Ambry Genetics®

Understanding Your Carrier Genetic Test Result (Limited Evidence Cancer Gene)

INFORMATION FOR PATIENTS WITH ONE PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

RESULT	Your testing shows that you have one variant that is either pathogenic (known to be disease causing) or likely pathogenic (likely to be disease causing) in an autosomal recessive gene. This type of result is called a "carrier" result. There is currently insufficient evidence to suggest an increased risk for cancer in people who carry only one pathogenic or likely pathogenic variant in this gene.
CARRIERS	Everyone has two copies of each gene. People with only one pathogenic or likely pathogenic variant in this gene are known to be carriers of an autosomal recessive condition, and people with two would be expected to have the autosomal recessive condition. For this reason, your children could be affected with the condition if their other biological parent is also a carrier. When both parents are carriers, the risk of having a child with the condition is 1 in 4, or 25%. It is important that you review your result with a genetic counselor or other healthcare provider so you can learn about the specific autosomal recessive condition related to your result and understand the risks that you and your family members may have.
LIMITED EVIDENCE GENES	Although there is limited evidence to suggest an increased risk for cancer in people who carry one pathogenic or likely pathogenic variant in a limited evidence cancer gene, collecting information about these genes is an ongoing process. It is possible that your result may be better understood in the future. Ambry regularly reviews the data and published evidence about each limited evidence gene, and your healthcare provider will be notified if enough new information becomes available to change the meaning of your result. For this reason, it is recommended that you continue to follow-up with your healthcare provider that ordered your genetic testing.
WHAT YOU CAN DO	Risk management decisions are very personal and depend on many factors. It is important to discuss these options with your healthcare provider and decide on a plan that works for you.
FAMILY	Family members may also have inherited this pathogenic or likely pathogenic variant.. You may wish to share this information with family members, **especially those of childbearing age**, so they can discuss their options with their healthcare provider and decide on a plan that works for them.
RESOURCES	▪ National Society of Genetic Counselors nsgc.org ▪ Canadian Society of Genetic Counsellors cagc-accg.ca

Please discuss this information with your health care provider. The cancer genetics field is continuously evolving, so updates related to your result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a provider and should not be considered or interpreted as medical advice.

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