

HoFH

Homozygous Familial Hypercholesterolaemia



Could your extremely high levels of bad cholesterol (LDL-C) be putting you at risk for a heart-related event?

You could be living with an ultra-rare
inherited disease called **Homozygous Familial
Hypercholesterolaemia (HoFH)**

LDL-C=low-density lipoprotein cholesterol.

Understanding HoFH

Approximately 1 in 300,000 people are affected by HoFH.² Many of them are not diagnosed or are only diagnosed later in life.²



HoFH is an inherited condition that makes it difficult for your body to clear bad cholesterol (LDL-C)²

Having HoFH means you may have extremely high levels of bad cholesterol (LDL-C), reaching or exceeding levels **5x higher than target** (>10 mmol/L with HoFH vs 1.8 mmol/L target for adults with no cardiovascular risk)¹

You could be living with an ultra-rare inherited disease called Homozygous Familial Hypercholesterolaemia (HoFH)

Recognising HoFH can help you potentially get diagnosed and treated early



BAD CHOLESTEROL (LDL-C) LEVELS

Extremely high levels of bad cholesterol that can be difficult to control



GENETIC CONDITION

HoFH is inherited from your parents



VISIBLE SIGNS

Cholesterol buildup:

- Around the joints and on the tendons (xanthomas)
- In the eyes (corneal arcus) and around the eyelids (xanthelasma)

Adapted from Rocha and Wang et al.

If untreated, HoFH can cause cardiovascular events, even from childhood. Events include:



Heart attack

Stroke

Damage to the heart

Knowing the signs of HoFH can help you get a diagnosis and start treatment right away

It's important to diagnose HoFH early because early treatment reduces risk and prevents complications

Living with high levels of bad cholesterol (LDL-C) from HoFH can increase heart-related issues

AT BIRTH


Bad cholesterol levels are much higher than they should be, up to 18 mmol/L

Disease begins at birth⁴


DURING CHILDHOOD

Cholesterol begins to build up throughout the blood vessels and valves

In severe cases of HoFH, a first heart attack may occur before age 10⁴



The earlier you receive a diagnosis, the earlier you can start treatment to help lower your bad cholesterol levels



DURING ADOLESCENCE

Cholesterol buildup continues, narrowing and hardening the arteries and valves, and compromising the heart's function

Even with less severe cases of HoFH, major heart-related events may occur⁴

INTO ADULthood

Severe buildup hardens the arteries and valves, and may lead to heart attack and heart failure

Long-term outlook for survival gets worse, even with standard medical treatment⁴

2023 ESC/EAS LDL-C Reduction Target Recommendations¹

Children:

<3 mmol/L

Adults:

<1.8 mmol/L

Adults with ASCVD:

<1.4 mmol/L

ESC=European Society of Cardiology; EAS=European Atherosclerosis Society; ASCVD=atherosclerotic cardiovascular disease.

HoFH can have a serious effect on both your body and mind



In addition to heart-related risks, HoFH can affect you:



Physically:

Cholesterol buildup can cause joint pain and make moving painful³



Emotionally:

Getting an HoFH diagnosis can be overwhelming. It is normal to experience feelings of shame, fear, and loneliness

You could feel isolated because movement is painful and makes it hard to be active socially

These feelings are not a symptom of HoFH but a byproduct of it. Talk to your doctor if you need additional resources to help cope with the emotional side of HoFH³

