

# Understanding familial hypercholesterolemia



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# What you need to know about familial hypercholesterolemia

Learning how to manage familial hypercholesterolemia (FH) might feel hard, especially at first. This inherited type of high cholesterol might leave you wondering what you can do to improve your health. Understanding your diagnosis can help you take control. Staying on track with lifestyle changes and medication therapy can help you manage symptoms and live a full and active life.

## Cholesterol and the body

Cholesterol is a fat-like substance in the body. It is also found in some foods. The body needs some cholesterol to work properly. But too much of it can build up in the walls of the arteries.<sup>1</sup>

It is hard for blood to flow through hardened or blocked arteries. This can raise the risk for heart disease. It can also raise the risk for heart attack.<sup>1</sup>

There are two “types” of cholesterol in the body<sup>1</sup>:

- High-density lipoprotein (HDL), or “good” cholesterol
- Low-density lipoprotein (LDL), or “bad” cholesterol

Having more HDL can lower the risk of heart disease. But high levels of LDL can raise the risk for heart attacks. It also raises the risk for heart disease.<sup>1</sup>

The liver removes cholesterol from the body. In those with FH, the liver cannot remove LDL from the body.<sup>1</sup>

## Causes and risk factors

Unlike other types of high cholesterol, FH is not caused by diet. It is not caused by lifestyle. It is present at birth.<sup>2-3,5-6</sup> FH is caused by a defect in chromosome 19.<sup>1,5</sup> This affects how the body clears LDL from the blood.<sup>1</sup>

## Who develops FH?

FH is one of the world’s most common genetic diseases.<sup>2</sup>

It affects about **1 in every 250 people** across the globe.<sup>3-4</sup>

In the United States, about **1.3 million people** live with FH.<sup>3</sup> But only about 10 percent of them are diagnosed.<sup>3</sup>

Most people with FH are undiagnosed and undertreated.<sup>2</sup>

FH affects **men and women** equally.<sup>4</sup>

A child typically inherits FH from one parent.<sup>5</sup> This is heterozygous FH.<sup>1</sup> More rarely, a child might inherit FH from both parents. This is called homozygous FH.<sup>1</sup> This form of FH is more severe. It can raise the risk for heart attack and death before age 30.<sup>1</sup>

## Diagnosis

There are many ways to diagnose FH. (See Table 1.) A doctor might use a few methods and results to confirm a diagnosis.<sup>1-2,5-6</sup>

## What you need to know about familial hypercholesterolemia (cont.)

**Table 1. Diagnosing FH**

Methods	Results
Blood tests	May show: <ul style="list-style-type: none"><li>• Total cholesterol above 300 milligrams per deciliter (mg/dL) in adults or above 250 mg/dL in children<sup>1</sup></li><li>• LDL above 190 mg/dL in adults or above 160 mg/dL in children that does not go down with changes in diet or other cholesterol-lowering treatment<sup>6</sup></li></ul>
DNA tests	Genetic testing showing the chromosomal defect for FH <sup>2,5</sup>
Family history	<ul style="list-style-type: none"><li>• Another family member already diagnosed with FH<sup>2</sup></li><li>• Early heart disease or heart attacks in the family<sup>3,6</sup></li></ul>
Heart function tests	Abnormal results on heart tests, like a cardiac stress test <sup>1</sup>
Physical exams	Cholesterol deposits in or around the skin, joints and eyes <sup>1,6</sup> : <ul style="list-style-type: none"><li>• Corneal arcus, or a white arc in the outer layer of the eye<sup>1,6</sup></li><li>• Xanthelasmas, or yellow areas around the eyes<sup>1,6</sup></li><li>• Xanthomas, or bumps on the skin or tendons<sup>1,6</sup></li></ul>

### Symptoms

Some people with FH have many symptoms. Some have just a few. Others have none at all.<sup>4,5</sup>

Symptoms might include<sup>1,5</sup>:

