Ambry Genetics®

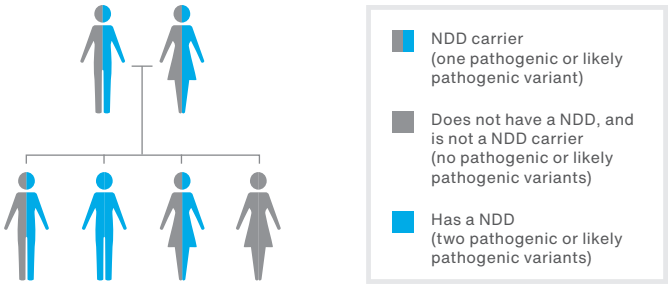
Understanding Your Neurodevelopmental Disorder Carrier Genetic Test Result

INFORMATION FOR PATIENTS WITH **ONE PATHOGENIC OR LIKELY PATHOGENIC VARIANT**

Result	The genetic testing done for you or your family member shows one pathogenic or likely pathogenic variant (a change in a gene that is disease-causing or likely to be disease-causing) in a recessive gene that causes neurodevelopmental disorders (NDDs), including epilepsy, intellectual disability (ID), and/or autism spectrum disorder (ASD). This means that you or your family member is a "carrier".
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 both copies of certain recessive genes can cause NDDs. Carriers have a change in just one copy of the gene and typically do not have symptoms of a NDD but can have children with a NDD.
Family Members	If a carrier's partner is also a carrier of a pathogenic or likely pathogenic variant in the same gene, there is a 1 in 4 (25%) chance for each child to have a NDD. Genetic testing for a carrier's partner may provide more information about the risk to have a child with a NDD. In addition, other family members may wish to be tested to determine if they also have the pathogenic or likely pathogenic variant found in your family. It is recommended that you share this information with family members so they can learn more and discuss with their healthcare providers.
Resources	▪ 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

How Recessive NDDs Are Inherited

There is a 50/50 random chance to pass on the pathogenic or likely pathogenic variant from a parent to a child. If the other parent also happens to carry a pathogenic or likely pathogenic variant in the same gene, there is a 25% chance that a child would have a NDD, a 50% chance that a child would be a carrier, and a 25% chance that a child would not have a NDD or be a carrier.



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 interpreted as medical advice.

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