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Hereditary Hemochromatosis (HFE-Related)

Hereditary hemochromatosis is a genetic condition characterized by absorption of too much dietary iron. This may lead to iron overload, which can cause damage to the joints and certain organs, such as the liver, skin, heart, and pancreas. This test includes the two most common variants linked to this condition.

Basic, you have **two copies** of a genetic variant we tested.

Women with this result have a slightly increased risk of developing iron overload related to hereditary hemochromatosis. Lifestyle and other factors can also affect your risk.

Variant detected
in the HFE gene

1 variant not determined

The test may not be able to determine a result for every variant tested. This can be caused by random test error or other factors that interfere with the test.

How To Use This Test

This test does not diagnose hereditary hemochromatosis 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 the **C282Y** and the **H63D** variants in the HFE gene linked to hereditary hemochromatosis.

Limitations

- Does **not** test for all possible variants linked to HFE-related hereditary hemochromatosis.
- Does **not** test for variants in other genes linked to hereditary hemochromatosis.

Important Ethnicities

- The variants included in this test are best studied in people of **European** descent.

You have a **slightly increased risk** of developing iron overload related to hereditary hemochromatosis based on your genetic result.

However, most women with this result do not develop iron overload. Because women lose iron through menstruation, iron overload is rare in women. When women do develop iron overload, it tends to be after menopause.

We detected two copies of the C282Y variant in the HFE gene.

Your result could not be determined for the H63D variant, which is most commonly found in people of **Northern European** descent.

See Scientific Details

Women with this result have a slightly increased risk of developing symptoms related to hereditary hemochromatosis.

Studies estimate that women of **European** descent with this result have a 4-14% chance of being diagnosed with signs or symptoms of hereditary hemochromatosis.

See Scientific Details

Since you share DNA with your family members, they may also be interested in this result.

Both of your parents and each of your children likely have this variant. Your siblings likely have at least a 75% chance of having one or more copies of this variant.

Lifestyle and other factors can also influence the chances of developing iron overload related to hereditary hemochromatosis.

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

Age

Although developing symptoms of iron overload related to hereditary hemochromatosis is rare in younger people, the risk increases as a person ages. Symptoms of iron overload are most often diagnosed in men between the ages of 40 and 60, and in women after the age of 60.

See Scientific Details for more information

Age | Sex | Alcohol consumption | Diet

About Hereditary Hemochromatosis (HFE-Related)

Also known as: HFE-HH, primary hemochromatosis, hemochromatosis type I

When it develops

Because it is a genetic condition, hereditary hemochromatosis is present at birth. Many people with this condition never develop iron overload. Of those who do develop iron overload, only a small number develop symptoms. If men develop symptoms, they typically appear between 40 and 60 years of age. Women rarely develop symptoms, and when they do it tends to be after menopause.

Typical signs and symptoms of iron overload

- Joint and abdominal pain
- Fatigue and weakness
- Darkening of the skin
- Liver disease
- Heart disease
- Diabetes

How common is the condition?

Hereditary hemochromatosis is most common in people of Northern European descent. Around 1 in 250 people of European descent has the genotype most commonly associated with hereditary hemochromatosis. However, only some of those people will go on to develop symptoms of iron overload related to hereditary hemochromatosis.

How it's treated

People with hereditary hemochromatosis are typically monitored for symptoms or complications. Iron overload related to hereditary hemochromatosis is a treatable condition. In some patients, having blood drawn on a regular basis can help lower iron levels. People with iron overload are encouraged to avoid drinking alcohol to minimize liver damage and to limit intake of iron-rich food.

Read more at: National Institute of Diabetes and Digestive and Kidney Diseases | GeneReviews | Genetics Home Reference

Consider sharing this result with a healthcare professional, especially if you have other risk factors.

If you have a family history of this condition or think you have symptoms, consult with a healthcare professional.

Print report

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

See our Frequently Asked Questions for more information.

FAQs

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Scientific Details

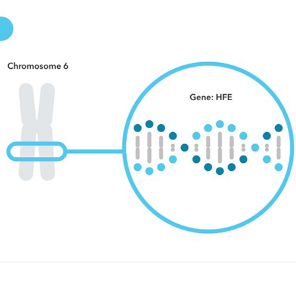
Hereditary hemochromatosis is a genetic condition characterized by absorption of too much dietary iron. This may lead to iron overload, which can cause damage to the joints and certain organs, such as the liver, skin, heart, and pancreas. This test includes the two most common variants linked to this condition.

