Familial hypercholesterolemia
Making sense of family heart disease

CardioSmart
AMERICAN COLLEGE of CARDIOLOGY

Info sheet

Many people develop high cholesterol as they get older. But if you have familial hypercholesterolemia, or FH, your low-density lipoprotein (LDL) cholesterol is dangerously high early in life – often from birth. This happens because the body isn't able to get rid of the excess LDL cholesterol, also called the "bad" cholesterol.

Too much LDL cholesterol can clog arteries, which can lead to heart attacks and other heart issues.

Use this handout to learn more about FH, the two types, how they are treated, and questions to ask your health care team.

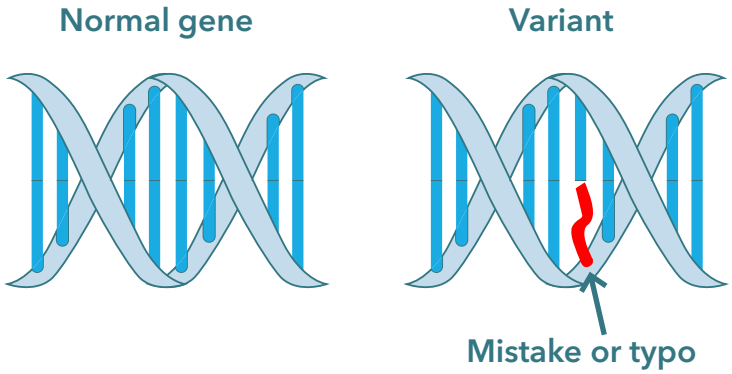
FH runs in families

FH is a genetic condition – that means it is passed down in families through the genes we inherit.

It happens when there is a change or typo (called a variant) in one of several genes that help instruct the body to recognize and clear cholesterol. You can get FH from one or both of your parents.

Without proper treatment, people with FH are much more likely to suffer a heart attack, cardiac arrest (when the heart stops suddenly) or stroke. Heart attacks and other heart issues happen at a much younger age for people with FH compared with others.

For some people, these events are the first clue that leads them to find out they have FH. For others, FH is suspected when LDL cholesterol levels aren't lowered as much as would be expected after starting treatment with lifestyle changes and medications.



Family screening

Finding out whether you or a family member has FH early on is important. Starting treatment as soon as possible can help prevent life-threatening events, such as heart attack or stroke.

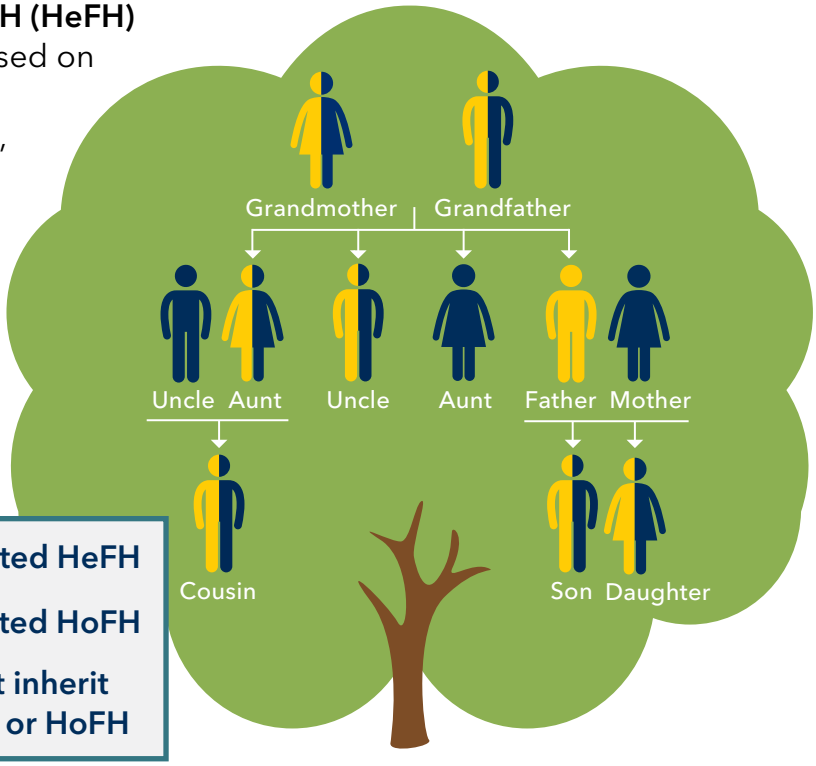
Ask about how to get family members tested for FH. Screening can save lives.

