CONDITION

Familial hypercholesterolemia

Familial hypercholesterolemia is an inherited condition that causes very high LDL cholesterol from birth.

WHAT IT IS
Inherited condition causing very high LDL cholesterol

BODY SYSTEM
Heart and circulation

MAIN SYMPTOMS
Usually none, chest pain, tendon lumps, corneal ring

WHAT CAUSES IT
Inherited changes affecting LDL removal

HOW IT IS DIAGNOSED
Cholesterol testing, clinical criteria, sometimes genetic testing

MAIN TREATMENT
Cholesterol-lowering medicines and heart-healthy habits

CAN IT BE CURED?
No - inherited tendency remains lifelong

IS IT AN EMERGENCY?
Sometimes - possible heart attack or stroke signs

LONG-TERM OUTLOOK
Often good with early, sustained cholesterol lowering

Overview

Familial hypercholesterolemia, often shortened to FH, is an inherited condition that causes a high level of low-density lipoprotein cholesterol from birth. This is often called LDL or "bad" cholesterol.

In FH, the body cannot remove LDL efficiently, so it builds up in the blood. Over many years, LDL can collect in artery walls and contribute to atherosclerosis, meaning narrowing and stiffening of the arteries.

Lifelong exposure to high LDL can cause heart and blood vessel problems at a younger age than usual, including angina and heart attacks. Early diagnosis and sustained treatment can substantially lower this risk.

Most people with FH inherit one altered gene copy, known as heterozygous FH. A much rarer and more severe form occurs when altered copies are inherited from both parents.

FH is not caused by eating the wrong foods. Diet and activity affect heart health, but the main cause of high LDL is inherited.

Symptoms

FH usually causes no day-to-day symptoms. Many people feel well for years even when their LDL is very high.

Possible visible signs of cholesterol buildup include:

These signs do not prove someone has FH. Xanthelasma and corneal arcus can occur for other reasons, particularly later in life. Tendon swellings linked to cholesterol are more suggestive.

If narrowed arteries limit blood flow to the heart, symptoms may include chest pressure during activity, shortness of breath, or unusual fatigue. Sometimes the first symptom is a heart attack.

Children with heterozygous FH usually have no symptoms. Those with a severe form can develop cholesterol deposits or heart and artery problems at a young age.

- Firm swellings over tendons, especially the Achilles or hand tendons
- Yellowish patches around the eyelids, called xanthelasma
- A pale or white ring around the coloured part of the eye, called corneal arcus

Causes and risk factors

FH is caused by an inherited genetic change affecting how the body handles LDL. Commonly affected genes include LDLR , APOB , and PCSK9 .

The liver normally removes LDL from the blood using LDL receptors. Many FH-related changes reduce the number of working receptors or prevent them from handling LDL normally.

Most FH follows a dominant inheritance pattern. If one parent has this form, each biological child has a 1 in 2 chance of inheriting it. Less common inheritance patterns have different risks.

A family history suggesting FH may include:

Smoking, high blood pressure, diabetes, kidney problems, low physical activity, and other cholesterol abnormalities do not cause FH, but they can increase cardiovascular risk.

- Very high LDL in several relatives

- Heart attacks, angina, or coronary procedures at unusually young ages
- A parent, sibling, or child with diagnosed FH
- Tendon cholesterol deposits in close relatives

Diagnosis

FH is diagnosed using cholesterol results, personal and family history, physical findings, and sometimes genetic testing.

A lipid profile measures LDL, high-density lipoprotein cholesterol, total cholesterol, and triglycerides. FH usually causes markedly raised LDL, but a single number is not enough to diagnose everyone.

A healthcare professional may ask about cholesterol levels and early heart problems in close relatives. They may also check for tendon swellings or deposits around the eyes.

Other causes of high cholesterol must be considered, including an underactive thyroid, kidney or liver conditions, some medicines, and certain dietary patterns. Another cause does not always exclude FH but may affect how results are interpreted.

Clinical scoring systems combine LDL levels, family history, early heart problems, and physical signs. The system used varies between healthcare settings.

A blood or saliva genetic test may identify an FH-related change. However, a negative result does not always rule out FH because current tests cannot identify every cause.

When one person is diagnosed, close biological relatives may be offered cascade testing. This can identify relatives with high LDL before complications develop.

Treatment and management

Treatment aims to lower LDL and reduce future heart and blood vessel problems. Because LDL has been raised since birth, treatment is usually lifelong.

Statins like atorvastatin or simvastatin are commonly used first. They reduce cholesterol production in the liver and help it remove more LDL from the blood. Blood tests and follow-up appointments assess the response and check for unwanted effects.

Many people need more than one medicine. Options may include cholesterol-absorption inhibitors, PCSK9-targeting medicines, bempedoic acid, or bile acid sequestrants. The choice depends on age, LDL level, other conditions, side effects, pregnancy plans, and local

availability.

