

What is Familial Hypercholesterolemia?

Familial Hypercholesterolemia (FH) is an inherited disorder that affects your body's ability to clear a specific type of cholesterol, known as LDL-C. Increased levels of LDL-C can cause atherosclerosis, which is deposits of plaque in your arteries. It can also affect how blood flows through your body leading to circulation abnormalities. This can lead to early heart attacks, strokes, and peripheral artery disease.



When is FH suspected?

- LDL-C cholesterol is 190 mg/dL or higher in adults
- LDL-C cholesterol is 160 mg/dL or higher in children
- If you or other family members have had a heart attack or stroke at an early age (under age 55 in men and under age 65 in women)
- Genetic testing **may** confirm a FH diagnosis, but is not required for diagnosis

There are two forms of FH: Heterozygous FH (HeFH) and Homozygous FH (HoFH). The table below shows the differences.

Heterozygous FH (HeFH)	Homozygous FH (HoFH)
Abnormal gene inherited from 1 parent	Abnormal genes inherited from both parents
Occurs in 1 in 250 persons	Occurs in 1 in 300,000 persons
LDL-C cholesterol >160 mg/dL in children LDL-C cholesterol >190 mg/dL in adults	LDL-C >400 mg/dL
May have physical symptoms, such as cholesterol deposits in the eye, tendons, knees, elbows and/or between fingers and toes	Likely physical symptoms, such as cholesterol deposits in the eye, tendons, knees, elbows and/or between fingers and toes. These deposits are not always present
Treatment as early as age 10	Treatment at time of diagnosis, no matter the patient's age, with a specialist

How is FH treated?

Several medicines are used to treat FH. The first is usually a statin. Other medicines can be added to reduce the LDL-C to recommended levels. Examples of these medicines include oral drugs (ezetimibe, bempedoic acid, and bile acid sequestrants) and injectable medications (alirocumab and evolocumab). For some people, a treatment called Lipoprotein Apheresis is used to remove excess LDL-C cholesterol from the body. This procedure is done every one to two weeks. For HoFH, there are additional treatment options available: evinacumab (an infusion once a month) and lomitapide (a daily oral medicine).



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If you have FH, what does this mean for your family?

- Screening all family members for FH is important.
 - Children, parents, and siblings have a 1 in 2 chance of having HeFH.
 - If both parents have FH, each child will have a 1 in 4 chance of having HoFH.
- Children with a family history of FH should have their cholesterol checked at age 2.
- Children need a heart-healthy diet and regular physical activity.
- Some children with FH will require medication at a young age.

Final Highlights

- While not curable, treating FH can help you lead a full life and lower your risk of:
 - Heart attack
 - Stroke
 - Circulation problems such as peripheral arterial disease (PAD)
- Follow up and taking prescribed cholesterol medicines through your lifetime is very important.
- A heart-healthy diet and regular physical activity will help to lower your heart disease risk.

FH Resources

- Family Heart Foundation: www.thefhfoundation.org
- The Foundation of the National Lipid Association: www.learnyourlipids.com
- Preventive Cardiovascular Nurses Association: <https://pcna.net/clinical-resources/patient-resources/familial-hypercholesterolemia/>
- American Society for Preventive Cardiology: www.aspconline.org



Ask your health care provider to help you:

- Make a plan for exercise.
- Quit smoking cigarettes or e-cigarettes, or stop using smokeless tobacco.
- Schedule a visit with a dietitian to help you make healthy diet changes.
- Schedule an appointment with a cholesterol or lipid specialist, if needed.