Ambry Genetics
A Konica Minolta Company

Understanding Your Positive Familial Hypercholesterolemia (FH) Genetic Test Result

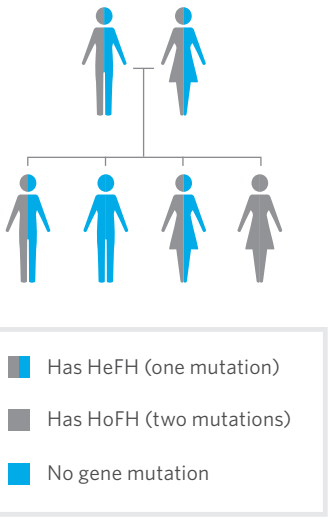
INFORMATION FOR PATIENTS WITH TWO **PATHOGENIC MUTATIONS** OR **VARIANTS THAT ARE LIKELY PATHOGENIC**

Result	**POSITIVE**	Your testing shows that you have two pathogenic (disease-causing) mutations or variants that are likely disease-causing in a gene that causes FH. Both of these should be treated as the same type of positive result.
Diagnosis	**FH**	This result confirms the diagnosis of FH. This means that high cholesterol runs in your family and requires close monitoring and treatment.
Gene	**DEFINITION**	Everyone has two copies of each gene. We get one copy of each gene from each of our parents. Two mutations (changes in the gene, like spelling mistakes) in any of the genes in this test can cause what is called homozygous FH (HoFH).
Homozygous Familial Hypercholesterolemia	**HOFH**	People with untreated HoFH often have total serum cholesterol levels >500mg/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). 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. 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

HoFH 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 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.



MKT-0346_v2 | 08.24.20
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