Ambry Genetics®

Understanding Your Positive Familial Hypercholesterolemia (FH) Genetic Test Result

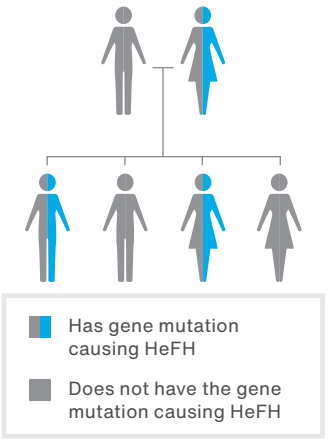
INFORMATION FOR PATIENTS WITH **ONE PATHOGENIC MUTATION** OR **VARIANT, LIKELY PATHOGENIC**

Result	POSITIVE	Your testing shows that you have a pathogenic (disease-causing) mutation or a variant that is 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 one mutation typically have heterozygous familial hypercholesterolemia (HeFH), and those with two mutations have homozygous familial hypercholesterolemia (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	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	One mutation 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 FH	People with HeFH usually respond well 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 for patients with HoFH. 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 FH. 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 this same mutation, 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 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 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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