- Chest pain that suggests heart disease<sup>1,5</sup>
- Cholesterol deposits in or around the skin, joints and eyes<sup>1,5</sup>
- Cramps in the calves when walking or sores on toes that don't heal because of poor blood flow to the limbs<sup>2,5</sup>
- High total cholesterol and LDL<sup>1</sup>
- Sudden stroke-like symptoms, such as trouble speaking or one-sided drooping of face muscles<sup>5</sup>

### Related conditions and complications

Diagnosing and treating FH early is important. This can lower cholesterol and prevent complications.<sup>2,4</sup> But untreated FH or more severe forms of FH can have serious health effects. These might include<sup>2</sup>:

- A 10- to 20-times higher risk of coronary artery disease<sup>2</sup>
- A 50 percent increased risk of heart attack by age 50 in men and a 30 percent increased risk of heart attack by age 60 in women<sup>2,5</sup>
- Heart disease<sup>5</sup>
- Peripheral vascular disease<sup>2,5</sup>
- Stroke<sup>2,5</sup>

# Living with FH

There is no cure for FH. But there are many ways to treat it. Early diagnosis and the right treatment plan are important.

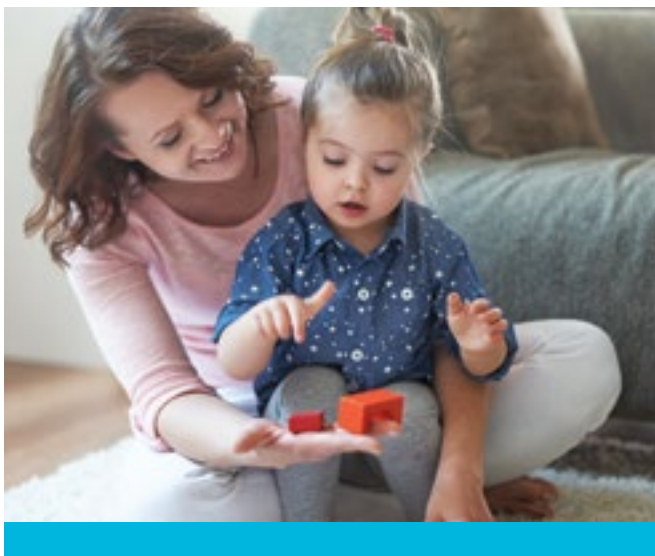
FH treatment is often more aggressive than treatment for noninherited high cholesterol.<sup>2</sup> This is especially true for those who inherit FH from both parents.<sup>1</sup> It also applies to those with other health issues, such as diabetes.<sup>2</sup>

FH treatment includes drug therapy and lifestyle changes. Lowering the risk for heart disease is the main goal. This means lowering LDL.<sup>1</sup>

Sometimes this means cutting cholesterol by half. Other times it means bringing it below 100 mg/dL.<sup>2</sup> The goal level can be different for each person.<sup>2</sup> Work with your doctor to build a treatment plan that is right for you.

## Medications

There are a few types of drugs for FH. These help lower cholesterol in different ways.<sup>1-2,7-8</sup> (See Table 2.) Some people take one drug. Others might take a combination of drugs.



**Table 2. Cholesterol-lowering drugs**

Type	Action
Bile acid sequestrant	Converts cholesterol into bile acid to lower cholesterol levels <sup>8</sup>
Cholesterol absorption inhibitor	<ul style="list-style-type: none"><li>• Lessens the amount of cholesterol absorbed from food<sup>7-8</sup></li><li>• Lowers triglycerides, another type of blood fat<sup>7</sup></li></ul>
Fibrate	<ul style="list-style-type: none"><li>• Lowers triglycerides<sup>7</sup></li><li>• Raises HDL<sup>7</sup></li></ul>
MPT inhibitor	Reduces cholesterol created by the body <sup>8</sup>
Nicotinic acid	<ul style="list-style-type: none"><li>• Lowers LDL<sup>7-8</sup></li><li>• Raises HDL<sup>7-8</sup></li></ul>
PCSK9 inhibitor	Helps the liver filter LDL from the body <sup>7</sup>
Statin	<ul style="list-style-type: none"><li>• Blocks the liver from making cholesterol<sup>7</sup></li><li>• Lowers LDL<sup>8</sup></li></ul>

Most people with FH might begin by taking statins.<sup>1-2</sup> But for many, this is not enough.<sup>2</sup> People often need more than one medication.<sup>2,7</sup>

Some people might have side effects, like muscle pain.<sup>2,7</sup> Others might not see better cholesterol levels with certain drugs.<sup>2</sup> These things might prompt a change to different medication. You can work with your doctor to adjust your treatment as needed.

