

Familial Hypercholesterolemia



What is FH?

- ◆ FH stands for Familial Hypercholesterolemia, an inherited disorder that causes severely elevated cholesterol levels. A normal total cholesterol level is approximately 200 mg/dL, but people with FH may have a cholesterol level in the 350 to 500 mg/dL range.
- ◆ Untreated individuals with FH have approximately 20 times higher risk of early onset heart disease compared with people with normal cholesterol levels.
- ◆ The type of cholesterol that is elevated is called LDL (low density lipoprotein) cholesterol. Excess LDL cholesterol collects in the blood vessels, eventually decreasing blood flow through these vessels. The blocked blood vessels may lead to chest pain and heart attacks.
- ◆ FH is common; about 1 in 200 people have FH.



How is FH diagnosed?

FH is usually diagnosed by a blood cholesterol test, called a "lipid panel," and consideration of personal and family history. Genetic testing may be done, but is usually not necessary. Signs of FH may also be visible during a physical exam. These can include yellow deposits on the skin around the eyes, a white ring on the colored part of the eye, and cholesterol deposits in the tendons of the hands, elbows and ankles.



Should I be concerned about my family?

Yes. Children, siblings and parents of individuals with FH should be screened for FH by having their cholesterol tested by a health care provider. Ideally, children should be tested after age 2 if someone in their family has FH.

Each time a person with FH has a child, there is a 50% chance that the gene for FH will be passed on to the child.

Rarely, a child inherits the FH gene from both parents. This results in a more severe form of FH, called homozygous FH (this occurs in 1 in about 250,000 to 1,000,000 people). If left untreated, a person with homozygous FH may die of heart disease in childhood or adolescence.

What can I do for my FH?

The goal of treating FH is to prevent or delay the onset of heart disease. This is done by lowering the LDL cholesterol to an acceptable level.

Medicines	A category of medicines, called statins, are the primary drugs to lower LDL. In addition, ezetimibe and bile acid sequestrants are other medicines used to lower LDL cholesterol. A new class of medications called PCSK9 Inhibitors are given as injections under the skin every 2 to 4 weeks. For homozygous FH, the medications lomitapide and mipomersen can be used.
Diet & Lifestyle	People with FH should follow a heart healthy diet that limits saturated fat, aim to maintain a healthy weight, get 2.5 hours of moderate physical activity or 75 minutes of vigorous physical activity weekly, limit alcohol intake and not smoke or use tobacco products.
LDL Apheresis	LDL apheresis is a medical therapy for patients who are not able to meet their LDL cholesterol goal with medicines and lifestyle changes. This procedure physically removes LDL cholesterol from the blood. Blood is removed from one arm and goes through a special filter that removes LDL cholesterol. The newly filtered blood, with a much lower LDL level, is then returned to the other arm. Medical professionals monitor patients during the procedure, which takes 2 to 3 hours and is generally performed once every other week.
Lipid Specialist	If you have FH and are not able to take a statin medication or to lower your LDL cholesterol adequately with medicines and lifestyle changes, it is important to see a lipid (cholesterol) specialist. In addition, children with FH should also see a lipid specialist.