HFE-related hereditary hemochromatosis is linked to variants in the HFE gene.

HFE

The HFE gene contains instructions for making a protein that helps control the amount of iron absorbed by the body. Certain variants in this gene disrupt the ability of the HFE protein to regulate iron absorption, causing too much iron to be absorbed.

Read more at Genetics Home Reference



You have two copies of a genetic variant we tested. Your result for the other tested variant could not be determined.

Variants Detected | View All Tested Markers

Marker Tested	Genotype*		Additional Information
C282Y Gene: HFE Marker: rs1800562	A Variant copy from one of your parents	A Variant copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [1, 2, 3, 5, 7, 9, 10, 11, 12, 13, 14, 18] ClinVar

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Interpretation

This report provides information about the risk of developing iron overload in people of Northern European descent who have the variants included in this test. Estimates for other ethnicities are not currently available.

Health Risk Estimates

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

For certain genotypes, quantitative risk estimates may not be available.

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

References [2, 9]

Risk estimates for developing signs or symptoms of hereditary hemochromatosis

The numbers in the table describe the percentage of people with the indicated genotype who are expected to be diagnosed with signs or symptoms of hereditary hemochromatosis, including iron overload.

Genotype	Men	Women
Two copies of C282Y variant	24-36%	4-14%
One C282Y variant and one H63D variant	3%	2%
Other genotypes	Not likely at risk	Not likely at risk

Other Factors

Hereditary hemochromatosis is a genetic condition. People with this condition have a higher risk of developing iron overload, which can lead to liver disease and other symptoms. In people with this condition, risk of developing iron overload can also be influenced by other factors.

This is not a complete list of other factors.

The factors described here include the most common and well-established risk factors associated with iron overload in people with hemochromatosis. Other factors not listed here may also influence risk for iron overload in people with the condition.

Typically, only people with certain combinations of variants in the HFE gene are at increased risk for iron overload. People with these genotypes in addition to other risk factors may have an even higher risk of developing iron overload.

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

Other Factors	References
Age In people with hemochromatosis, iron takes time to build up in the body. Thus, developing symptoms of iron overload related to hereditary hemochromatosis is rare in younger people. However, the risk increases as a person ages. Symptoms of iron overload are most often diagnosed in men between the ages of 40 and 60, and in women after the age of 60.	[17]
Sex Although most people with hereditary hemochromatosis do not develop noticeable symptoms, men with the condition are at higher risk than women for developing iron overload. Because iron takes many years to build up in the body, men usually don't experience symptoms of iron overload until their 40s or later. Women lose iron naturally through menstruation, reducing their risk. For women who do develop symptoms, they tend to develop them later than men, after menopause.	[2, 6]
Alcohol consumption In general, excessive alcohol consumption can lead to liver disease. In people with hemochromatosis, the liver is already at risk for damage from iron overload. For these individuals, the risk for liver damage is further increased with excess alcohol consumption. For example, studies have shown that people with two copies of the C282Y variant who have more than three to four drinks daily are significantly more likely to develop liver disease than those who drink less.	[6, 8, 16]
Diet Iron is an essential nutrient that the body needs to function properly. Consuming foods high in iron or taking certain supplements can increase the amount of iron stored in the body. For people with hemochromatosis, this may increase the chances of developing symptoms of iron overload. Consult with a healthcare professional before making any major dietary changes.	[6]

Test Details

Indications for Use

The 23andMe PGS Genetic Health Risk Report for Hereditary Hemochromatosis (HFE-Related) is indicated for reporting of the C282Y and H63D variants in the HFE gene. This report describes if a person has variants linked to hereditary hemochromatosis and a higher risk for iron overload, but it does not describe a person's overall risk of developing iron overload. This report is most relevant for people of Northern European descent.

Special Considerations

- Genetic testing for hereditary hemochromatosis is recommended under certain circumstances by several health professional organizations, including the American Association for the Study of Liver Diseases and the European Association for the Study of the Liver. [11]

Test Performance Summary

Clinical Performance

About 91% of all cases of HFE-related hereditary hemochromatosis are caused by the two variants included in this test.

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. All 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.*
- 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.

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Change Log

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

Date	Change
July 25, 2017	Hereditary Hemochromatosis (HFE-Related) report created.

