

# Understanding Your Positive Familial Hypercholesterolemia (FH) Genetic Test Result

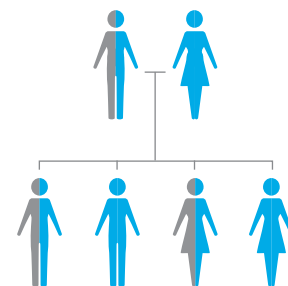
INFORMATION FOR PATIENTS WITH **ONE PATHOGENIC MUTATION OR VARIANT THAT IS LIKELY PATHOGENIC**



Result	<b>POSITIVE</b>	Your testing shows that you have a pathogenic (disease-causing) mutation or a variant that is likely disease-causing in a gene that causes FH. Both of these should be treated as the same type of positive result.
Diagnosis	<b>FH</b>	People with one mutation typically have HeFH, and those with two mutations have HoFH. However, if your cholesterol levels are consistent with HoFH and only one mutation was found, you could actually have HoFH (and the second mutation was unable to be found from the testing).
Gene	<b>DEFINITION</b>	Everyone has two copies of each gene. We get one copy of each gene from each of our parents.
Heterozygous Familial Hypercholesterolemia	<b>HEFH</b>	One mutation (change in the gene, like spelling mistakes) in one copy of any of the genes in this test can cause HeFH. Adults with untreated HeFH often have total serum cholesterol levels >310mg/dL. Children or adolescents with untreated HeFH often have total serum cholesterol levels >230mg/dL.
Homozygous Familial Hypercholesterolemia	<b>HOFH</b>	Two mutations (changes in the gene, like spelling mistakes) in any of the genes in this test can cause HoFH. People with untreated HoFH often have total serum cholesterol levels >500mg/dL.
Management Options	<b>FOR PATIENTS WITH FH</b>	People with HeFH usually respond well to a combination of diet change and medications (e.g. statins and PCSK9 inhibitors). Lipoprotein apheresis treatment, and sometimes even surgery like a liver transplant, might be needed for patients with HoFH. Talk to your doctor about treatment that may be right for you.
Screening Options	<b>FAMILY MEMBERS</b>	Careful monitoring of cholesterol levels is important for all close relatives of patients with FH. Talk to your doctor about which options may be right for you and/or your family.
Next Steps	<b>DISCUSS</b>	Please share this with family members so they can talk with their doctors and learn more. They can now be tested for this same mutation, if they choose to.
Reach Out	<b>RESOURCES</b>	<ul style="list-style-type: none"> <li>National Society of Genetic Counselors <a href="http://nsgc.org">nsgc.org</a></li> <li>Canadian Association of Genetic Counsellors <a href="http://cagc-accg.ca">cagc-accg.ca</a></li> <li>The FH Foundation <a href="http://thefhfoundation.org">thefhfoundation.org</a></li> <li>Genetic Information Nondiscrimination Act (GINA) <a href="http://ginahelp.org">ginahelp.org</a></li> </ul>

## HeFH in the Family

Your close family members (like your parents, brothers, sisters, children) have a 50/50 chance of having the mutation that you carry, and other family members (like your aunts, uncles, cousins) may also have it. Your relatives can now be tested for this same mutation, if they wish.

Please talk with your doctor or genetic counselor about this. The field of genetics is continuously changing, so updates related to your 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 taken as medical advice.



	Has gene mutation causing HeFH		Does not have the gene mutation causing HeFH
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