

Understanding Your Positive Exome or Genome Sequencing Test Result

INFORMATION FOR PATIENTS WITH ONE OR MORE POSITIVE RESULTS

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 pathogenic or likely pathogenic variants in one or more genes that are known to be associated with your existing medical condition.</p>
Cause	<p>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.</p>
Patient for Life	<p>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.</p>
Management Options	<p>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.</p>
Family Members	<p>Many people with a genetic condition are the first person in their family to have it. Often, genetic testing can find a genetic variant in someone even if the variant was not found in other family members and was not passed down from a parent. In other families, genetic variants 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.</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.