

Understanding Your VUS Neurodevelopmental Disorder 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 genes that cause a neurodevelopmental disorder (NDD), including epilepsy, intellectual disability (ID), and/or autism spectrum disorder (ASD). A VUS is a change in a gene that may or may not cause a NDD. It is classified as uncertain because there is not enough information currently available about the variant to know if the variant causes a NDD or not.
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 a NDD and may be passed down in families or be brand new in the person who has a NDD. Even if there is no history of a NDD in your family, it can still be caused by a change in a gene. Also, some NDDs are not caused by changes in a gene.
Diagnosis	This testing does not change your/your family member's NDD diagnosis. If you or your family member has been diagnosed with an NDD, 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	Management options vary by condition and other factors. Talk to your healthcare provider about which treatment options may be helpful for you or your family member.
Reclassification	Collecting information about a VUS is an ongoing process. 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	<ul style="list-style-type: none"> ▪ American Epilepsy Society aesnet.org ▪ Autism Speaks autismspeaks.org ▪ The Arc thearc.org ▪ Child Neurology Foundation childneurologyfoundation.org ▪ 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 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.