Two types of FH

There are two types of FH: **heterozygous FH (HeFH)** and **homozygous FH (HoFH)**, which are based on whether you inherited one or two FH gene variants from your birth parents. In general, if one of your parents has FH, you have a 50/50 chance of having it.

It's important to know which type of FH you have because treatments and the risk of heart problems can be different.

The type of FH you have also affects the likelihood of passing it on to a child.



How HeFH and HoFH differ

	Heterozygous FH	Homozygous FH
How common it is	• More common • Occurs in 1 in 250 adults	• Fairly rare • Occurs in 1 in 250,000 adults
What you inherit from your parents	• 1 healthy gene, 1 FH gene from a parent	• 2 FH genes – one from each parent
LDL cholesterol numbers Cholesterol levels are measured in milligrams (mg) of cholesterol per deciliter (dL) of blood.	• Higher than normal • Usually over 190 mg/dL	• Very high, often 4 times higher than normal levels • Usually over 400 mg/dL
How it affects heart health	• Heart attack, blocked arteries or stroke occur at a young age	• Heart issues and events early in life, sometimes in children

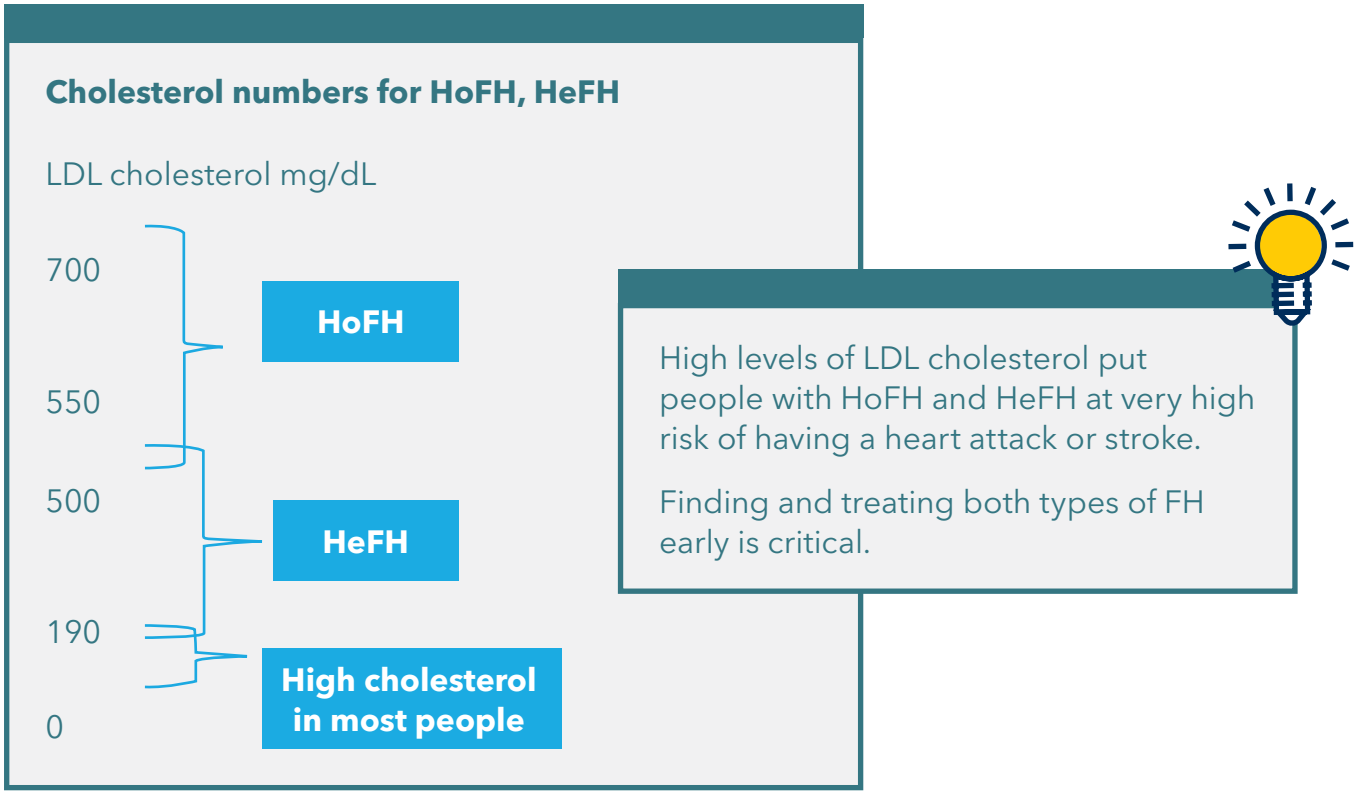
More about HoFH

HoFH is rarer than HeFH and often more severe. With HoFH:

- Symptoms and related heart events, including heart attack, usually show up earlier and may occur in childhood
- It tends to be harder to treat than HeFH
- Apheresis, a treatment to remove blood and filter LDL cholesterol out before returning it into the body (similar to dialysis for kidney disease), is commonly used
- A liver transplant may be needed in some cases

When FH might be suspected

- **A very high LDL cholesterol level** – above 190 mg/dL in adults, and above 160 mg/dL in children
- You or a family member develops heart disease, or has a **heart attack or stroke at a very young age**
- **Fatty deposits or bumps under the skin**, on the Achilles tendon (attaches your calf and heel), on the joints of the hand (called xanthomas), or under the skin of the eyelids (xanthelasmas)
