Familial Hypercholesterolemia

Familial hypercholesterolemia (FH) is a genetic disorder that causes high levels of cholesterol in the blood, which increases the risk of heart disease. FH is caused by a defect in the gene that codes for the LDL receptor, a protein that plays a crucial role in the removal of LDL (bad) cholesterol from the bloodstream.

In FH, the LDL receptor is either absent or does not function properly, allowing more LDL cholesterol to enter the cells, where it can form plaques on the inside of the arteries. This can lead to heart disease, including coronary artery disease, and other complications.

Testing for FH

To test for FH, doctors often use a combination of family history, physical examination, and laboratory tests. The most common laboratory test is the measurement of total cholesterol and LDL cholesterol levels.

How to Use This Test

This service is offered through laboratory testing. Please contact your healthcare provider for more information.

Interpreted by

[Name]

Limitations

These tests are not recommended for the general public. They are intended for healthcare providers and their patients with FH.

Important Risks

There are no known serious, long-term risks associated with this test.

In the U.S., about 2% of people who are 20 years old and older have high LDL cholesterol levels (levels ≥190 mg/dL). This is why it is important to talk to a healthcare provider about your FH risk and how to manage it.

If you have a personal or family history of FH, you may want to talk to a healthcare professional about your FH risk.

Lifestyle and other factors can also influence the chances of developing heart disease. It is important to make healthy lifestyle choices, such as eating a healthy diet, getting regular exercise, and maintaining a healthy weight.

Learn more about FH.

See a Family History Questionnaire for FH.

About Familial Hypercholesterolemia

Familial hypercholesterolemia is a genetic disorder that causes high levels of cholesterol in the blood, which increases the risk of heart disease. It is inherited in an autosomal dominant pattern, meaning that if you inherit one defective copy of the gene from a parent, you will have the disease.

People with FH have a 50% chance of passing the gene to their children. If both parents have FH, each child has a 75% chance of inheriting the gene.

A person with FH may have symptoms such as:

- High levels of cholesterol in the blood
- Painless, smooth, yellow nodules on the skin called xanthomas
- Thickening and narrowing of the arteries, which can lead to heart disease

If you or a family member has FH, it is important to talk to a healthcare professional about your FH risk and how to manage it.

Learn more about FH.