An **early diagnosis** can make a lifetime of difference

Quest Diagnostics™

Most FH goes undiagnosed. **Genetic testing** can change that.

9 in 10 familial hypercholesterolemia (FH) cases go undiagnosed[1]

Diagnose today to treat tomorrow—and control risk long-term*

Early genetic testing can benefit affected patients and their families.

Genetic testing for FH from Quest examines 3 actionable[2-6] FH genes: **LDLR**, **APOB**, and **PCSK9**, to enable a definitive diagnosis of FH.

Treatment: The consensus conclusion is that if diagnosed and treated early in childhood, individuals with FH can have normal life expectancy (Wiegman, *et al*., 2015).[7]

FH testing from Quest Diagnostics provides actionable information that can lead to treatment and lower lifetime risk of cardiovascular disease.

"Early identification and control of dyslipidemia throughout youth and into adulthood will substantially reduce clinical CVD risk beginning in young adult life."

— Expert panel, *Pediatrics*, 2011[8]

* These points are provided for informational purposes only and are not intended as medical advice. A physician's test selection and interpretation, diagnosis, and patient management decisions should be based on his/her education, clinical expertise, and assessment of the patient.

FH testing can lead to knowing sooner so cardiovascular care can be optimized

Why family members of affected individuals should be tested:

- 50% of first-degree relatives of heterozygous FH index cases are affected[9]
- American Academy of Pediatrics (AAP) recommends screening affected children after age 2, but no later than age 10[10]

Earlier treatment may include pharmacologic intervention[10]

Guidelines state that statins should be considered for patients 8 years and older with:

An LDL-C of **≥190 mg/dL** **OR** An LDL-C of **≥160 mg/dL** with a family history of early heart disease **OR** ≥2 additional risk factors present or LDL-C **≥130 mg/dL** if diabetes mellitus is present

... if dietary and lifestyle modifications fail to achieve treatment goals.[11] Also, for patients who need additional LDL-C lowering after being on a controlled diet and maximally tolerated statin therapy, PCSK9 inhibitors should be considered.[12]

FH cascade screening is highly recommended by multiple evidence groups to enable earlier insight and intervention[13]

Cascade screening for FH:

- Can reduce the average age at which individuals with FH are diagnosed
- Can increase the percentage of individuals taking lipid-lowering therapies, potentially resulting in reductions in LDL-C and coronary heart disease

Early **FH testing** and cascade screening can give patients and their families a chance for healthier cardiovascular futures.

A diagnosis that may ensure a **long-term difference**

Make an early and definitive diagnosis of familial hypercholesterolemia to start therapy sooner.

Ordering information

Test Name	Test Code	CPT Code(s)*
Familial Hypercholesterolemia (FH) Panel	94877	81405, 81406(x2), 81479(x1)
FH Single Site	94878	81403

* The CPT codes provided are based on AMA guidelines and are for informational purposes only. CPT coding is the sole responsibility of the billing party. Please direct any questions regarding coding to the payer being billed.

For more information, including other cardiometabolic testing, contact your Quest Diagnostics representative or visit us at **QuestDiagnostics.com/Education** and **4myheart.com.**

References

1. Nordestgaard BG, Chapman MJ, Humphries SE, *et al.* Familial hypercholesterolaemia is underdiagnosed and undertreated in the general population: guidance for clinicians to prevent coronary heart disease: consensus statement of the European Atherosclerosis Society. *Eur Heart J.* 2013;34(45):3478-3490a.
2. Varret M, Abifadel M, Rabès JP, Boileau C. Genetic heterogeneity of autosomal dominant hypercholesterolemia. *Clin Genet.* 2008;73:1-13.
3. Ahmad Z, Adams-Huet B, Chen C, Garg A. Low prevalence of mutations in known loci for autosomal dominant hypercholesterolemia in a multiethnic patient cohort. *Circ Cardiovasc Genet.* 2012; 5:666-75.
4. Rader DJ, Cohen J, Hobbs HH. Monogenic hypercholesterolemia: new insights in pathogenesis and treatment. *J Clin Invest.* 2003;111:1795-804.
5. Wang J, Ban MR, Hegele RA. Multiplex ligation-dependent probe amplification of LDLR enhances molecular diagnosis of familial hypercholesterolemia. *J Lipid Res*. 2005;46(2):366-72.
6. Fouchier SW, Dallinga-Thie GM, Meijers JC, *et al.* Mutations in STAP1 are associated with autosomal dominant hypercholesterolemia. *Circ Res*. 2014;115:552-5.
7. Wiegman A, Gidding SS, Watts GF, *et al*. Familial hypercholesterolaemia in children and adolescents: gaining decades of life by optimizing detection and treatment. *Eur Heart J.* 2015;36:2425-37.
8. Expert panel on integrated guidelines for cardiovascular health and risk reduction in children and adolescents: summary report. *Pediatrics*. 2011;128 Suppl 5:S213-56. doi: 10.1542/peds.2009-2107C.
9. Ned RM, Sijbrands EJG. Cascade screening for familial hypercholesterolemia (FH). Version 1. *PLoS Curr*. 2011 May 23; 3: RRN1238; doi: 10.1371/currents.RRN1238.
10. Daniels SR, MD, Greer FR and the Committee on Nutrition. Lipid screening and cardiovascular health in childhood. *Pediatrics*. 2008;122(1):199-208.
11. Gidding SS, Wiegman A, Watts G, Chapman J. Pediatric familial hypercholesterolemia: expert analysis. American College of Cardiology. Available at acc.org/latest-in-cardiology/articles/2015/11/24/14/32/pediatric-familial-hypercholesterolemia. Accessed October 20, 2017.
12. Chaudhary R, Garg J, Shah N, *et al.* PCSK9 inhibitors: A new era of lipid lowering therapy. *World J Cardiol*. 2017;9(2):76-91.
13. Centers for Disease Control and Prevention. Cascade screening for familial hypercholesterolemia in the United States: public health impact and challenges. Available at blogs.cdc.gov/genomics/2017/07/25/cascade_screening. Accessed September 29, 2017.

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