

Understanding Your Positive Genetic 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 be brand new for 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 change in a gene.
Result	The testing 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. Either type of variant should be treated as a positive result.
Cause	The test result confirms a genetic cause of your symptoms. 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.
Management Options	Management 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 inherited from earlier generations. Talk to your healthcare provider about how the specific genetic condition may run in your family based on your test results and family history, 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.