Ambry Genetics®

Understanding Your Uncertain Exome Sequencing Test Result

INFORMATION FOR PATIENTS WITH ONE OR MORE UNCERTAIN 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, but it is not clear if this test result is the cause for your existing medical condition.
Diagnosis	This testing does not change your diagnosis. If you have been diagnosed with a specific condition, that remains the same.
Reclassification	Collecting information about an uncertain result is an ongoing process. It is possible that your result may be better understood in the future. The healthcare provider that ordered your test will be notified if new information becomes available about your uncertain result.
Patient for Life	As part of Ambry's Patient for Life program, we keep your results on file. While we did not find a genetic cause of your medical condition today, future genetic discoveries may provide enough information to update your result. We will notify your healthcare provider if any clinically significant results are identified in the future.
Family Members	Your report will indicate if testing family members may help us learn more about your specific result.
Management Options	Management options vary by condition and other factors. Talk to your healthcare provider about which may be right for you.

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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