

Understanding Your Uncertain Exome or Genome Sequencing Test Result

INFORMATION FOR PATIENTS WITH ONE OR MORE UNCERTAIN RESULTS

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| Chromosomes and Genes | <p>Chromosomes are packages of DNA. They are made up of genes that provide instructions for how our bodies develop. Almost everyone has two copies of each chromosome, one from each parent. Variants in our genes or chromosomes can cause genetic conditions. These genetic variants may be passed down in families or occur for the first time in the person who has a genetic condition. Even if there is no history of the specific condition in your family, it can still be caused by a variant in a person's genes or chromosomes.</p> |
| Exome and Genome Sequencing | <p>Exome and genome sequencing tests are designed to look for genetic variants in genes that may be the cause of an existing medical condition. Exome sequencing is a test that analyzes all genes known to cause medical conditions. Genome sequencing is another test that analyzes all genes known to cause medical conditions; it can also detect rare types of genetic variants that can not be detected by exome sequencing.</p> |
| Result | <p>This testing found one or more variants in one or more genes, but it is not clear if this test result is the cause for your existing medical condition.</p> |
| Diagnosis | <p>This test result does not change your diagnosis. If you have been diagnosed with a specific condition, that remains the same.</p> |
| Reclassification | <p>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.</p> |
| Patient for Life | <p>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.</p> |
| Family Members | <p>Your report will indicate if testing family members may help us learn more about your specific result.</p> |
| Management Options | <p>Management options vary by condition and other factors. Talk to your healthcare provider about which may be right for you.</p> |

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.