Ambry Genetics®

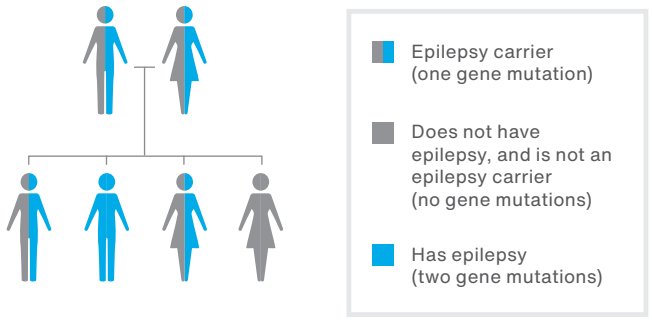
Understanding Your Epilepsy Carrier Genetic Test Result

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 a variant that is likely pathogenic, in a gene that causes epilepsy. This means you/your family member is a "carrier" for this epilepsy gene. It is unlikely that having only one mutation in this gene causes epilepsy.
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 epilepsy. Carriers have **one** mutation in a copy of their gene. Carriers do not usually have the symptoms of epilepsy, but can have children with the condition.
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 epilepsy 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 with their healthcare providers. They can be tested for the same mutation if they choose.
Reach Out	**RESOURCES**	American Epilepsy Society aesnet.org Citizens United for Research in Epilepsy cureepilepsy.org Epilepsy Foundation epilepsy.com National Society of Genetic Counselors nsgc.org Canadian Association of Genetic Counsellors cagc-accg.ca

How Some Epilepsy is Inherited

For some epilepsy disorders, with what is known as recessive inheritance, people who have a mutation in only one gene are considered "carriers" and do not usually have symptoms of the condition. If they have children with someone else who is a carrier for a mutation in the same gene, there is a 1 in 4 (25%) chance for them to have a child with epilepsy 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 epilepsy, 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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