bodies work and develop. Everyone has two copies of each gene, one from each parent. Mutations (changes in the gene, like spelling mistakes) in certain genes can cause an ASD. These mutations may be passed down in families or be brand new for the person who has an ASD. Even if there is no history of ASD in your family, it can still be caused by a change in a gene.
Cause	GENETIC	The testing done confirms a genetic cause for the ASD in you/your family member. Sometimes an ASD can be the first sign that a person may have other medical problems. Talk with your healthcare provider to learn more about whether you/your family member might need any additional medical screening.
Management Options	FOR PEOPLE WITH AN ASD	Some genetic test results can help identify treatments that are more or less likely to be helpful for people with an ASD. Treatment options can include: medication, speech/ language therapy, occupational therapy, physical therapy, behavioral intervention, and educational support/assistance. Knowing the genetic cause for the ASD in you/your family member may also help to avoid some tests or procedures. Talk to your healthcare provider about which may be right for your family member.
Screening Options	FAMILY MEMBERS	In some families, gene mutations that cause ASDs can be inherited. Your healthcare provider can talk with you more about the inheritance pattern of ASD based on this test result and your family history, and what this means for the rest of your family.
Next Steps	DISCUSS	It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers. Talk to your healthcare provider about who in your family might benefit from considering genetic testing.
Reach Out	RESOURCES	<ul style="list-style-type: none"> ▪ Ambry's Neurology Site for Families ambrygen.com/patients/neurology ▪ Autism Speaks autismspeaks.org ▪ Talk About Curing Autism tacanow.org ▪ National Society of Genetic Counselors nsgc.org ▪ Canadian Association of Genetic Counsellors cagc-accg.ca

How ASDs Can Run in a Family

Many people with an ASD are the first person in their family to have it. Often, genetic testing can find a gene mutation for someone with an ASD that is not found in other family members and was not passed down from a parent. In other families, gene changes that cause ASDs can be inherited from earlier generations.

Please discuss this information with your healthcare provider. The field of genetics is continuously changing, so updates related to your genetic testing result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or taken as medical advice.