

Understanding Your Positive Epilepsy Genetic Test Result

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

Result	MUTATION	The testing done for you/your family member shows a pathogenic mutation (a disease-causing change in the gene, like a spelling mistake), or a variant that is likely disease-causing, in a gene that causes epilepsy. Either pathogenic mutations or variants that are likely disease-causing should be treated as a 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 medical management options that are more or less likely to be helpful. Management options can include: medication, specific diet, 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 healthcare provider 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 healthcare provider can talk with you more about how epilepsy runs in your family based on your test results and family history.
Next Steps	DISCUSS	It is recommended that you share this information with your family members so they can learn more and discuss this with their healthcare providers. Talk to your healthcare provider about who in your family might benefit from genetic 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 ▪ National Society of Genetic Counselors nsgc.org ▪ Canadian Association of Genetic Counsellors cagc-accg.ca

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.