

Understanding Your VUS Epilepsy Genetic Test Result

INFORMATION FOR PATIENTS WITH ONE OR MORE VARIANTS OF UNCERTAIN SIGNIFICANCE (VUS)

| Result | The genetic testing done for you or your family member shows one or more variants of uncertain significance (VUS) in one or more genes that cause epilepsy. A VUS is a change in a gene that may or may not cause epilepsy. It is classified as uncertain because there is not enough information currently available about the variant to know if the variant causes epilepsy or not. |
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| 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 certain genes can cause epilepsy and may be passed down in families or be brand new in 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. Also, some types of epilepsy are not caused by changes in a gene. |
| Diagnosis | This testing does not change you or your family member's epilepsy diagnosis. If you or your family member have been diagnosed with epilepsy, that remains the same. |
| Family Members | Your report will indicate if testing family members may help us learn more about your specific VUS. Please speak with your healthcare provider to determine if they might also benefit from a test to evaluate their personal risk of developing a disease. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers. |
| Management Options | Medical management options can include: medication, specific diet, surgery, or avoiding certain situations that can bring on seizures. Talk to your healthcare provider about which treatment options may be helpful for you/your family member. |
| Reclassification | Collecting information about a VUS is an ongoing process, so it is possible that your result may be better understood in the future. The healthcare provider that ordered your test will be notified if new information becomes available about your VUS. |
| 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 |

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.