

Understanding Your Positive Neurodevelopmental Disorder Genetic Test Result

INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

Result	MUTATION	The testing done for you/your family member found 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 neurodevelopmental disorders (NDDs), including epilepsy, intellectual disability (ID), and/or autism spectrum disorders (ASDs). Either a mutation or a variant that is likely disease-causing should be treated as a positive result.
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 NDDs. These mutations may be passed down in families or be brand new for the person who has an NDD. Even if there is no history of NDDs 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 NDD in you/your family member. Sometimes NDDs 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 additional medical screening.
Management Options	FOR PEOPLE WITH AN NDD	Some genetic test results can help identify medical management options that are more or less likely to be helpful for someone with an NDD. This can include options such as: medication, specific diet, surgery, speech/language therapy, occupational therapy, physical therapy, behavioral intervention, and educational support/assistance. Knowing the genetic cause of an NDD for you/your family member may also help to avoid some tests or procedures. Talk to your healthcare provider about which management options may be right for you/your family member.
Family Members	VARIES	Many people with an NDD are the first person in their family to have it. Often, genetic testing can find a gene mutation for someone with an NDD that is not found in other family members and was not passed down from a parent. In other families, gene changes that cause NDDs can be inherited from earlier generations. Your healthcare provider can talk with you about how an NDD may run in your family based on your test results and family history, and what this means for the rest of your family.
Next Steps	DISCUSS	It is recommended that you share this information with your 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 patients.ambrygen.com/neurology ▪ American Epilepsy Society aesnet.org ▪ Autism Speaks autismspeaks.org ▪ The Arc thearc.org ▪ Child Neurology Foundation childneurologyfoundation.org ▪ National Society of Genetic Counselors nsgc.org ▪ Canadian Association of Genetic Counsellors cagc-accg.ca

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.