

Understanding Your Negative Familial Hypercholesterolemia (FH) Genetic Test Result

INFORMATION FOR PATIENTS

Result	NEGATIVE	Your testing did not find any disease-causing gene changes (or mutations). Only 50-60% of people with FH have a mutation in one of the genes in this test. You may have a mutation in a gene that was not included in this test. If someone in your family has a specific mutation in one of the genes in this test, it is likely that you are not at increased risk for FH.
Diagnosis	NO CHANGE	This testing does not rule out the diagnosis of FH for you. If you have been diagnosed with FH based on your cholesterol levels and/or family history, that remains the same.
Management Options	FOR PATIENTS WITH FH	People with the most common form of FH 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 those with more extreme cholesterol levels that occur less often.
Screening Options	FOR PATIENTS WITH A FAMILY HISTORY OF FH	If you have a family history of FH, and no mutation has been found in your family, it is likely that you are still at increased risk for FH. Talk with your doctor about regular cholesterol screening.
Next Steps	DISCUSS	Please share this with family members so they can talk with their doctors and learn more.
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

Even though your genetic testing was negative, FH can run in families. All close family members of someone with FH (like parents, brothers, sisters, children) should talk with their doctor about cholesterol screening.

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.