- **White ring around the cornea** of the eye
- Results of **genetic testing** show one of the gene variants that can cause FH



Treatments to control cholesterol levels

The good news is that if you have FH, there are now many treatments available to help keep you healthy. It's important to start therapies to lower LDL cholesterol as soon as possible, even in childhood.

Treatment will include a combination of lifestyle changes, one or more medicines and/or a procedure called apheresis.

Getting treatment for FH early on can lower your risk of heart disease by about 80%.

Treating HeFH and HoFH

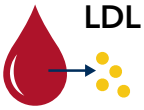
Lifestyle changes

- Eating a heart healthy diet
- Moving your body more, exercising
- Watching your weight
- Not smoking

Medications taken daily for the rest of your life

For example

- High-dose statins
- Ezetimibe
- Bile acid sequestrants
- Bempedoic acid
- Inclisiran
- PCSK9 inhibitor
- Lomitapide
- Evinacumab
- Mipomersen



LDL apheresis

Therapy to filter and remove LDL cholesterol from the blood if medications don't work well enough

HeFH → **HoFH**

Also important parts of care for HeFH and HoFH:

Ongoing monitoring, including frequent blood tests to check cholesterol levels and imaging to look for cholesterol and plaque buildup in the arteries.

Mental, emotional, and social support.

Questions to ask about your FH

- What type of FH do I have? How might this impact my care?
- Who in my family should be screened and why?
- How often will I need follow-up blood tests and checkups? What about imaging tests?
- Should we be adding some of the newer medications to my treatment? How do they work?
- Would taking part in a clinical study or registry be a good option for me?
- I know lifestyle changes are important to protect my heart. What should I focus on first?
- Is there more advice or support for living healthier and making behavior changes?