

Understanding Your Positive NeuropathySelect™ Genetic Test Result

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

Result	POSITIVE	The result of your genetic testing shows you either have a change that is known to be disease-causing (pathogenic mutation) or a change or gene variant that is likely to be disease-causing (likely pathogenic), in a gene that causes polyneuropathy (peripheral and/or autonomic) and/or hereditary amyloidosis. Either should be considered as the same type of positive result.
Mutation	DEFINITION	An alteration or change in a gene, like a spelling mistake, that can affect the function of that gene.
Gene Variant	DEFINITION	Everyone has two copies of each gene; one gene copy from each parent. Mutations or likely pathogenic variants in certain genes can cause polyneuropathy and/or hereditary amyloidosis. These mutations or gene variants may be passed down in families (inherited) or be brand new for the person who has polyneuropathy and/or hereditary amyloidosis. Even if there is no history of polyneuropathy and/or amyloidosis in your family, it can still be caused by a change in a gene variant.
Medical Management Options	FOR PATIENTS WITH POLYNEUROPATHY AND/OR AMYLOIDOSIS	Medical management options may include: medications, supportive therapies, supportive devices, surgery and organ transplantation. Talk to your healthcare providers about which may be right for you.
Screening Options	FOR FAMILY MEMBERS	Options for screening and early detection include: physical exams, imaging studies, laboratory tests, biopsies and other specialized analysis (electromyography, nerve conduction velocity testing, etc.). Talk to your healthcare providers about whether these medical management options are right for you and/or your family.
Family Members	VARIES	In some families, gene mutations that cause polyneuropathy and/or hereditary amyloidosis can be inherited. Your healthcare provider can talk with you more about the inheritance pattern of polyneuropathy and/or hereditary amyloidosis based on this test result and your family history, and what this means for the rest of your family.
Next Steps	DISCUSS	Please share this with family members so they can talk with their healthcare providers and learn more about genetic testing with Ambry Genetics at ambrygen.com .
Reach Out	RESOURCES	<ul style="list-style-type: none"> • Amyloidosis Foundation amyloidosisresearchfoundation.org • National Society of Genetic Counselors nsgc.org • Canadian Association of Genetic Counsellors cagc-accg.ca • Genetic Information Nondiscrimination Act (GINA) ginahelp.org

Please discuss this information with your healthcare providers. The field of genetics is continuously changing, so updates related to your genetic testing results and/or medical management options may be available over time. This information is not meant to replace a discussion with a healthcare providers, and should not be considered or taken as medical advice.