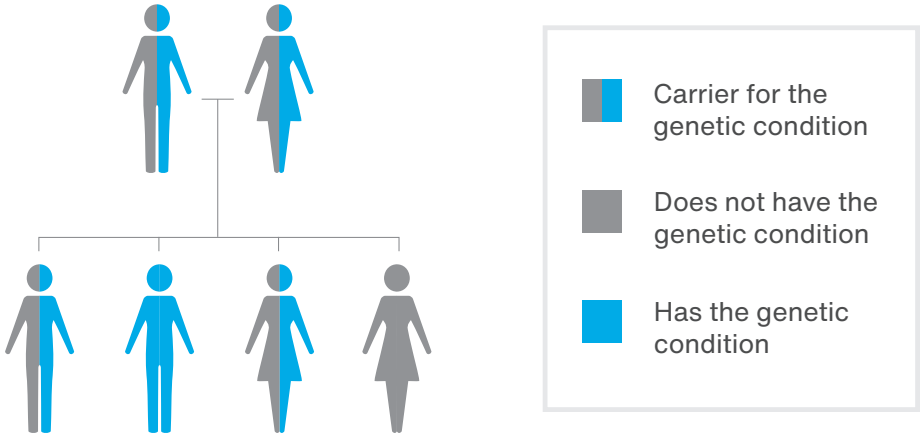
Ambry Genetics®

Understanding Your Carrier Genetic Test Result

INFORMATION FOR PATIENTS WITH ONE PATHOGENIC MUTATION OR LIKELY PATHOGENIC VARIANT

Genes	Genes are instructions for how our bodies work and develop. Everyone has two copies of each gene, one from each parent. Variants (changes) in **both** copies of certain genes can cause specific genetic conditions. Carriers are people who have a change in **only one** copy of their gene. Carriers do not usually have the symptoms of a genetic condition, but can have children with a genetic condition.
Result	The testing found a variant that is either pathogenic (known to be disease-causing) or likely pathogenic (likely to be disease-causing) in a gene known to cause a genetic condition. This means that you are a "carrier" for a genetic condition. It is unlikely that having only one pathogenic or likely pathogenic variant in this gene causes a genetic condition.
Family Members	If a carrier's partner is also a carrier of a pathogenic mutation or likely pathogenic variant in the same gene, there is a 1 in 4 (25%) chance to have a child with a specific genetic condition in each pregnancy. Genetic testing for a carrier's partner may provide more information about this risk. In addition, other adult family members may wish to be tested to see if they carry the pathogenic mutation or likely pathogenic variant found in your family. It is recommended that you share this information with them so they can learn more and discuss with their healthcare providers.
Recessive Inheritance	For certain types of genetic conditions (recessive conditions), people who have a pathogenic mutation or a likely pathogenic variant in only one copy of the gene are "carriers". Usually, carriers do not have any symptoms. If they have children with a partner who is also a carrier of a pathogenic mutation or likely pathogenic variant in the same gene, there is a 1 in 4 (25%) chance for them to have a child with a specific genetic condition in each pregnancy together. There is a 1 in 2 (50%) chance for them to have a child who is a carrier, and a 1 in 4 (25%) chance for them to have a child who is not a carrier and does not have the specific genetic condition.



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 interpreted as medical advice.

One Enterprise, Aliso Viejo, CA 92656 USA Toll Free +1.866.262.7943 Fax +1.949.900.5501 **ambrygen.com**

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