University of Iowa Health Care

Inheriting High Cholesterol

What you need to know about familial hypercholesterolemia

High cholesterol is one of many risk factors that puts you at higher risk for heart disease. About 7 out of 100 adults in the U.S. have high cholesterol, or hypercholesterolemia. It is defined as LDL, or "bad" cholesterol, of 190 mg/dL or greater. Most cases of high cholesterol are due to a combination of diet, exercise, smoking, or other factors.

What is familial hypercholesterolemia?

Familial hypercholesterolemia (hi-per-ko-les-ter-ol-e-me-a) is also called FH. It is a genetic disease that causes very high cholesterol in the blood, starting from birth. Since it is genetic, it can be passed down from parent to child.

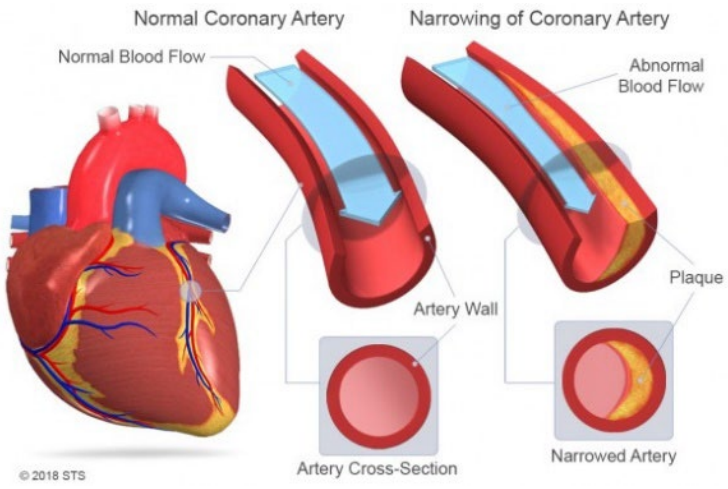
FH is one of the most common genetic diseases. It impacts 1 in 250 people worldwide. In the U.S., about 1.3 million people have FH. While it is common, not every person with high cholesterol will have FH.

FH is very underdiagnosed in the U.S., where 9 out of 10 people with FH do not know that they have it.

Why is it important to diagnose FH?

The risk of developing heart disease (also known as coronary artery disease) at an early age is very high for people with FH who are not treated. This risk is much higher for people with FH compared to people with high cholesterol due to other causes.

If it is not treated, 5 in 10 men with FH will have a heart attack by age 50, and 3 in 10 women will by age 60**.** There is no cure, but FH is very treatable.



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People with FH have high cholesterol levels at birth. This causes plaque to build up in arteries, leading to early heart disease, heart attacks, and strokes.

Could my high cholesterol be due to FH?

If you have high cholesterol, it could be due to FH if you have more than one of the below:

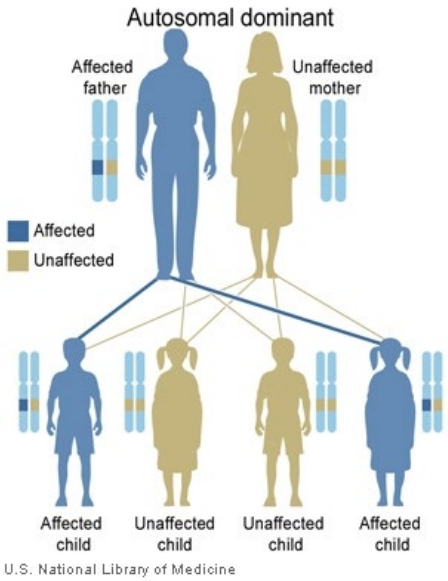
- Very high LDL, more than 190 mg/dL.
- Diagnosed with heart disease at a young age (before age 55 for men and age 60 for women).
- 1 or more close relatives (a parent, sibling, or child) with very high cholesterol or early heart disease, heart attacks, or strokes.
- Swollen or painful deposits under the skin on the elbows, joints, tendons, knees, hands, feet, or buttocks, or skin tags on the eyelids.
- A gray or bluish ring or arc around the colored part of the eye before age 45.

What causes FH?

FH is caused by changes in certain genes. The genes associated with FH make proteins that help control the levels of LDL in the blood. Some changes in these genes can make proteins that do not work the right way or do not work at all. This leads to the buildup of LDL in people with FH.

There are 2 main types of FH:

- Heterozygous FH (HeFH): This is the most common type. A child has a 50% chance of having HeFH if 1 of their parents has HeFH.
- Homozygous FH (HoFH): This is very rare. It causes very high LDL. Often over 500 mg/dL. This causes aggressive heart disease at a very early age, such as the teens or early 20s. HoFH can happen when **both** parents have HeFH.



HeFH is an inherited autosomal dominant trait.

- Autosomal = males and females equally likely to be affected
- Dominant = child has a 50% chance of being affected in one of their parents is affected.

How can I find out if I have FH?

Often, your doctor can diagnose you with FH in 2 ways:

1. They can analyze your cholesterol levels, ask about your family history of high cholesterol and heart disease, and do an exam to look for signs (such as skin changes) that some people with FH have. Your doctor may be able to diagnose you this way if enough signs are seen in both you and your family.
2. They can also diagnose you with genetic testing. They check the genes associated with FH: LDLR, APOB, PCSK9, and LDLRAP1, to see if there are harmful genetic changes. This testing can be done with blood or saliva.

How is FH treated?

If you are diagnosed with FH, your provider may suggest lifestyle changes, such as diet, exercise, and quitting smoking. But lifestyle changes alone are usually not enough to lower your LDL to safe levels. Doctors often prescribe medicine (like statins) to help lower LDL even more.

When should a person with FH start treatment?

Since FH is due to genetic causes, people with FH often have very high levels of LDL starting from birth. The longer a person is exposed to high LDL levels, the higher the lifetime risk of heart disease.

Experts suggest children with FH start cholesterol-lowering treatment (such as a low-dose statin) by 8 to 10 years of age. Studies show people with FH who are treated as a child have the same lifetime risk of heart disease as a person without FH.

How can genetic testing help?

Genetic testing for FH can help in many ways:

- It may help clarify if you have FH. It may be good to do if your cholesterol levels, family history, or other findings paint an unclear picture.
- It can make testing your family for FH easier, compared to relying on cholesterol levels alone.
- It may motivate you to take your medicine and make lifestyle changes.
- It may help you get insurance coverage for some cholesterol-lowering medicines that are often expensive, such as PCSK9 inhibitors (Repatha®, Praluent®).
- Confirming a genetic diagnosis leads to psychological relief for some people. They think, "my high cholesterol is due to a genetic condition, not because I have a poor diet or don't exercise enough."

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Genetic testing for FH has some limits and disadvantages:

- It is not able to find a genetic answer for each person or family with FH.
- Some results of genetic testing are uncertain.
- Insurance may not cover the cost of testing. However, most people whose insurance does not cover the cost pay no more than a few hundred dollars out of pocket. People can also get their estimated out of pocket costs before doing the test.
- Results of genetic testing may disqualify or increase the costs of certain types of insurance, such as life, long-term care, and/or disability.

Contact information

Talking with a genetic counselor may help you decide whether genetic testing for FH is right for you.

To schedule a visit at UI Health Care, call **319-356-7102** or visit **uihc.org/appointment**.

To learn more, go to:

- uihc.org/cardiovascular-genetics
- uihc.org/services/genetic-counseling
- thefhfoundation.org/
- nsgc.org/page/find-a-genetic-counselor