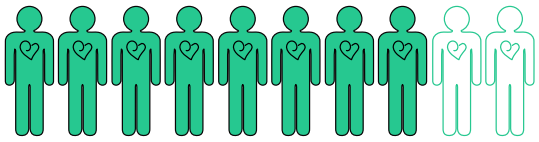
Innovative Gx
Familial Hypercholesterolemia Genetic Testing



80%

Patients with familial hypercholesterolemia who have yet to be diagnosed. Genetic testing provides a definitive diagnosis for patients with high LDL cholesterol levels.

What is Familial Hypercholesterolemia (FH)?

High cholesterol is a multifactorial condition influenced by both genetic and environmental factors, such as diet and lifestyle. There are dozens of genes that regulate levels of cholesterol in the blood. For some patients with high cholesterol, their disease is driven by a single gene. FH is an inherited form of high LDL cholesterol. An estimated 1 in 250 people have FH across all ethnicities. Most patients with FH have yet to be diagnosed and show no signs beyond high LDL cholesterol levels. Genetic testing is the gold standard of care for diagnosing patients with suspected FH and their at-risk relatives. When a patient is diagnosed with FH, genetic testing should then be offered to their first-degree relatives, including children, regardless of the their LDL cholesterol level.[1]

Who should get tested?

	SHOULD BE OFFERED:		CAN BE CONSIDERED:	
ADULTS	>250 mg/dl LDL-C level*	>190 mg/dl LDL-C level* AND At least 1 first-degree relative similarly affected or with premature CAD OR family history unavailable	Personal history of premature CAD, no LDL-C levels* available, and family history of both hypercholesterolemia and premature CAD	>160 mg/dl LDL-C levels*, either a personal history or family history of premature CAD, and a family history of hypercholesterolemia
CHILDREN	>190 mg/dl LDL-C level*	>160 mg/dl LDL-C level* AND At least 1 first-degree relative similarly affected or with premature CAD or family history unavailable	>160 mg/dl LDL-C level* AND >190 mg/dl LDL-C level* in at least 1 parent or a family history of hypercholesterolemia and premature CAD	

**Pretreatment levels without an apparent secondary cause of hypercholesterolemia; Premature coronary artery disease (CAD) defined as male subjects 55 years or younger and female subjects 65 years or younger*

Test Name	Genes
Innovative Gx Familial Hypercholesterolemia Panel	*APOB, LDLR, LDLRAP1* and *PCSK9*

References

1. Sturm AC, Knowles JW, Gidding SS, Ahmad ZS, Ahmed CD, Ballantyne CM, Baum SJ, Bourbon M, Carrié A, Cuchel M, de Ferranti SD, Defesche JC, Freiberger T, Hershberger RE, Hovingh GK, Karayan L, Kastelein JJP, Kindt I, Lane SR, Leigh SE, Linton MF, Mata P, Neal WA, Nordestgaard BG, Santos RD, Harada-Shiba M, Sijbrands EJ, Stitziel NO, Yamashita S, Wilemon KA, Ledbetter DH, Rader DJ; Convened by the Familial Hypercholesterolemia Foundation. Clinical Genetic Testing for Familial Hypercholesterolemia: JACC Scientific Expert Panel. J Am Coll Cardiol. 2018 Aug 7;72(6):662-680. doi: 10.1016/j.jacc.2018.05.044. PMID: 30071997.