

# FAMILIAL HYPERCHOLESTEROLEMIA

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## What is Familial Hypercholesterolemia?

- Familial Hypercholesterolemia (FH) is an inherited disease, in which a genetic alteration causing high blood cholesterol is transmitted from generation to generation
- **Familial** means it runs in families; sometimes it is possible to trace the disease over several generations
- **Hypercholesterolemia** means high blood cholesterol
- The type of cholesterol that is specifically increased in Familial Hypercholesterolemia is Low Density Lipoprotein-Cholesterol (LDL-C)
- Individuals with FH may look perfectly healthy and are able to have a normal, active life

## Why is This Important?

FH is the most common genetic disorder. Approximately 1 in 500 people in the world has a genetic alteration that causes FH. If one parent has FH, there is a 50% chance that their son or daughter will also have it. FH is associated with an increased risk of heart disease.

**There is no cure for FH but it can be successfully treated.**

## What is Cholesterol?

Cholesterol is a fatty substance needed to build cells, make hormones and bile acids.

## What is LDL-Cholesterol?

- Often referred to as “bad cholesterol”
- LDL-cholesterol floats in the blood stream and transports cholesterol from one cell in your body to another cell
- Too much LDL-cholesterol in your blood stream is not good for you; excess cholesterol can be deposited in the walls of blood vessels making them narrower and causing the onset of heart disease

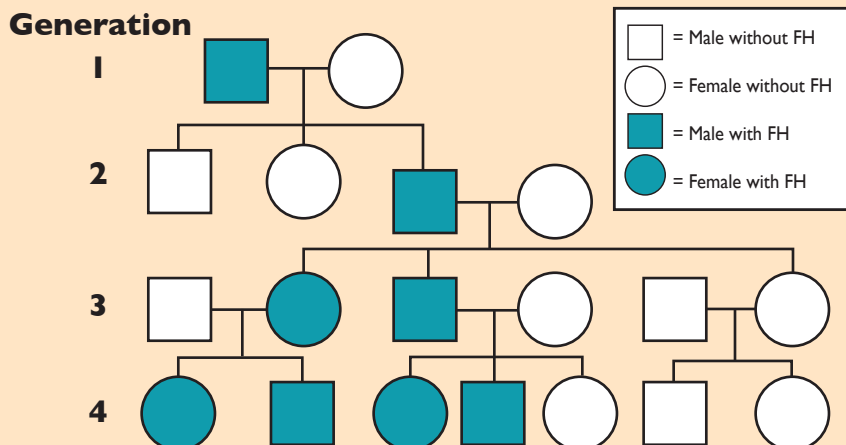
## Cholesterol Levels in FH

Blood tests may show:

- High levels of total cholesterol
  - Greater than 250 mg/dL in children
  - Greater than 300 mg/dL in adults
- High LDL levels
  - Greater than 170-200 mg/dL in children
  - Greater than 220 mg/dL in adults

## When to Suspect FH

**This family tree shows four generations affected by FH**



## Diagnosis/Symptoms

- A routine blood test shows high cholesterol
- A heart attack before the age of 50-60
- Family history of cardiovascular disease early in life
- Swollen tendons on the heels and hands
- Yellowish areas (cholesterol deposits) around the eyes

## What About My Children?

If you have FH your children should be screened and tested as early as possible. People with FH are born with it. An early diagnosis and early changes in diet and eating habits can help reduce the impact later in life.

# Treatment Options for Familial Hypercholesterolemia

The aim of treatment is to reduce your LDL-cholesterol to an acceptable level, thereby preventing or delaying heart disease. A lipidologist, a healthcare provider who specializes in treating diseases like FH, can develop the best treatment plan for you. To locate a lipidologist in your area, visit [www.learnyourlipids.com](http://www.learnyourlipids.com).

## Medication

The most important cholesterol-reducing drugs used to treat FH are **statins**. These medications work by reducing cholesterol production in cells. Other medications such as ezetimibe, niacin and bile acid sequestrants may also be taken to lower LDL levels.

It is important to continue maintaining a healthy diet and lifestyle even if taking a medication.

## Lifestyle Modification

You can reduce your cholesterol level and protect your heart health by:

- Stopping smoking
- Eating a healthy diet
- Not drinking excessive amounts of alcohol
- Regular physical activity
- Having healthy body weight and shape
- Controlling blood pressure if high

A change in diet is the first step in reducing cholesterol levels. Studies show that cholesterol levels can be reduced by 5-10% simply by changing what you eat.

Physical activity will help reduce your risk of heart disease. Adults should aim for at least 30 minutes of activity five days a week. If this is too difficult, break it down into three 10-minute periods. Any activity you choose should make you feel warm and slightly out of breath, but you should still be able to carry on a conversation. It is a good idea to vary the activity so you do not get bored.

## Apheresis

In extreme cases of FH where other treatments have failed it may be necessary to mechanically remove LDL-cholesterol from the blood. Apheresis is a treatment similar to kidney dialysis where the patient is connected to a machine where the blood is "cleaned" and then returned to the patient.

## Why is Lifelong Treatment Necessary?

Once LDL-cholesterol has decreased as a result of treatment it is important to prevent it from rising again. Your body makes cholesterol on a continuous basis and is exposed to fat and cholesterol in food every day. People with FH cannot regulate cholesterol properly and will need to maintain a healthy diet and lifestyle and continue to take lipid-lowering medication throughout life to keep levels under control.

### Guidelines for a Heart-Friendly Diet

- Eat less fat, particularly less saturated fat
- Replace saturated fat with unsaturated fat
- Eat more foods containing fiber, vegetables and fruit everyday
- Eat less cholesterol-rich food
- Limit food and drinks high in sugar or alcohol

**Next Steps—to learn more visit [www.learnyourlipids.com](http://www.learnyourlipids.com)**

Name: \_\_\_\_\_ Date: \_\_\_\_\_ Health Care Provider: \_\_\_\_\_

LDL Goals: \_\_\_\_\_ Weight Loss Goals: \_\_\_\_\_

Activity/Exercise Goals: \_\_\_\_\_

Medications Recommended: \_\_\_\_\_

Provided by your healthcare provider and the Foundation of the National Lipid Association  
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