HoFH is often underdiagnosed or misdiagnosed



Family history



A physical exam



A blood test

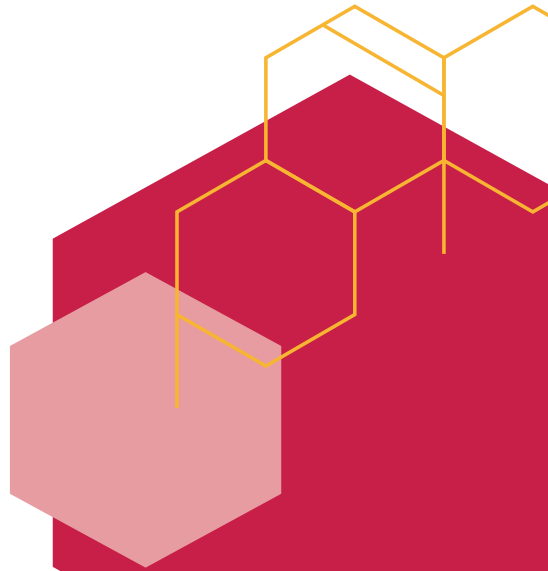
Once diagnosed, talk to your family members about HoFH

HoFH is an **inherited disorder**. There is a 1 in 4 chance that your siblings have HoFH and a 1 in 2 chance they have HeFH (a less severe form of FH)

If you suspect that you or a family member might have HoFH, consult your healthcare provider to screen for it

Guidelines suggest that healthcare providers use a process called reverse cascade screening to see which of your family members have FH

HeFH=heterozygous familial hypercholesterolaemia; FH=familial hypercholesterolaemia.



Treating HoFH early can lower serious risks to your health⁴

Here are a few things you can do:



Recognise HoFH early to help lower the risk of heart-related events, such as a heart attack



Get diagnosed as early as possible to begin the right treatment plan



Treat early to lower bad cholesterol (LDL-C) levels and help lower heart-related risks

Treatment options for HoFH may include:

- Lifestyle changes (diet, exercise, etc.)
- Medicines
- Emerging therapies
- Filtering out bad cholesterol (apheresis)
- Liver transplant (in extreme cases)



If your HoFH is undertreated, you may be at a more than 12x higher risk* for mortality from a heart-related event⁴

Talk to your healthcare provider about getting a diagnosis and creating an LDL-C treatment plan that is right for you

* Risk varies with patient characteristics, total cholesterol, and prior treatment.

HoFH may seem overwhelming, but you are not alone

Interested in learning more about how to live with HoFH?

The familial hypercholesterolaemia (FH) community can help you through your HoFH journey.



**FH Europe
Foundation**

Contact FH Europe Foundation to learn more about HoFH and the support that the community is providing to people living with HoFH and their families.

<https://fheurope.org/>

FH Europe Foundation is a patient network actively working to improve Europe-wide awareness, understanding, and access to diagnosis and treatment of inherited lipid conditions, including HoFH.

Check out its HoFH section (<https://fheurope.org/about-hofh/homozygous-familial-hypercholesterolaemia/>) **to find out more about living with HoFH.**

To find a FH patient group near you, see

<https://fheurope.org/who-we-are/network/>



HEART UK
THE CHOLESTEROL CHARITY

HEART UK

<https://www.heartuk.org.uk/>
ask@heartuk.org.uk

These organisations are listed for informational purposes only and are not intended to replace your healthcare professional's medical advice. Ask your doctor or nurse any questions you may have about your disease or treatment plan.

If untreated, HoFH can cause heart attacks and even mortality before age 30³

Lower your risk with an early diagnosis

Know the signs and symptoms of HoFH so you can start managing your condition:



Extremely high levels of bad cholesterol (LDL-C)



Heart attack or heart-related event at a young age



Family history of heart attacks at a young age and/or family history of FH



Cholesterol buildup in skin, tendons and eyes

Talk to your doctor about a diagnosis; with the right early diagnosis, you can take action to lower your bad cholesterol (LDL-C) with a treatment plan tailored to your condition

1. Cuchel M et al. *Eur Heart J*. 2023;00:1–15. 2. Tromp TR et al. *Lancet*. 2022;399:719–28. 3. Cuchel M et al. *Eur Heart J*. 2014;35:2146–2157. 4. Thompson GR et al. *Eur Heart J*. 2018;39:1162–1168. 5. Rocha V et al. *J Am Coll Cardiol*. 2013;61(21):2193. 6. Wang N et al. *J Med Case Rep*. 2022;16(1):290.