Severe FH may require specialist treatment, including medicines for particular genetic forms or LDL apheresis. During apheresis, blood passes through a machine that removes LDL before being returned to the body. Liver transplantation is rarely considered for exceptionally severe cases.

Children with FH should be managed by a clinician experienced in childhood cholesterol conditions. Heart-healthy habits begin early, and medicine may be recommended based on age, LDL level, family history, and overall risk.

Lifestyle measures support treatment but usually do not lower LDL enough by themselves. Helpful measures include:

Pregnancy requires planning because some cholesterol-lowering medicines are usually paused. People at exceptionally high cardiovascular risk may need a different approach. Do not stop prescribed medicine without speaking to the clinician managing your FH. If pregnancy is possible, ask for advice before trying to conceive.

- Avoiding smoking and second-hand smoke
- Eating more vegetables, fruit, whole grains, beans, nuts, and fish
- Replacing saturated fats with unsaturated fats where practical
- Staying physically active in ways suited to your health
- Managing blood pressure, diabetes, and body weight where relevant

Complications

The main complication is atherosclerosis in the arteries supplying the heart. This can cause angina, a heart attack, or the need for a procedure to improve blood flow.

FH can also narrow arteries supplying the brain, legs, and other organs, increasing the risk of stroke or peripheral artery problems. Severe forms can affect the aorta and aortic valve, sometimes at a young age.

Risk varies with the genetic form, lifetime LDL exposure, smoking, blood pressure, diabetes, and how early effective treatment begins.

Prevention

FH itself cannot usually be prevented because it is inherited. Healthy eating and exercise

cannot remove the underlying genetic tendency.

Complications can often be delayed or prevented through early cholesterol testing, consistent treatment, avoiding smoking, and controlling other cardiovascular risk factors.

Cascade testing can identify affected relatives before artery damage causes symptoms. Genetic counselling can explain inheritance and testing options for people planning a family.

Living with it and what to expect

FH is lifelong. Without treatment, LDL generally remains high even if you feel well and follow a healthy diet. Some people need several medicines or specialist care to reach an appropriate level.

Regular follow-up usually includes cholesterol tests, a medicine review, and checks for other heart risk factors. The frequency depends on age, treatment changes, LDL response, and whether heart disease is already present.

Do not use symptoms to judge whether treatment is working. High LDL is usually silent, so blood tests are needed.

Many people with heterozygous FH lead active lives without an early heart complication, especially when diagnosed and treated promptly. Treatment lowers risk but cannot guarantee that a heart or circulation problem will never occur.

Severe inherited forms require closer lifelong care, sometimes including frequent appointments, several medicines, and apheresis. Treatment plans may change as health needs and available therapies change.

When to get medical help

Arrange a routine appointment if you have repeatedly high LDL, a close relative with FH, or a family pattern of heart attacks or coronary procedures at young ages. Swellings over the Achilles or hand tendons also need assessment.

If you have FH, contact your healthcare team if you develop possible medicine side effects, have difficulty taking treatment consistently, become pregnant, or plan a pregnancy.

Seek emergency help for possible signs of a heart attack, including:

Call emergency services for possible stroke signs, such as sudden facial drooping, weakness or numbness on one side, trouble speaking, loss of vision, severe imbalance, or an unusual sudden headache.

Do not drive yourself if you may be having a heart attack or stroke.

- New chest pressure, tightness, squeezing, or pain that does not settle
- Discomfort spreading to an arm, shoulder, jaw, back, or upper abdomen
- Chest discomfort with sweating, nausea, breathlessness, or faintness

Common questions

Is familial hypercholesterolemia the same as ordinary high cholesterol?

No. Many factors can raise cholesterol, including age, diet, medicines, and other conditions. FH is inherited, usually causes high LDL from birth, and often affects several family members.

Can I lower FH cholesterol with diet alone?

Usually not enough. A heart-healthy diet can improve cholesterol levels and lower risk, but it cannot correct the inherited problem with LDL removal. Most people also need medicine.

Why do I need treatment if I feel fine?

High LDL rarely causes symptoms before an artery becomes significantly narrowed or blocked. Treatment reduces LDL exposure and its buildup in artery walls.

Should my family be tested?

Close biological relatives are commonly offered cholesterol testing and, when a familial genetic change is known, targeted genetic testing. Parents, siblings, and children are usually considered first.

Does a negative genetic test mean I do not have FH?

Not always. Genetic testing does not find every cause. FH may still be diagnosed from LDL levels, family history, physical findings, and early heart problems.

Can I have children if I have FH?

Yes. FH does not usually prevent pregnancy or parenthood, but it can be passed to biological children. Medicines may need review before and during pregnancy, so pre-pregnancy planning is important.

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