Ambry Genetics®

Understanding Your Positive Familial Hypercholesterolemia (FH) Genetic Test Result

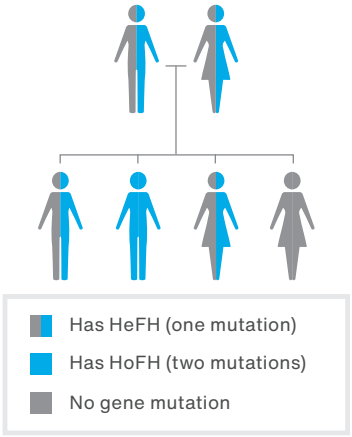
INFORMATION FOR PATIENTS WITH **TWO PATHOGENIC MUTATIONS** OR **VARIANTS, LIKELY PATHOGENIC**

Result	**POSITIVE**	Your testing shows that you have two pathogenic (disease-causing) mutations or variants that are likely pathogenic in a gene that causes FH. Both of these should be treated as the same type of positive result.
Diagnosis	**FH**	People with two mutations typically have have homozygous familial hypercholesterolemia (HoFH), and those with one mutation have heterozygous familial hypercholesterolemia (HeFH). However, if your cholesterol levels are more consistent with HeFH, your mutations could be located on the same copy of the gene, and you could actually have HeFH. Familial testing may be necessary to determine whether mutations are on the same or different copies of the gene. High cholesterol levels over time can cause an increased risk for heart disease, but treatment can help reduce risk.
Gene	**DEFINITION**	Genes are instructions for how our bodies work and develop. Everyone has two copies of each gene. We get one copy of each gene from each of our parents. Mutations (changes in the gene, like spelling mistakes) in any of the genes in this test can cause FH.
Heterozygous Familial Hypercholesterolemia	**HeFH**	Mutations in one copy of any of the genes in this test can cause HeFH. Adults with untreated HeFH often have LDL cholesterol levels of >190mg/dL. Children or adolescents with untreated HeFH often have LDL cholesterol levels of >160mg/dL.
Homozygous Familial Hypercholesterolemia	**HoFH**	Two mutations in any of the genes in this test can cause HoFH. People with untreated HoFH often have LDL cholesterol levels of >400mg/dL.
Management Options	**FOR PATIENTS WITH HOFH**	People with HoFH may respond to a combination of diet change and medications (e.g. statins and PCSK9 inhibitors). Additional medications, lipoprotein apheresis treatment, and sometimes even surgery like a liver transplant, might be needed. Talk to your doctor about treatment that may be right for you.
Screening Options	**FAMILY MEMBERS**	Careful monitoring of cholesterol levels is important for all close relatives of patients with HoFH. Monitoring should begin in childhood for many families. Talk to your doctor about which options may be right for you and/or your family.
Next Steps	**DISCUSS**	Please share this with family members so they can talk with their doctors and learn more. They can now be tested for these same mutations, if they choose to.
Reach Out	**RESOURCES**	National Society of Genetic Counselors nsgc.org Canadian Association of Genetic Counsellors cagc-accg.ca The FH Foundation thefhfoundation.org Genetic Information Nondiscrimination Act (GINA) ginahelp.org

FH in the Family

Your close relatives (like your parents, brothers, sisters, children) have at least a 3/4 chance of having at least one of the mutations that you carry, and other family members (like your aunts, uncles, cousins) may also have it. Your relatives can now be tested for these same mutations, if they wish.

Please discuss this information with your healthcare provider. 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 interpreted as medical advice.



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