

Understanding Your Positive Exome Sequencing Test Result

INFORMATION FOR PATIENTS WITH ONE OR MORE POSITIVE RESULTS

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.
Result	Exome sequencing found one or more variants (or changes) in one or more genes that are known to be associated with your existing medical condition.
Cause	The test result confirms a genetic cause of your medical condition. Sometimes, genetic conditions can put a person at increased risk of other medical problems later in life. Talk with your healthcare provider to learn more about whether additional medical screening may be considered.
Patient for Life	As part of Ambry's Patient for Life program, we keep your results on file. Future genetic discoveries may provide enough information to update your result. We will notify your healthcare provider if any other clinically significant results are identified in the future.
Management Options	Management and treatment options vary by condition and other factors. Knowing the genetic cause of your medical condition 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.