Homozygous Familial Hypercholesterolemia (HoFH)
Is HoFH Treatable?

HoFH is treatable. Managing HoFH requires consistent therapeutic intervention and the expertise of a lipid specialist.

What is HoFH?

HoFH (or Homozygous Familial Hypercholesterolemia) is an inherited disorder. HoFH severely or completely disrupts the body’s ability to clear LDL-cholesterol from the blood. This leads to atherosclerosis (narrowing and blocking of blood vessels), early coronary artery disease and valvular heart disease. If left untreated, HoFH can cause heart attacks or sudden death as early as childhood and young adulthood.

HoFH is the most severe and least common form of a disease known as FH (Familial Hypercholesterolemia). It occurs when the FH gene is inherited from both parents.

90% of individuals with FH are not properly diagnosed. Therefore, they may not know if they have passed it on to their children.

How do I know if I have HoFH?

• Abnormally high LDL-cholesterol levels from an early age (usually above 500 mg/dL if cholesterol untreated).

• Family history of early heart disease and heart attacks.

• Bumps or lumps on the skin around the knuckles, elbows and knees (xanthomas). These may be noticed by a dermatologist.

• Swollen or painful Achilles tendons (tendon xanthomas).

• Yellowish areas around the eyes (xanthelasmas) or a white arc near the colored part of the eye (corneal arcus). These may be noticed by an ophthalmologist.

Remember that not everyone with HoFH has the same signs and symptoms. If you recognize any of the above signs, please consult your health care provider.

“It is not easy being diagnosed with FH, but this is a time of great hope. Many people with HoFH can lead happy, healthy lives with treatment.”

- Samir Elias
  FH Foundation Advocate & HoFH patient
It’s important to remember that HoFH is a serious medical condition and is life-threatening. HoFH leads to progressive and early heart disease. That’s why if you or your child are diagnosed with HoFH, one of the first steps to take – as soon as possible – is to consult a lipid specialist (an expert in lipid/cholesterol disorders). If a couple knows they both have FH, they should consult a lipid specialist to check their children’s cholesterol by 6 months of age. Prenatal diagnosis is also possible.

How Do You Treat HoFH?

There are treatments specifically for HoFH:

• LDL-apheresis - a mechanical procedure that removes cholesterol from the bloodstream
• Juxtapid (lomitapide) - a daily pill
• Kynamro (mipomersen) - a weekly injection

*Some cholesterol-lowering drugs have not yet been tested on young children. Consult your healthcare provider for more information.

Early diagnosis and treatment can be crucial to saving a life.

Be sure to speak with your doctor about what therapy is right for you.

HoFH RUNS IN FAMILIES

Think about your own family history.

Are early heart attacks, early heart disease, or very high cholesterol common in your family? If so, speak with a cardiologist, lipid specialist or genetic counselor to investigate whether you may have FH or HoFH in your family.
THE FH FOUNDATION IS HERE FOR YOU

The FH Foundation is a patient-centered nonprofit organization dedicated to education, advocacy, and research of all forms of Familial Hypercholesterolemia (FH). Our mission is to raise awareness and save lives by increasing the rate of early diagnosis and encouraging proactive treatment. If left untreated, FH leads to aggressive and early heart disease in women, men and children of all racial and ethnic backgrounds.

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