Familial Hypercholesterolemia

Familial hypercholesterolemia (FH) is a genetic condition associated with very high levels of cholesterol in the blood, specifically low-density lipoprotein (LDL), or "bad" cholesterol. High cholesterol due to FH increases the risk for early cardiovascular disease, which can lead to a heart attack. This test includes 24 genetic variants linked to FH.

zero_variants, you **do not have** the genetic variants we tested.

However, this test does not include the majority of variants linked to FH, so you could still have a variant not included in this test.

If you have a personal or family history of high cholesterol or early heart disease, you should talk to a healthcare professional about cholesterol screening options.

How To Use This Test

This test does not diagnose familial hypercholesterolemia or any other health conditions.

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Review the Genetic Health Risk tutorial
See Scientific Details
See Frequently Asked Questions

Intended Uses

- Tests for one variant in the APOB gene and 23 variants in the LDLR gene. These variants are linked to having very high LDL cholesterol levels, which is associated with an increased risk for heart disease.

Limitations

- Does **not** test for all possible variants in the APOB and LDLR genes. More than 1,000 variants in these genes are linked to FH. Only 24 of those variants are included in this test.
- Does **not** test for variants in other genes linked to FH and other forms of inherited high cholesterol.
- Does **not** report if someone has two copies of a tested variant.

Important Ethnicities

- The majority of the variants included in this test are most commonly found in people of **European** and **Lebanese** descent, as well as in the **Old Order Amish**. In addition, some of these variants have also been found in other ethnicities.
- However, more than 1,000 variants have been linked to FH in people of European descent, as well as in people of other ethnicities. This test does not include the majority of those variants.

You **do not have** the genetic variants we tested linked to FH.

People without these variants are still at risk for high cholesterol and heart disease due to other factors, including lifestyle, family history, and other genetic factors.

There is still a chance of having a variant linked to FH.

More than 1,000 variants are linked to FH and are known to be associated with very high LDL cholesterol levels. This test only includes 24 of those variants.

See Scientific Details

In the U.S., about 30% of people who are 20 years and older have high LDL cholesterol levels.

Most cases of high LDL cholesterol are not caused by the genetic variants in this report. Your risk is influenced by many other factors, including lifestyle, family history, and other genetic factors.

If you have a personal or family history of high cholesterol or early heart disease, it's important to talk to a healthcare professional about your cholesterol screening options.

A healthcare professional can help you assess your overall heart disease risk. Learn more from the American Heart Association.

Lifestyle and other factors can also influence the chances of developing heart disease.

Consult with a healthcare professional before making any major lifestyle changes.

Age

In general, the risk for heart disease increases as a person gets older. With age, blood vessels become less flexible, which can impede blood flow to the heart. Also, deposits called plaques can build up over time along blood vessels and restrict blood flow to the heart.

See Scientific Details for more information

Family history
Age
High blood pressure
Smoking
Other genetic factors
Lifestyle

About Familial Hypercholesterolemia

Also known as: FH, familial hypercholesterolæmia, autosomal dominant hypercholesterolemia, hyperlipoproteinemia type IIA

When it develops

Because it is a genetic condition, FH is present at birth, meaning most people with this condition have high LDL cholesterol levels from a young age. Since many people with FH show no physical symptoms, this condition is typically diagnosed with a blood test for cholesterol. However, some people with FH may not be diagnosed until after experiencing symptoms related to early heart disease, including chest pain or heart attack.

Typical signs and symptoms

- Elevated total and LDL cholesterol levels
- Heart disease, heart attack, or chest pain
- In some cases, cholesterol deposits may build up in the skin or tendons (xanthomas), under the skin in the eyelids (xanthelasmas), or around the cornea of the eye (corneal arcus)

How common is this condition?

FH affects people of all ethnicities. About 1 in 200 to 1 in 250 people in the world have FH.