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Frequently Asked Questions

Hereditary hemochromatosis is a genetic condition characterized by absorption of too much dietary iron. This may lead to iron overload, which can cause damage to the joints and certain organs, such as the liver, skin, heart, and pancreas. This test includes the two most common variants linked to this condition.

Hereditary Hemochromatosis (HFE-Related)

What is hereditary hemochromatosis and how is it related to iron overload?

Hemochromatosis is a condition in which the body absorbs too much dietary iron. As iron builds up in the body over time, it may lead to a condition called iron overload. This can cause damage to the joints and certain organs, including the liver, skin, heart, and pancreas. Hemochromatosis can be caused by genetics, also known as hereditary hemochromatosis, or can be acquired over a person's lifetime due to non-genetic factors, such as blood transfusions or overuse of iron supplements. Most cases of hereditary hemochromatosis are due to mutations in the HFE gene, although mutations in other genes have also been found. This test includes the two most common variants linked to hereditary hemochromatosis – the C282Y and H63D variants, both in the HFE gene. Learn more about HFE-related hereditary hemochromatosis.

Is this answer helpful? Yes No

What does this test do?

This test looks for two specific genetic variants in the HFE gene called C282Y and H63D. These variants are linked to hereditary hemochromatosis.

People with certain combinations of the genetic variants included in this test are more likely to develop iron overload, a sign of hereditary hemochromatosis. However, not everyone with these genetic variants will develop iron overload.

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

Is this answer helpful? Yes No

What does this test not do?

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

This test does not tell you if you have iron overload or if you will definitely develop the condition in the future.

This test does not include all possible genetic variants linked to hereditary hemochromatosis.

This test does not provide a complete assessment of risk for iron overload.

Is this answer helpful? Yes No

The report says the variants included in this test are best studied in people of European descent. What if I'm not of European descent?

The effect of these variants on a person's risk of developing iron overload is best studied in people of European descent. If a person who is not of European descent has one or more of these genetic variants, he or she is still expected to have a similar risk of developing the condition, but the exact risk numbers are not well understood. See Scientific Details for more information.

Is this answer helpful? Yes No

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

You can learn more about hereditary hemochromatosis and iron overload from the following resources:

- American Liver Foundation
- Iron Disorders Institute
- The Haemochromatosis Society
- Canadian Hemochromatosis Society
- 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 two copies of one variant called C282Y were detected. What does this mean?

This means you have two copies of the genetic variants we tested. Women with this result have a slightly increased risk of developing iron overload related to hereditary hemochromatosis.

Studies estimate that women of European descent with this variant have a 4-14% chance of being diagnosed with symptoms of hereditary hemochromatosis. See Scientific Details for more information.

However, this does not mean you have developed or definitely will develop symptoms. Other factors can also affect your risk. Learn more about other factors.

Is this answer helpful? Yes No

My report says two copies of one variant called C282Y were detected. What are some things I could do?

This genetic result is associated with a slightly increased risk of developing iron overload related to hereditary hemochromatosis.

Consider sharing this result with a healthcare professional, such as a physician or a genetic counselor. This is especially important if you have other family members who have had iron overload or if you have other factors that may increase your chances of developing the condition. Learn more about other factors.

Is this answer helpful? Yes No

What does it mean if the result for a variant could not be determined?

This means we could not tell if you have or do not have the tested genetic variant. This can be caused by random test error or other factors that interfere with the test.

Is this answer helpful? Yes No

What does slightly increased risk mean?

A "slightly increased risk" means that, based on your genetic result for the test, your chances of developing iron overload related to hereditary hemochromatosis are slightly higher than average. See Scientific Details for more information.

Consider sharing this result with a healthcare professional, such as a physician or a genetic counselor. This is especially important if you have other family members who have had iron overload or if you have other factors that may increase your chances of developing the condition. Learn more about other factors.

Is this answer helpful? Yes No

How could my result affect my family?

Since you share DNA with your family members, they may also be interested in your result. If you are thinking about talking to family members about your results, see this article for a discussion of things to consider before having the conversation.

Because you have two copies of one variant, it is expected that:

- Each of your children will inherit this variant from you.
- Each of your parents has at least one copy of this variant.
- Each of your siblings has at least a 75% chance of having one or more copies of this variant.

If your partner has a variant linked to iron overload related to hereditary hemochromatosis, your children could inherit a combination of variants that may put them at risk of developing the condition.

Is this answer helpful? Yes No

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