Some people with very high LDL might also need LDL apheresis. This treatment filters LDL from the blood. It can decrease LDL by about 50 percent. But it must be done regularly to keep LDL levels under control.<sup>1-2,4-5</sup>

### **Staying on track with treatment**

It is important to stay on track with treatment. This can help you manage FH, control your symptoms and improve your health. No matter which medications are part of your treatment, take them exactly as prescribed by your doctor. This means taking them at the right times and the correct doses.

If you are having a hard time staying on track with treatment, ask your doctor or pharmacist for help. Do not stop treatment on your own. This can affect how well your cholesterol is controlled. Discuss any concerns with your doctor first. You can work with your doctor to adjust your treatment if needed.

## **Lifestyle changes**

Along with medication therapy, certain lifestyle changes can be part of FH treatment. Eating well, staying active and avoiding smoking can lower the risk for heart disease.<sup>1,2,5,9</sup>

### **Eating well**

Changing the way you eat might help control the effects of FH.<sup>1,5</sup> Your doctor might suggest a diet low in saturated and trans fats.<sup>3,10</sup> These types of fat can raise LDL. They are usually found in meat, dairy and convenience foods.<sup>9</sup> Most people with FH are instructed to aim for a diet with no more than 30 percent of total daily calories coming from fat.<sup>1,5</sup>

Certain changes might help<sup>1,5,9-10</sup>:

- Avoid packaged and fast foods as well as fried foods and baked goods.<sup>9-10</sup>
- Limit or avoid beef, pork and lamb.<sup>1,10</sup>
- Skip egg yolks, organ meats and full-fat dairy products.<sup>1,5,10</sup>

Healthful food choices can lower the cholesterol in your diet. Some healthful choices include<sup>2,9-10</sup>:

- Beans, nuts and seeds
- Fish
- Fruits
- Lean poultry
- Low-fat dairy
- Vegetables
- Whole grains

Eating this way can also add fiber to your diet. Soluble fiber can help lower LDL.<sup>9-10</sup> Work with your doctor or a dietitian to create a healthful eating plan that is right for you. You can monitor your progress together and adjust your plan as needed.

## Staying active

Staying active is also a good idea.<sup>1-2,10</sup> Being inactive can lower HDL, which is needed to remove LDL from your body.<sup>10</sup> But finding ways to move each day can help you<sup>9-10</sup>:

- Lower cholesterol
- Reach and keep a healthy weight
- Strengthen your heart

Most adults need about 150 minutes of moderate aerobic activity or 75 minutes of vigorous aerobic activity each week.<sup>9,11</sup> This can be broken up into shorter sessions of time each day of the week. But each session should last at least 10 minutes.<sup>11</sup>

Find an activity you enjoy. This can help make exercise a regular part of each day. Some examples of moderate aerobic activities include<sup>10-11</sup>:

- Cycling on level ground
- Brisk walking
- Doubles tennis
- Mowing the lawn
- Water aerobics

Some examples of vigorous aerobic activities include<sup>10-11</sup>:

- Basketball
- Cycling fast or on hills
- Dancing
- Jogging or running
- Singles tennis
- Swimming

Ask your doctor how much exercise is right for you. You might also ask for a referral to an exercise specialist. Together, you can build a plan to stay active and boost your heart health.