How it's treated

Early and active treatment of FH can substantially reduce the risk for heart disease. FH treatment focuses on lowering LDL cholesterol levels, and FH is usually treated with cholesterol-lowering medications. Lifestyle modifications, including diet, exercise, and weight control can help lower LDL cholesterol levels. But these changes are generally not enough to effectively manage the condition. In extreme cases of FH, LDL-apheresis, a procedure that filters cholesterol out of the blood, can be used when other treatments have failed.

Read more at: National Human Genome Research Institute GeneReviews Genetics Home Reference FH Foundation

Learn more about FH.

See our Frequently Asked Questions for more information.

FAQs

If you have a personal or family history of high cholesterol or early heart disease, consult with a healthcare professional.

Print report

Learn more about cholesterol screening to help you and your doctor create a screening plan that's right for you.

Learn more

Scientific Details

Rs4477212 (Intergenic) (rs4477212) is a genetic variation associated with very large levels of cholesterol in blood. Cholesterol is a fatty substance that is found in the bloodstream and in cells throughout the body. It is needed to make cell membranes and other hormones and vitamins, which are made in small amounts. This locus includes 20 genetic variants related to this.

FH is linked to variants in multiple genes.

This report includes variants in the FH gene and 19 other genes related to this condition.

LDLR | APOB | LDLRAP1

Chromosome 2

The APOB gene provides instructions for making a protein called apolipoprotein B. This protein plays a key role in the transport of fat molecules such as cholesterol on the surface of cells, particularly in the liver. Cancer risk is also associated with the APOB gene. Rs693 is a common variant in the APOB gene that causes a DNA mutation resulting in the amino acid from Arginine to Glutamine.

Read more at Genetics Home Reference

You do not have the genetic variants we tested.

Genotype Reported	Risk Allele Reported	Allele Frequency	Genotype Frequency	Allele Frequency	Genotype Frequency	Allele Frequency	Genotype Frequency

View All Tested Markers

RS5370
Gene: EDN1
Rs5370 - G/T
Genotype: GG/TGG

Allele Frequency: G
Typical Genotype: GG
Allele Frequency: T
Typical Genotype: TT

Biological explanation
The gene tested is a change from G to A in the 2 Kb DNA sequence of the EDN1 gene. It results in a change from Glycine to Glutamic acid in the protein. Endothelin-1 is a potent vasoconstrictor and is only found in the bloodstream. This variant is also known as G201R.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

LTB4DR-R
Gene: LTB4DR
Rs1805192 - A/G
Genotype: AG/GG

Allele Frequency: A
Typical Genotype: AA
Allele Frequency: G
Typical Genotype: GG

Biological explanation
The gene tested is a change from A to G in the 2 Kb DNA sequence of the LTB4DR gene. It results in a change of the protein that is not fully understood.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

MFN2
Gene: MFN2
Rs2349 - T/C
Genotype: TT/CT

Allele Frequency: T
Typical Genotype: TT
Allele Frequency: C
Typical Genotype: CC

Biological explanation
The gene tested is a change from T to C in the 2 Kb DNA sequence of the MFN2 gene. It results in a change of the protein that is not fully understood. This variant is also known as R707W.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

ZP90
Gene: ZP3
Rs2699313 - A/G
Genotype: AG/GG

Allele Frequency: A
Typical Genotype: AA
Allele Frequency: G
Typical Genotype: GG

Biological explanation
The gene tested is a change from A to G in the 2 Kb DNA sequence of the ZP3 gene. It results in a change of the protein that is not fully understood. This variant is also known as R388H.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

EDN1R
Gene: EDNRA
Rs5335 - G/C
Genotype: GG/CG

Allele Frequency: G
Typical Genotype: GG
Allele Frequency: C
Typical Genotype: CC

Biological explanation
The gene tested is a change from G to A in the 2 Kb DNA sequence of the EDNRA gene. It results in a change of the protein that is not fully understood. This variant is also known as G167S.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

