

# HoFH

Homozygous  
Familial  
Hypercholesterolemia



**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 Hypercholesterolemia (HoFH)**

***Know the signs of HoFH so you can talk to your healthcare provider about a possible diagnosis and treatment options.***

# Understanding HoFH

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



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

**Your bad cholesterol could be more than 5x the normal level.**

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

† Lower LDL-C levels, especially in children or in people already on treatment, do not rule out HoFH. LDL-C=low-density lipoprotein cholesterol; HeFH=heterozygous familial hypercholesterolemia.

# Recognizing HoFH can help you 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 (xanthelasmas)

Untreated, HoFH can cause cardiovascular events, even at a young age. Events include:



Damage to the heart



Stroke



Heart attack

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

# It's important to diagnose HoFH early because the risks can get worse over time

**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 birth**



## 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 10**



**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 the arteries and compromising the heart's function

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

## INTO ADULTHOOD

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

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



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## ESC/EAS guideline recommendations:

LDL-C level goal  
for children:

**<3.5 mmol/L**

LDL-C level goal  
for adults:

NORMAL  
**<2.5 mmol/L**

HIGH CV RISK  
**<1.8 mmol/L**

**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 painful



### **Emotionally:**

You could experience feelings of shame, fear, and being alone

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

# HoFH is often underdiagnosed or misdiagnosed



Family history



A physical exam



A blood test

## Once diagnosed, talk to your family members about HoFH

Guidelines suggest healthcare providers use a practice called **cascade screening** to see if your family members could have FH, including HoFH. This process involves using genetic testing to determine if other family members have HoFH.

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

# Treating HoFH early can lower serious risks to your health

Here are a few things you can do:



Recognize 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 to lower bad cholesterol (LDL-C) levels to help lower heart-related risks

## Treatment options for HoFH may include:

- Lifestyle changes (diet, exercise, etc)
- Medicines (statins, PCSK9 inhibitors, others)
- Emerging therapies
- Filtering out bad cholesterol (apheresis)
- Liver transplant (extreme cases)



**If your HoFH is undertreated, you may be at a more than 10x higher risk\* for death from a heart-related event.**

**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. PCSK9=proprotein convertase subtilisin/kexin type 9.





# HoFH may seem overwhelming, but you are not alone

**Many tools and resources are available to help you along the way**

The familial hypercholesterolemia (FH) community is active and supportive, and can help you throughout your HoFH journey.

**Use these resources to learn more about HoFH and connect with the FH community:**

**FH Canada:**

<https://www.FHcanada.net>

**Family Heart Foundation:**

<https://familyheart.org/>

**EAS Consensus on HoFH:**

<https://eas-society.org/page/consensus-on-hofh/>

**EAS Familial Hypercholesterolaemia Studies Collaboration (FHSC):**

<https://beta.clinicaltrials.gov/study/NCT04272697>

<https://fheurope.org/about-hofh/homozygous-familial-hypercholesterolaemia/>


**Worldwide experience of HoFH, retrospective cohort study published in The Lancet:**

<https://eas-fhsc.org/publications/>

**Find an FH specialist to help you along your treatment journey**

**FH Canada:**

<https://www.FHcanada.net>



These organizations are an incomplete listing of rare disease advocacy groups. The list is meant for informational purposes only and is 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.

# Untreated, HoFH can cause heart attacks and even death before age 30

**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 joints 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**



**Scan the QR code to learn more at [HoFHdisease.ca](https://www.HoFHdisease.ca)**

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