Ambry Genetics®

Understanding Your Carrier NeuropathySelect™ Genetic Test Result

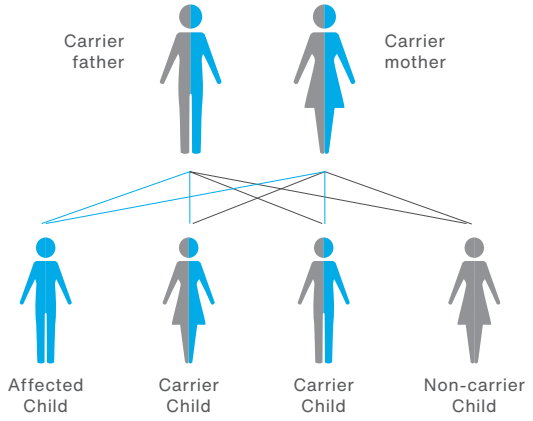
INFORMATION FOR PATIENTS WITH ONE PATHOGENIC MUTATION OR VARIANT THAT IS LIKELY PATHOGENIC FOR A RECESSIVE CONDITION

Result	CARRIER	The result of your genetic testing shows that you have a pathogenic (disease-causing) mutation, or a variant that is likely disease-causing, in one copy of a gene that causes polyneuropathy (peripheral and/or autonomic) or another condition. This means you are a "carrier" of a mutation that is related to polyneuropathy or another condition.
Carrier	DEFINITION	Everyone has two copies of each gene; one gene copy from each parent. A mutation (change in the gene, like a spelling mistake) in both copies of certain genes can cause polyneuropathy or another condition. Carriers have a mutation in one copy of their gene. Carriers do not usually have symptoms but can have children with the condition.
Diagnosis	NO CHANGE	This testing does not change your clinical diagnosis. If you have been diagnosed with polyneuropathy and/or amyloidosis, that remains the same.
Further Testing	DISCUSS	More genetic testing may be right for you. Please talk about this with your healthcare provider.
Medical Management Options	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	PATIENTS WITH A FAMILY HISTORY OF POLYNEUROPATHY AND/OR AMYLOIDOSIS, BUT NO PERSONAL SIGNS	Options for screening and early detection include: physical exams, imaging studies, laboratory tests, biopsies and specialized analysis (electromyography, nerve conduction velocity testing, etc.). Talk to your healthcare providers about whether these options are right for you.
Family Members/ Partner	TESTING	If your partner is also a carrier of a mutation in the same gene, you could have a 1 in 4 (25%) chance to have an affected child in each pregnancy together (see below). Genetic testing for your partner may help you both learn more about this. In addition, your adult blood-related family members may wish to be tested to see if they carry the mutation(s) found in your family.
Next Steps	DISCUSS	Please share this with family members so they can talk with their doctors and learn more.
Reach Out	RESOURCES	• 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

How are Genetic Diseases Inherited?

Individuals who have a mutation in only **one** copy of a gene are "carriers," and do not usually have symptoms. If they have children with someone who is also a carrier for a mutation in the same gene, there is a 1 in 4 (25%) chance for them to have an affected child in each pregnancy together. There is a 2 in 4 (50%) chance for them to have a child who is a carrier (usually without symptoms), and a 1 in 4 (25%) chance for them to have a child who is not affected and is not a carrier.

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.



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