EDN1
Gene: EDN1
Rs1800541 - T/C
Genotype: TT/CT

Allele Frequency: T
Typical Genotype: TT
Allele Frequency: C
Typical Genotype: CC

Biological explanation
The gene tested is a change from T to C in the 2 Kb DNA sequence of the EDN1 gene. It results in a conservative protein change, which may lead to a change in protein function. This variant is also known as G289S.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

S2B4
Gene: SCARB2
Rs6480370 - C/T
Genotype: CC/CT

Allele Frequency: C
Typical Genotype: CC
Allele Frequency: T
Typical Genotype: TT

Biological explanation
The gene tested is a change from C to T in the 2 Kb DNA sequence of the SCARB2 gene. It results in a conservative protein change, which may lead to a change in protein function. This variant is also known as R183C.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

GDF5
Gene: GDF5
Rs143383 - G/T
Genotype: GG/TGG

Allele Frequency: G
Typical Genotype: GG
Allele Frequency: T
Typical Genotype: TT

Biological explanation
The gene tested is a change from G to A in the 2 Kb DNA sequence of the GDF5 gene. It results in reduced protein activity, which may lead to a change in protein function. This variant is also known as R53H.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

EDNRB
Gene: EDNRB
Rs5351 - G/A
Genotype: GG/AG

Allele Frequency: G
Typical Genotype: GG
Allele Frequency: A
Typical Genotype: AA

Biological explanation
The gene tested is a change from G to A in the 2 Kb DNA sequence of the EDNRB gene. It results in reduced protein activity, which may lead to a change in protein function. This variant is also known as G167S.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

LGR4
Gene: LGR4
Rs7938748 - G/A
Genotype: GG/AG

Allele Frequency: G
Typical Genotype: GG
Allele Frequency: A
Typical Genotype: AA

Biological explanation
The gene tested is a change from G to A in the 2 Kb DNA sequence of the LGR4 gene. It results in a conservative protein change, which may lead to a change in protein function. This variant is also known as R265H.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

MCT3
Gene: SLC16A3
Rs1049434 - A/G
Genotype: AA/AG

Allele Frequency: A
Typical Genotype: AA
Allele Frequency: G
Typical Genotype: GG

Biological explanation
The gene tested is a change from A to G in the 2 Kb DNA sequence of the SLC16A3 gene. It results in a conservative protein change, which may lead to a change in protein function. This variant is also known as R156Q.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

ZP90
Gene: ZP3
Rs2699314 - G/A
Genotype: GG/AG

Allele Frequency: G
Typical Genotype: GG
Allele Frequency: A
Typical Genotype: AA

Biological explanation
The gene tested is a change from G to A in the 2 Kb DNA sequence of the ZP3 gene. It results in a change of the protein that is not fully understood. This variant is also known as R407C.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

EDN1R
Gene: EDNRA
Rs5333 - G/A
Genotype: GG/AG

Allele Frequency: G
Typical Genotype: GG
Allele Frequency: A
Typical Genotype: AA

Biological explanation
The gene tested is a change from G to A in the 2 Kb DNA sequence of the EDNRA gene. It results in a change of the protein that is not fully understood. This variant is also known as R347H.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

CETP
Gene: CETP
Rs1800775 - C/A
Genotype: CC/CA

Allele Frequency: C
Typical Genotype: CC
Allele Frequency: A
Typical Genotype: AA

Biological explanation
The gene tested is a change from C to A in the 2 Kb DNA sequence of the CETP gene. It results in a change of the protein that is not fully understood. This variant is also known as D497 and I405V.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

PAR1
Gene: F2RL1
Rs2227537 - C/T
Genotype: CC/CT

Allele Frequency: C
Typical Genotype: CC
Allele Frequency: T
Typical Genotype: TT

Biological explanation
The gene tested is a change from C to T in the 2 Kb DNA sequence of the F2RL1 gene. It results in the stop codon, or the protein, is not produced. This variant is also known as R389*. This variant is also known as R389*.

