

Understanding Your Positive Epilepsy Genetic Test Result

INFORMATION FOR PATIENTS WITH A **PATHOGENIC MUTATION OR VARIANT THAT IS LIKELY PATHOGENIC**

Result	NEGATIVE	The testing done for you/your family member shows a pathogenic (disease-causing) mutation, or a variant that is likely disease-causing, in a gene that causes epilepsy. Both mutations and variants that are likely disease-causing should be treated as the same type of positive result.
Gene	DEFINITIONS	Genes are instructions for how our bodies work and develop. Everyone has two copies of each gene, one from each parent. Mutations (changes in the gene, like spelling mistakes) in certain genes can cause epilepsy. These mutations may be passed down in families or be brand new for the person who has epilepsy. Even if there is no history of epilepsy in your family, it can still be caused by a change in a gene.
Cause	GENETIC	There are many causes for epilepsy. However, the testing done confirms a genetic cause for the epilepsy in you/your family member. Sometimes epilepsy can be the first sign that a person may have other health or medical problems. Talk with your healthcare provider to learn more about whether you/your family member might need any additional screening.
Management Options	FOR PATIENTS WITH EPILEPSY	Some epilepsy genetic test results can help identify specific treatments that are more or less likely to be helpful. Treatment options can include: medication, specific diet, vagus nerve stimulation, surgery, or avoiding certain situations that can provoke seizures. Knowing the genetic cause of epilepsy for you/your family member may help to avoid some tests or procedures. Talk to your doctor about which may be right for you/your family member.
Screening Options	FAMILY MEMBERS	In some families, gene mutations that cause epilepsy can be inherited. Your doctor or genetic counselor can talk with you more about the inheritance pattern of epilepsy in your family based on your test results and your family history.
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. Talk to your doctor or genetic counselor about who in your family might benefit from considering this testing.
Reach Out	RESOURCES	<ul style="list-style-type: none"> • 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 Can Run in a Family

Many people with epilepsy are the first person in their family to have it. Often, genetic testing can find a gene mutation for someone with epilepsy that is not found in other family members and was not passed down from a parent. In other families, gene changes that cause epilepsy can be inherited from earlier generations.



Please discuss this information with your healthcare provider. The field of genetics is continuously changing, so updates related to your epilepsy 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.