Ambry Genetics®

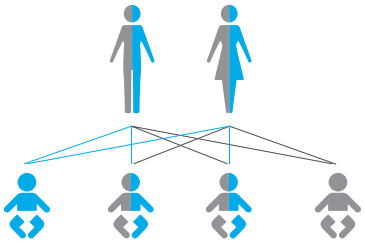
Understanding Your Epilepsy Carrier Genetic Test Result

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

Result	**MUTATION**	The testing done shows that you/your family member has one pathogenic (disease-causing) mutation, or a variant that is likely disease-causing, in a gene that causes epilepsy. This means you/your family member is a "carrier" for epilepsy. 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 (change in the gene, like a spelling mistake) 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 your partner is also a carrier of a mutation in the same gene, you could have a 1 in 4 (25%) chance to have a child with epilepsy in each pregnancy together (see below). Genetic testing for your partner may help you both learn more about this. In addition, your adult family members may wish to be tested to see if they carry the mutation(s) 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 now be tested for the same mutation(s), if they choose to.
Reach Out	**RESOURCES**	• American Epilepsy Society aesnet.org • Citizens United for Research in Epilepsy cureepilepsy.org • Epilepsy Foundation epilepsy.com • Rare Epilepsy Network ren.rti.org • National Society of Genetic Counselors nsgc.org • Canadian Association of Genetic Counsellors cagc-accg.ca

How Epilepsy is Inherited

People who have a mutation in only one copy of an epilepsy gene are "carriers," and do not usually have symptoms. 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 2 in 4 (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.



- Epilepsy carrier (one gene mutation)
- Does not have epilepsy, and is not an epilepsy carrier (no gene mutations)
- Has epilepsy (two gene mutations)

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.

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