- **Typical vs. variant DNA sequences:**
  Typical Sequence: ... Variant Sequence: ...
- **Percent of Chromosomes carrying each variant:**

Variant 1	
European	0.00%
African American	0.00%
Ashkenazi Jewish	0.00%
East Asian	0.00%
South Asian	0.00%
Middle Eastern	0.00%

References: 16, 45, 199, 200 | Citations

Test Interpretation

This report provides risk estimates for heart disease based on LDL cholesterol levels and the presence or absence of variants linked to FH. In most cases, these estimates represent a general risk for individuals with any variant linked to FH, not specific risk estimates associated with the 24 variants in this report.

For reference, LDL cholesterol ranges for adults are provided based on the classification by the National Heart, Lung, and Blood Institute.

Health Risk Estimates

Risk estimates are based on clinical studies that identify an association between a genotype and a health condition.

Numerical risk estimates are not available for people who have more than one variant linked to FH. An interpretation of "increased risk" is provided to people with these results, because it is likely that their risk is at least as high as the risk for people with just one variant.

Consider talking to a healthcare professional if you have any concerns about your results.

References [68, 86, 120]

Odds ratios | Likelihood ratios | LDL cholesterol ranges

An "odds ratio" estimates how strongly a genetic variant is associated with a condition. An odds ratio greater than 1 means that people with that variant are more likely to develop the condition, while an odds ratio less than 1 means that people with that variant are less likely to develop the condition, compared to people without the variant.

The table below shows the odds ratios for heart disease based on LDL cholesterol levels and the presence or absence of variants linked to FH.

LDL cholesterol level (mg/dL)	Without FH variant	With FH variant
<130	1	2.2
≥130-160	1.8	3.8
≥160-190	2.9	5.6
≥190-220	5.2	17.0
≥220	7.7	25.8

Other Factors

The genetic variants in this report are linked to FH and associated with an increased risk for heart disease. However, other factors besides the genetic variants in this report can influence your chances of developing heart disease.

This is not a complete list of other factors.

People with multiple risk factors may have a higher risk of developing heart disease. Overall risk depends on family history and other factors.

Keep in mind that in general, a healthy lifestyle (eating a healthy diet, maintaining a healthy weight, and exercising regularly) has been associated with many health benefits.

Consult with a healthcare professional before making any major lifestyle changes.