## Avoiding smoking

Smoking raises the risk for heart disease. When paired with other factors, like family history for heart disease, its effects are even worse.<sup>10,12</sup> Smoking also lowers HDL and makes it hard to exercise—two things that can help manage the effects of FH.<sup>10,12</sup>

If you don't smoke, don't start. Avoid secondhand smoke.<sup>12</sup> If you do smoke, take steps to quit. You can find help for quitting:

- Ask your doctor about any behavioral therapies, drugs or programs that might help you quit.
- Find out if your health plan offers a program to quit.
- Get free resources for quitting from the National Institutes of Health at [BeTobaccoFree.gov](http://BeTobaccoFree.gov) or 800-QUIT-NOW (784-8669).

### **Pregnancy and breastfeeding with FH**

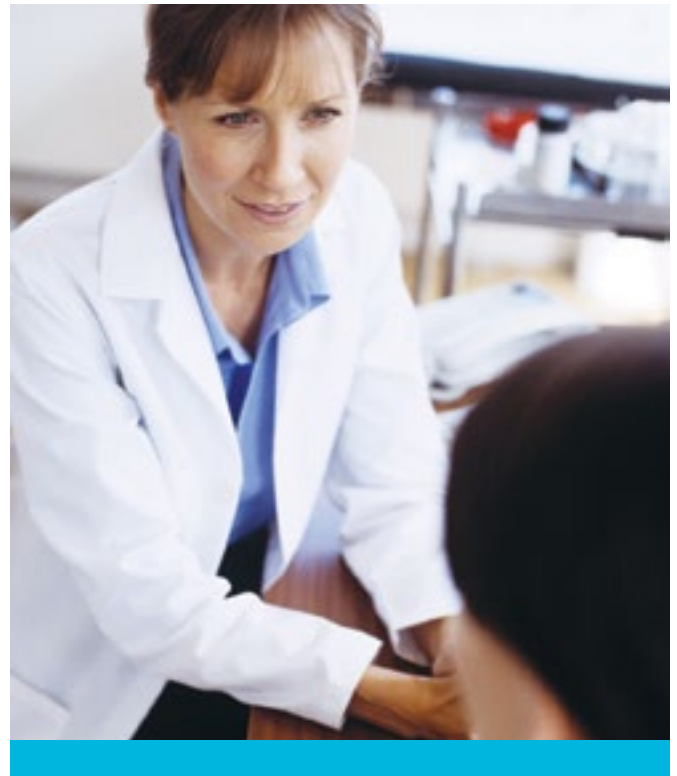
Cholesterol levels can rise during pregnancy for almost all women. This happens, in part, because of rising hormone levels.<sup>13</sup> Total cholesterol can rise by 25 to 50 percent. LDL can rise as much as 66 percent.<sup>14-15</sup> Because women with FH already have high cholesterol, managing rising levels during pregnancy is vital.

However, most cholesterol-lowering drugs should not be used when trying to get pregnant, during pregnancy or while breastfeeding.<sup>14-15</sup> It might be necessary to pause drug therapy during these times.<sup>15</sup> Ask your doctor what is right for you. Certain other medications and treatments might be possible. For example, LDL apheresis might be an option for women with more severe forms of FH or existing heart disease.<sup>15</sup>

Otherwise, healthy habits can support heart health during pregnancy<sup>14</sup>:

- Avoid tobacco smoke.
- Follow a low-fat, low-cholesterol diet.
- Get 30 to 60 minutes of moderate activity most days of the week

If you are trying to get pregnant or if you become pregnant during FH treatment, ask your doctor what treatments are safe. Talk about whether pausing medication therapy is right for you. Ask about how it might affect your overall health.<sup>15</sup> Together, you can choose the path to good health for you and your baby.



### **Ongoing care**

Work with your doctor to keep track of your progress. You can discuss how well your medications and lifestyle changes are working. Your doctor can also monitor side effects and adjust your treatment as needed. Each visit can help you stay on track with treatment and better manage FH and lower your risk for heart disease.



We've provided this information because we believe the more you know about FH, the better prepared you will be to monitor your health and manage your treatment. In addition, AllianceRx Walgreens Prime specialty pharmacy Care Team is here to provide you with dependable, personalized support to help you manage medication side effects and stay on track with your prescribed therapy. We look forward to being a member of your healthcare team and helping you get the best results from your treatment.

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