



Confirm a diagnosis of Homozygous Familial Hypercholesterolemia (HoFH)

Sponsored Genetic Testing

Extremely high levels of low-density lipoprotein cholesterol (LDL-C) may be a sign of homozygous familial hypercholesterolemia (HoFH). A rare condition, HoFH is often undiagnosed or misdiagnosed. HoFH occurs when an individual inherits two FH gene variants; one from each parent. Clinical findings are variable but may include cholesterol deposits in the tendons and joints (xanthomas) or in the eye (corneal arcus). HoFH can be diagnosed clinically or genetically; for patients and families, genetic testing may confirm an HoFH diagnosis in instances where there is insufficient information to diagnose clinically.^{1,2,3}

Regeneron Pharmaceuticals is sponsoring no-cost genetic testing through Ambry Genetics to help confirm a diagnosis of HoFH.

Testing Eligibility

Patients must be in the United States and meet one criteria from Part A and one criteria from Part B

Part A:

- Untreated LDL-C >300 mg/dL
- Treated LDL-C >250 mg/dL on one lipid-lowering therapy
- Treated LDL-C >200 mg/dL on two or more lipid-lowering therapies
- Other LDL-C level and treatment profile consistent with HoFH

Part B:

- Family history of FH or premature coronary artery disease (<55 yrs for males and <65 yrs for females)
- Personal history of premature coronary artery disease (<55 yrs for males and <65 yrs for females)
- Personal history of tendonous and or cutaneous xanthomas
- Personal history of corneal arcus before age 45 years
- Personal history of aortic stenosis

Find the underlying genetic cause of a patient's elevated cholesterol.



How Does the Program Work?

This program utilizes Ambry's FHNext 4-gene panel aimed at identifying an underlying genetic cause of unexplained elevated cholesterol levels.

The 4 genes included are associated with both heterozygous (HeFH) and homozygous (HoFH) forms of familial hypercholesterolemia. Specimens are collected via an easy-to-use saliva collection kit. The program includes no-cost genetic counseling for the patient to discuss the test, review results and guide next steps.

Genes tested with FHNext:

APOB LDLR PCSK9 LDLRAP1

Points for Your Practice

1. For patients and families, genetic testing may confirm an HoFH diagnosis in instances where there is insufficient information to diagnose clinically.^{1,2}
2. Cases of HoFH have been identified through genetic testing that had been previously misdiagnosed *clinically* as heterozygous FH (HeFH).³
3. Genetic counseling for patients is recommended before and after genetic testing for FH.⁴

How it Works

1. Genetic testing is done using a saliva sample, which can be collected in the office or by shipping a kit to the patient.
2. Genetic testing analyzes DNA to identify mutations in the 4 known genes associated with HoFH.
3. It takes 2-3 weeks for the testing to be completed and results are sent to the ordering healthcare provider via Ambry Portal or fax.

References

1. Gidding SS et al. "The Agenda for Familial Hypercholesterolemia, A Scientific Statement from the American Heart Association", *Circulation*, Vol 132, No 22, 2015.
 2. Cuchel M. et al. "2023 Update on European Atherosclerosis Society Consensus Statement on Homozygous Familial Hypercholesterolemia: new treatments and clinical guidance," *European Heart Journal*, 2023; ehad197
 3. Chaudhry A et al. "Genetic Identification of Homozygous Familial Hypercholesterolemia by Long-Read Sequencing Among Patients With Clinically Diagnosed Heterozygous Familial Hypercholesterolemia", *Circulation*, Vol. 16, No. 2, 2023.
 4. Brown EE et al. "Genetic testing in dyslipidemia: A scientific statement from the National Lipid Association", *Journal of Clinical Lipidology*, Vol 14, No 4, 2020.
- Abbreviations:** APOB, apolipoprotein B; LDLR, low-density lipoprotein receptor; PCSK9, proprotein convertase subtilisin/kexin type 9; LDLRAP1, low-density lipoprotein receptor adaptor protein 1

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