Other Factors	References
Family history First-degree relatives of an individual with early heart disease (younger than 55 years in men and younger than 65 years in women) have a higher chance of developing heart disease themselves. This may be primarily explained by genetic factors, but could also be explained by family members sharing a similar lifestyle and environment.	[7, 80, 87, 92]
Age In general, the risk for heart disease increases as a person gets older. With age, blood vessels become less flexible, which can impede blood flow to the heart. Also, deposits called plaques can build up over time along blood vessels and restrict blood flow to the heart. Men who are 45 or older and women who are 55 or older have a greater risk of developing heart disease.	[7, 31, 85, 87, 100]
Smoking Smoking increases the risk for heart disease. The chemicals found in tobacco smoke can damage the inner lining of blood vessel walls, increase heart rate, and cause irregularities in the timing of heartbeats, making a person more susceptible to having heart disease. Smoking also interacts with other risk factors for heart disease, including high blood pressure, high cholesterol, and obesity, to increase the risk for heart disease even more. Exposure to secondhand smoke, including smoke from a burning cigarette and smoke exhaled by smokers, also increases the risk for heart disease since it contains many of the same harmful substances found in the smoke inhaled by smokers.	[9, 87, 94, 108, 134]
Sex Men are generally at higher risk than women for developing heart disease earlier in life, but women's risk increases after menopause. Certain risk factors around the time of menopause, including weight gain and high blood pressure, increase a woman's risk for heart disease.	[7, 81, 87, 100]
High blood pressure Chronic high blood pressure increases the risk for heart disease. High blood pressure puts excess strain and increased workload on the heart and blood vessels, which can damage them over time. Damaged blood vessels are more vulnerable to plaque deposits, which can limit blood flow to the heart. This can lead to chest pain or heart attack.	[6, 50, 87, 93, 100, 108, 123, 134]
Diet A diet high in saturated fat, trans fat, dietary cholesterol, and added sugars is associated with increased heart disease risk. In contrast, studies have found foods rich in soluble fiber such as fruits, vegetables, and legumes are associated with reduced heart disease risk.	[7, 35, 41, 45, 50, 87, 134]
Physical activity In general, being physically active can help reduce the risk for heart disease by lowering a person's LDL cholesterol levels and blood pressure. Current U.S. guidelines recommend that adults should get at least 150 minutes a week of moderate-intensity activity or 75 minutes a week of vigorous-intensity activity for a healthier life. Learn more from the American Heart Association.	[5, 7, 35, 45, 50, 87, 121, 134]
Obesity Being overweight increases a person's risk of developing heart disease. In addition, excess weight typically worsens other risk factors for heart disease, including high blood pressure and diabetes.	[36, 45, 50, 87, 108, 134]
Other genetic factors Studies have identified more than 1,000 variants linked to FH and associated with an increased risk for heart disease. These variants are in the LDLR and APOB genes, as well as other genes including PCSK9. This report includes one variant in the APOB gene and 23 variants in the LDLR gene. In addition, there are other genetic variants that may impact LDL cholesterol levels. However, most of these variants are either rare or have only a small effect on risk.	[30, 68, 75]
Diabetes Having diabetes, especially type 2 diabetes, increases a person's risk for heart disease. People with diabetes have a two to four times greater risk of dying from heart disease than people without diabetes. The risk is greater in women than in men. High blood sugar can damage blood vessels, so it is important to control blood sugar levels and manage other modifiable risk factors, such as obesity and physical activity, to minimize risk for heart disease.	[7, 44, 50, 62, 98, 134]
High lipoprotein(a) levels Lipoprotein(a), also called Lp(a), is a lipoprotein similar to LDL. High Lp(a) in the blood is associated with an increased risk for heart disease. Lp(a) may deposit in blood vessels and contribute to plaque buildup, which may reduce blood supply to the heart. Lp(a) levels are primarily determined by inherited genetic factors.	[19, 65]

Cardiovascular Disease Risk Assessment and Cholesterol Screening Guidelines

Cholesterol screening can help you take action to reduce your risk for heart disease. The guidelines below may help you and your doctor create a screening plan that's right for you.

The American Heart Association and American College of Cardiology recommend that adults 20 years of age and older, who are free from cardiovascular disease, should have their cholesterol checked every four to six years and blood pressure checked every one to two years. Additionally, guidelines recommend that people should talk to their healthcare provider about heart disease risk factors, including diet, physical activity, smoking, and weight management. Learn more from the American Heart Association.

Consider talking to a doctor if you have a personal or family history of high cholesterol or heart disease. Your doctor may have specific screening recommendations for you.

Note that guidelines from different healthcare professional organizations may differ slightly in their recommendations.

Keep in mind that you could still have a variant in the APOB or LDLR genes or other genes not included in this report that could affect your risk for heart disease. In that case, different screening and actions may be recommended. Consult with a healthcare professional to learn more.

Test Details

Indications for Use

The 23andMe PGS Genetic Health Risk Report for Familial Hypercholesterolemia is indicated for reporting of one variant in the APOB gene and 23 variants in the LDLR gene. This report describes if a person's genetic result is associated with an increased risk of having very high LDL cholesterol, which can lead to heart disease. This test does not describe a person's overall risk of developing heart disease, and the absence of a variant tested does not rule out the presence of other variants that may be linked to familial hypercholesterolemia. The majority of the variants in this report are found in and have been most studied in people of European and Lebanese descent, as well as in the Old Order Amish.

