Familial Hypercholesterolemia

Your result

Austin, this test found a difference in your DNA linked to familial hypercholesterolemia (FH).

People with FH can have very high cholesterol and an increased chance of developing serious heart problems as early as 20.

Key takeaways

- For people with FH, diet and lifestyle changes are typically not enough to lower cholesterol levels. A healthcare provider usually prescribes medicine to help people with FH lower their cholesterol to clinically prescribed levels (medical report). This is one of the reasons which FH can be different from the more typical causes of high cholesterol.
- This does not mean you have high cholesterol or will develop heart problems—just that your chances are higher than typical.
- At least one of your parents likely has this same DNA difference linked to FH. Each of your children and siblings has a 1 in 2 chance of having it, too.
- Learn more about FH by reviewing this report and screening educational videos, webinars, and genetic counseling resources from PWNHealth, the independent clinician network that provided clinical oversight for your test.
- If you only do one thing, do this: Schedule an appointment as soon as possible with a healthcare provider to check your cholesterol levels. Download your physician report to take with you.

Download physician report

Scientific Results
Austin, this test found a difference in your LDLR or PCSK9 gene. Further details about your result that would be helpful to a healthcare provider to know are included in the physician report you can download and bring to them.

What this means for you

You have a much higher chance of having seriously high levels of cholesterol and heart problems at a younger age.

This higher chance is because you have a difference in your DNA that is linked to familial hypercholesterolemia (FH).[1] Most people with FH have very high levels of the bad form of cholesterol, known as LDL. If untreated, high levels of LDL cholesterol can increase the chances of having a heart attack and other serious heart problems.[2]

You can work with a healthcare provider to take steps to lower your cholesterol levels and your chances of FH-related heart disease, there are medicines that can help lower LDL cholesterol levels.[3] Maintaining healthy cholesterol levels will make you less likely to develop heart problems.[4]

Chance of developing heart problems with untreated FH

Medications can help people with FH lower their cholesterol to decrease their chance of developing serious heart problems.

What is FH?

Around 1 in 250 people in the United States has FH.

You're far from alone. FH is a genetic condition that can result in very high levels of bad form of cholesterol, known as LDL. It is thought that more than one million people in the United States have FH, but only about 10% of them are aware of it.[6] [7] Knowing that you have FH gives you information a healthcare provider can use to try to improve your cholesterol levels, which significantly lower the chances of developing heart problems.[8]

You need a certain amount of cholesterol to keep your body working properly.[9] Cholesterol can be found in the blood in two different forms, LDL and HDL. Ideally, you should have high levels of HDL and low levels of LDL.[10] When your LDL levels are high—as can happen in people with FH—cholesterol can build up in your bloodstream. This can lead to a sudden blood clot, heart attack, or stroke.[11]

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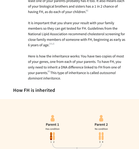
The heart and bad cholesterol



Our bodies are constantly working to clear extra LDL cholesterol out of our blood, and there are several genes that make this happen. Some people with FH have a difference in one or more of these genes that causes LDL cholesterol to not be cleared as well.[12] This can cause higher levels of LDL cholesterol in the blood.

Sometimes, people with FH can have yellowish patches under the skin or a light ring around the pupil of the eye.[13] But usually there aren't any visible signs of high cholesterol.[14] A healthcare provider can order a blood test for you to find out whether your cholesterol levels are within a normal range.

Most people with FH require medication to lower their cholesterol levels.[15] In addition, lifestyle changes like diet and exercise can help further improve their cholesterol levels.[16]

Ethnicity

People of certain ethnicities have a higher chance of having FH. For example, about 1 in 200 people of Ashkenazi Jewish or Ashkenazi Jewish ancestry have such difficulty,[17] [18] a higher for people of Old Order Amish descent, whose chances may be as high as 1 in 2.[19]

How inheritance works:

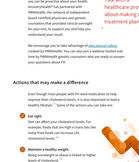
Each child of someone with FH has a 1 in 2 chance of having it as well.

In almost all cases of FH, people inherit the condition from one of their biological parents. This means that if you have FH, at least one of your parents probably has it too. It also means each of your biological brothers and sisters has a 1 in 2 chance of having FH, as do each of your children.

It is important that you share your result with your family members so they can get tested for FH. Guidelines from the National Lipid Association recommend cholesterol screening for close family members of someone with FH, beginning as early as 4 years of age.[20]

Here is how the inheritance works: You have two copies of most of your genes, one from each of your parents. To have FH, you only need to inherit a DNA difference linked to FH from one of your parents. This type of inheritance is called autosomal dominant inheritance.

