Ambry Genetics®

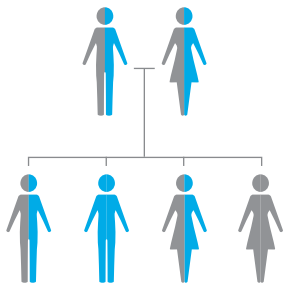
Understanding Your Epilepsy Carrier Genetic Test Result

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 epilepsy. 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 epilepsy. Carriers have a change in just one copy of the gene and typically do not have epilepsy but can have children with epilepsy.
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 epilepsy. Genetic testing for a carrier's partner may provide more information about the risk to have a child with epilepsy. 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 ▪ 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 Recessive Epilepsy is 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 epilepsy, a 50% chance that a child would be a carrier, and a 25% chance that a child would not have epilepsy or be a carrier.



- Epilepsy carrier (one pathogenic or likely pathogenic variant)
- Does not have epilepsy, and is not an epilepsy carrier (no pathogenic or likely pathogenic variants)
- Has epilepsy (two pathogenic or likely pathogenic variants)

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.

One Enterprise, Aliso Viejo, CA 92656 USA Toll Free +1.866.262.7943 Fax +1.949.900.5501 **ambrygen.com**

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