

Understanding Your VUS Neurodevelopmental Disorder Genetic Test Result

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

Result	VUS	The testing for you/your family member found one or more variants of unknown significance (VUS) in genes that cause neurodevelopmental disorders (NDDs), including epilepsy, intellectual disability (ID), and/or autism spectrum disorders (ASDs). A VUS is a gene change, but there is not enough information available to know if it causes NDDs or not.
Gene	DEFINITION	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 are known to cause NDDs. Even if there is no one else with an NDD in your family, it can still be caused by a change in a gene.
Diagnosis	NO CHANGE	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	POSSIBLE TESTING	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.
Management Options	FOR PEOPLE WITH AN NDD	Management options vary by condition and other factors. Talk to your healthcare provider about which may be right for you/your family member.
Reclassification	POSSIBLE	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.
Next Steps	DISCUSS	Please share this information with your family members so they can talk with their healthcare providers and learn more.
Reach Out	RESOURCES	<p>Ambry's Neurology Site for Families patients.ambrygen.com/neurology</p> <p>American Epilepsy Society aesnet.org</p> <p>Autism Speaks autismspeaks.org</p> <p>The Arc thearc.org</p> <p>Child Neurology Foundation childneurologyfoundation.org</p> <p>National Society of Genetic Counselors nsgc.org</p> <p>Canadian Association of Genetic Counsellors cagc-accg.ca</p>

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.