Special Considerations

- Genetic testing for FH in the general population is not currently recommended by any healthcare professional organizations.
- However, the U.S. CDC recommends that screening using cholesterol testing with or without DNA analysis should be conducted on relatives of people with familial high cholesterol.
- Heart disease risk associated with FH variants varies from person to person. Overall risk depends on family history and other factors.

Test Performance Summary

Clinical Performance [1, 2, 10, 13, 21, 49, 52, 64, 68, 70, 82, 105, 117, 122, 127]

The variants included in this report represent a small subset of all those linked to FH. Over 1,000 variants have been linked to FH. The 24 variants included in this test are linked to having very high LDL cholesterol, which is associated with an increased risk for heart disease. About 1 in 50 people with high LDL cholesterol have FH.

- Approximately 30-35% of people of European descent with a genetic variant linked to FH have one of the 24 variants included in this test.
- Approximately 15-20% of people of Hispanic/Latino or East Asian descent with a genetic variant linked to FH have one of the 24 variants included in this test.
- For people of Lebanese descent, the test covers about 80% of people who have a variant linked to FH.
- About 10% of the Old Order Amish have the APOB R3527Q variant linked to FH.

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.* The absence of a variant tested does not rule out the presence of other genetic variants that are linked to FH.
- This test does not diagnose any health conditions.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the Package Insert for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

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4. Alonso R et al. (2014). "Lipoprotein(a) levels in familial hypercholesterolemia: an important predictor of cardiovascular disease independent of the type of LDL receptor mutation." J Am Coll Cardiol. 63(19):1982-9.*
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9. Barnoya J et al. (2005). "Cardiovascular effects of secondhand smoke: nearly as large as smoking." Circulation. 111(20):2684-98.*
10. Bañares VG et al. (2017). "Preliminary spectrum of genetic variants in familial hypercholesterolemia in Argentina." J Clin Lipidol. 11(2):524-531.*

See all references

Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
[redacted]	Familial Hypercholesterolemia report created.

Familial Hypercholesterolemia

Frequently Asked Questions

Familial hypercholesterolemia (FH) is a genetic condition associated with very high levels of cholesterol in the blood, specifically low-density lipoprotein (LDL), or "bad" cholesterol. High cholesterol due to FH increases the risk for early cardiovascular disease, which can lead to a heart attack. This test includes 24 genetic variants linked to FH.

Familial Hypercholesterolemia

How is familial hypercholesterolemia (FH) different from typical high cholesterol?

In the general population, LDL cholesterol levels tend to start increasing in middle age and are often influenced by diet and other lifestyle choices. In contrast, FH is a genetic condition in which LDL levels can be elevated from birth and may also continue to rise throughout life. In both people with and without FH, high LDL cholesterol increases the risk for heart disease, but in people with FH the risk is higher because their cumulative exposure to elevated LDL is longer.

In addition, it's worth noting that because FH is a genetic condition, it is passed down through families. This means that people with a genetic variant can pass down the variant to their children. Also, it means that people with a variant have inherited that variant from one of their parents.

Is this answer helpful? Yes No

What does this test do?

This test looks for 24 specific variants linked to FH, one in the APOB gene and 23 in the LDLR gene.

People with the genetic variants included in this test are likely to have elevated LDL cholesterol levels, which is associated with an increased risk for heart disease.

This test does not include all possible genetic variants that may increase your risk for FH.

Is this answer helpful? Yes No

What does this test **not** do?

This test does not diagnose FH. Only a healthcare professional can do that.

This test does not measure cholesterol or tell you if you have elevated LDL cholesterol levels.

This test does not include all possible genetic variants linked to FH. Over 1,000 variants are linked to FH, and this test only includes 24 of them.

