Ambry Genetics®

Understanding Your Positive Secondary Findings Test Result

INFORMATION FOR PATIENTS WITH A 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 certain genes can cause genetic conditions. These gene changes may be passed down in families or not. Even if there is no history of the specific condition in your family, it can still be caused by a change in a gene.
Exome Sequencing	Exome sequencing is a test designed to look for genetic changes in genes that may be the cause of an existing medical condition. Some genetic tests just look for common mutations, while others may just look for changes in common genes. Exome sequencing analyzes all genes known to cause medical conditions. Because this test looks at all of the genes at once, medically important genetic changes can be found that have nothing to do with your current medical condition. These results are often referred to as “Secondary Findings” or “Incidental Findings”.
Secondary Findings Result	Exome sequencing 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 result was found in a gene that is unrelated to your existing medical condition, but may affect your health in the future.
Management Options	There may be medical management or treatment options related to your positive Secondary Findings result. Management and treatment options vary by condition and other factors. Knowing the genetic cause of your symptoms may also help to avoid some tests or procedures. Talk to your healthcare provider about which management options may be right for you.
Family Members	Many people with a genetic condition are the first person in their family to have it. Often, genetic testing can find a gene change in someone even if the gene change was not found in other family members and was not passed down from a parent. In other families, gene changes can be passed down from parent to child. Talk to your healthcare provider about how the specific genetic condition may run in your family and what this means for the rest of your family. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.

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.

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