Ambry Genetics®

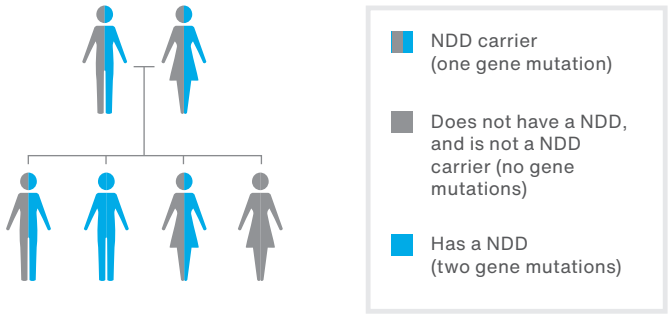
Understanding Your Neurodevelopmental Disorder Carrier Genetic Test Result

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

Result	CARRIER	The testing done found that you/your family member has one pathogenic mutation (a disease-causing change in the gene, like a spelling mistake) or 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). This means you/your family member is a "carrier" for a NDD. It is unlikely that having only one mutation in this gene causes a NDD.
Gene	DEFINITION	Genes are instructions for how our bodies work and develop. Everyone has two copies of each gene, one from each parent. A mutation in **both** copies of certain genes can cause NDDs. Carriers have a mutation in **only one** copy of their gene. Carriers do not usually have the symptoms of a NDD, but can have children with a NDD.
Screening Options	FAMILY MEMBERS	If a carrier's partner is also a carrier of a mutation in the same gene, there is a 1 in 4 (25%) chance to have a child with a NDD in each pregnancy. Genetic testing for a carrier's partner may provide more information about this risk. In addition, other adult family members may wish to be tested to see if they carry the mutation found in 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. They can be tested for the same mutation, if they choose to.
Reach Out	RESOURCES	Ambry's Neurology Site for Families ambrygen.com/patients/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

How Some NDDs Are Inherited

For some NDDs, people who have a mutation in only one gene are "carriers" and do not usually have symptoms. If they have children with a partner who is a carrier of a mutation in the same gene, there is a 1 in 4 (25%) chance for them to have a child with a NDD in each pregnancy together. There is a 1 in 2 (50%) chance for them to have a child who is a carrier (usually without symptoms), and a 1 in 4 (25%) chance for them to have a child who does not have a NDD, and is not 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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