

Understanding Your Positive Neurodevelopmental Disorder Genetic Test Result

INFORMATION FOR PATIENTS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT

Result	The genetic testing done for you or your family member shows a pathogenic or likely pathogenic variant (a change in a gene that is disease-causing or likely to be disease-causing) in a gene that causes neurodevelopmental disorders (NDDs), including epilepsy, intellectual disability (ID), and/or autism spectrum disorder (ASD).
Genes and Inheritance	Genes are instructions for how our bodies work and develop. Everyone has two copies of each gene, one from each parent. Changes in certain genes can cause NDD and may be passed down in families or be brand new in the person who has NDD. Even if there is no history of NDD in your family, it can still be caused by a change in a gene.
Cause	There are many causes for NDD; however, the genetic testing 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	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 with your healthcare provider to learn more about how treatment options may change based on the genetic test result.
Family Members	Many people with NDD are the first person in their family to have it. Often, genetic testing can find a gene change in someone even if the gene change was not found in other family members and was not passed down from a parent. In other families, gene changes can be inherited from earlier generations. Talk to your healthcare provider about how NDD may run in your family based on your test results and family history, and what this means for the rest of your family. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.
Resources	<ul style="list-style-type: none"> • 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.