Familial Hypercholesterolemia

Familial hypercholesterolemia (FH) is a genetic disorder that increases one’s risk for high cholesterol. High cholesterol is a “silent” disease that does not typically result in any noticeable symptoms. However, this condition can lead to coronary artery disease (CAD) which increases the risk for serious health complications such as heart attack and stroke. Other common features of FH include growths on tendons (Achilles tendon, hands, fingers) called xanthomas, cholesterol deposits under the skin of the eye, and a gray-colored ring on the cornea of the eye. Individuals with untreated FH may encounter cardiovascular disease before the age of 50. It is important to regularly monitor and manage cholesterol levels when FH is diagnosed.

WHAT CAUSES FAMILIAL HYPERCHOLESTEROLEMIA?

FH is caused by pathogenic (disease-causing) variants in genes that affect the production and function of low-density lipoproteins (LDLs). These variants hinder the body’s ability to remove LDL cholesterol from the blood. As a result, there is an excess amount of cholesterol that circulates through the bloodstream which can lead to symptoms described above.

WHO IS THIS TEST FOR?

This panel may be appropriate for individuals with a personal or family history of FH or high levels of cholesterol, particularly young patients with above-average levels. Additionally, those who have a personal or family history of early-onset coronary artery disease, stroke and/or peripheral vascular disease could benefit from testing. Individuals who exhibit common characteristics of FH may also be a candidate for testing.
BENEFITS OF GENETIC TESTING

Genetic testing for familial hypercholesterolemia can:

- Establish or confirm the appropriate diagnosis
- Identify risks for additional health-related symptoms
- Assist in modifying lifestyle changes, including diet and exercise
- Result in more personalized treatment and symptom management
- Inform family members about their own risk factors
- Provide options for family planning

TEST SPECIFICATIONS

**Acceptable Sample Requirements**

- Blood, two 4-mL EDTA tubes (lavender top)
- Extracted DNA, 3 μg in EB buffer
- Buccal swab or saliva

**Turnaround Time** 3–5 weeks

**Coverage** ≥96% at 20x

**Reporting**

VUS, likely pathogenic, and pathogenic variants

**Customization**

Customizable Gene list, VUS opt-out

RELATED TESTS

Visit Fulgent website for most up-to-date list

- Familial Hypercholesterolemia NGS Panel
- Hyperlipidemia NGS Panel
- Comprehensive Cardiovascular NGS Panel

GET CONNECTED

- The FH Foundation - thefhfoundation.org
- National Organization for Rare Disorders - rarediseases.org/rare-diseases/familial-hypercholesterolemia
- American Heart Association - heart.org
- Global Genes - globalgenes.org

REFERENCES

- medlineplus.gov/genetics/condition/familial-hypercholesterolemia
- sciencedirect.com/science/article/abs/pii/S0021915003004878
- ncbi.nlm.nih.gov/books/NBK174884
A Patient’s Guide to Genetic Testing

What does a genetic test check for?

Genetic testing checks the order of one’s DNA sequence (coded by the letters A, T, G, C) in specific genes linked to genetic conditions. Letters that were added, missing, or changed, are known as variants and can sometimes be harmful to one’s health, increasing the risk for a genetic condition.

What are the potential results?

There are three possible results from genetic testing:

Positive
A pathogenic/likely pathogenic variant is detected in one’s DNA. This type of variant is known to increase one’s risk of a genetic condition. Identifying the specific gene involved can help confirm a diagnosis, inform screening and management, and reveal risk factors for an individual and/or their family.

Negative
No variation known to be associated with a genetic condition was detected in one’s DNA. While a result may not show an increased risk for the condition(s) tested for, one can still be at risk for disease, especially if there is a family history.

Variant of Uncertain Significance (VUS)
A variant was detected in one’s DNA, however, not enough information is known about this variant to determine whether or not it is known cause the condition(s) tested for. More research is needed to better understand this variant.

What about family members?

Children, siblings, and parents of individuals who have a variant(s) identified in genetic testing could carry the same variant(s) and benefit from testing. Regardless of whether or not a variant was identified, individuals can still be at an increased risk for a genetic condition, especially with a family history.

Do genetic test results affect health insurance or employment?

No, the Genetic Information Nondiscrimination Act (GINA) was signed into law in 2008. It protects individuals from discrimination by an employer or a health insurance company based on genetic testing results and genetic information. GINA does not protect against life and disability insurance discrimination. For more information on GINA, go to www.ginahelp.org.

Where can I learn more?

Medline Plus/Genetics Home Reference - medlineplus.gov/genetics/understanding
National Society of Genetic Counselors - nsgc.org
Fulgent Genetics - fulgentgenetics.com/products/carrierscreening/learning.html