This test does not provide a complete assessment of risk for high cholesterol or heart disease.

Is this answer helpful? Yes No

The report says the variants included in this test are most common in certain ethnicities. What does that mean?

The majority of the variants we tested are most common in people of European or Lebanese descent, and in the Old Order Amish. In addition, many of the variants have also been observed in people of other ethnicities.

However, more than 1,000 variants have been linked to FH in people of European descent, as well as in people of other ethnicities. This test does not include the majority of those variants.

The risk estimates for elevated LDL cholesterol and early heart disease associated with these variants are expected to be the same regardless of ethnicity. See Scientific Details for more information.

Is this answer helpful? Yes No

Where can I learn more about FH, support groups, and other resources?

You can learn more about FH from the following resources:

- FH Foundation
- The Foundation of National Lipid Association
- American Heart Association
- National Organization for Rare Disorders

If you have questions about your results or how they might affect you or your family, a genetic counselor may be able to help. Learn more about genetic counseling.

Is this answer helpful? Yes No

My report says **zero variants** were detected. What does this mean?

This means you do not have any of the genetic variants we tested. However, you could still have another genetic variant not included in this test.

More than 1,000 variants have been linked to FH, which is associated with an increased risk for developing early heart disease. This report only includes 24 of those variants.

Other factors can also affect your risk of developing heart disease, even if you do not have any of the genetic factors. Learn more about other factors.

Regardless of your genetic result, if you have a family history of high LDL cholesterol or early heart disease, it's important to talk to a healthcare professional about cholesterol screening.

Is this answer helpful? Yes No

My report says **zero variants** were detected. Does this mean I'm not at risk of having high LDL cholesterol?

No. People with zero variants detected still have a risk of having high LDL cholesterol levels.

More than 1,000 variants have been linked to FH, which is associated with an increased risk for developing early heart disease. This report only includes 24 of those variants.

In addition, lifestyle and environment, as well as other factors, including age and family history, can influence your LDL cholesterol levels and overall risk for heart disease. Learn more about other factors.

Talk to a healthcare professional about cholesterol screening options. This is especially important if you have a family history of high cholesterol or heart disease.

Is this answer helpful? Yes No

My report says **zero variants** were detected, but I have a personal or family history of high cholesterol or early heart disease. What does this mean for me?

People with a family history of high cholesterol or early heart disease have a higher risk of having high cholesterol themselves. Even though you do not have any of the genetic variants we tested, you could still have a variant that is not included in this test. More than 1,000 variants in the LDLR and APOB genes as well as other genes have been linked to FH.

In addition, non-genetic factors, including lifestyle and environmental factors, may also influence your LDL cholesterol levels and overall risk for heart disease. Learn more about other factors.

It is important to discuss your personal or family history of high cholesterol or early heart disease with a healthcare professional, who can help you determine if additional cholesterol screening is appropriate.

Is this answer helpful? Yes No

My report says **zero variants** were detected. What are some things I could do?

Your genetic result means you do not have the genetic variants we tested. However, because there are more than 1,000 genetic variants linked to FH, your result doesn't give you much new information about your risk of having very high LDL cholesterol levels related to FH and the associated risk for heart disease.

There are many other genetic and non-genetic factors that can affect your risk, which this test does not take into account. Learn more about other factors.

It is important to continue with any cholesterol screenings your healthcare provider recommends. The American Heart Association and American College of Cardiology recommend that people in the general population should start regular screening at age 20. Learn more from the American Heart Association.

If you have a family history of high cholesterol or early heart disease, consider discussing your result with a healthcare professional to learn more about options for cholesterol screening and assessment of heart disease risk factors.

Talk to a healthcare professional if:

- You have a personal or family history of high cholesterol or early heart disease.
- You think you might have high cholesterol.
- You have questions about other risk factors you may have.

Is this answer helpful? Yes No

Have more questions? Check out our Customer Care Help Center.