How FH is inherited



What you can do

This information about your genes is only the beginning.

There are actions people with FH can take. The first thing to do is to see a healthcare provider about getting a blood test to check your cholesterol levels.

If your cholesterol is high, talk with a healthcare provider about appropriate next steps. A common recommendation for people with high cholesterol levels is to lower their cholesterol.[21]

It's important to learn more about FH so that you can be proactive about your health. Resources provided here are primarily from PWNHealth, an independent board-certified physicians and genetic counselors that provided clinical oversight for your test. To support you and help you understand your result.

Talk with a healthcare provider about making a treatment plan.

We encourage you to take advantage of educational videos created by PWNHealth. You can also join a webinar hosted twice by PWNHealth genetic counselors who are ready to answer your questions about FH.

Actions that may make a difference

Even though most people with FH need medication to help improve their cholesterol levels, it is also important to lead a healthy lifestyle.[22] Some of the actions you can take are:

- **Eat right.** Diet can affect your cholesterol levels. For example, foods that are high in trans fats like many fried foods can increase LDL cholesterol levels.[23]
- **Maintain a healthy weight.** Being overweight or obese is linked to higher levels of cholesterol.[24]
- **Stop smoking.** Physical activity can increase your levels of the good cholesterol, known as HDL.[25] This cholesterol helps drive down LDL levels. Diet and exercise also helps keep your heart healthy.
- **Keep diabetes under control.** People with diabetes have a higher chance of high cholesterol and the heart problems it can cause.

Limitations

This test cannot tell you whether you have high cholesterol, just whether you have a higher chance of developing it.

This test informs you about differences in your DNA and your chance of developing high levels of LDL cholesterol at some point in your life because of those DNA differences. There are technical limitations to this test:

Read more about limitations

Incorrect results can happen, although they are rare. They might be due to technical problems or like delivering, you or a family member accidentally provided a sample from someone else.

This test will not cover every gene.[26] This technology is capable to find, more than 2,400 DNA differences in the LDLR and APOB genes,[27] but a specific DNA difference in your genes may be missed. Because of this limitation, technology can miss about 1 in 5 possible cases (called false-differences, linked to FH but this test did not look for. In some cases, having additional DNA differences in these genes increases the chance that cholesterol levels are higher.

Occasionally, this test fails to detect some of the DNA differences that it is designed to look for.

There are other genes the test did not look at that may contain a DNA difference that is known to increase the chance of high LDL cholesterol levels.

The interpretation of your result may change as more is learned about which DNA differences are connected to high LDL cholesterol levels in the future.

This test is not a substitute for diagnostic genetic testing. For further information about the limitations of this test, download your physician report.

Genetic testing for FH is intended to be interpreted by board-certified physicians and independent board-certified genetic counselors, and with oversight from a board-certified physician. Genetic testing results should be interpreted by a board-certified genetic counselor or physician in your area. The results should not be interpreted, and do not determine your overall chance of developing a disease, or your ability to pass on a disease-causing change to your children. Only healthcare professionals who have been trained in the interpretation of genetic test results, and who are aware of the limitations of this test, should be used to interpret your results. Genetic testing results should be used in the context of other medical information, including before making any treatment, dietary, or lifestyle changes.

If you only do one thing, do this:

Schedule an appointment as soon as possible with a healthcare provider to check your cholesterol levels. Download your physician report to take with you.

You do not have to wait to do all of this on your own. A healthcare provider can help you with FH and can answer questions about getting a blood test that can check your cholesterol. A healthcare provider may recommend having your cholesterol levels checked regularly if it's especially important to monitor those levels, or you get sicker.

Download physician report

What else can you do?

Get support from a PWNHealth genetic counselor

Genetic counselors are board-certified physicians and independent board-certified physicians and genetic counselors that provided clinical oversight for your test. You can conditionally use a PWNHealth genetic counselor questions during a webinar hosted with the genetic test results.

You can also schedule a question to a PWNHealth genetic counselor below.

Sign up for an informational webinar

Ask a genetic counselor a question

powered by Lifetrack

Share your results with your family

Your DNA difference linked to FH may be inherited from a parent and higher chance of having FH. That's why it's important for you to share your result with them. That way they can decide whether they want to have genetic testing to learn more about their health. We have created a document to help you.

Just remember to be sensitive.

Download fact sheet

Resources

Glossary FAQ References Resources

Here are some places to start:

- The FH Foundation
- Foundation of the Nutrition Lipid Association
- American Heart Association
- National Organization of Rare Disorders
- Genetic and Rare Diseases Information Center

Download report