Ambry Genetics®
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Hereditary Lipid Disorders and Associated Tests

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INDICATIONS FOR TESTING	TEST NAME	GENES	CHARACTERIZED BY:	GENETIC TESTING SUGGESTED FOR PATIENTS WHO HAVE:
Familial hypercholesterolemia	FHNext	*APOB, LDLR, PCSK9, LDLRAP1*	• High levels of LDL-C in the blood • Increased risk of heart attacks and stroke at a younger than usual age	• Children: LDL-C levels >160 mg/dL and > 1 first-degree relative similarly affected or LDL-C levels >190 mg/dL even in the absence of family history • Adults: LDL-C levels >190 mg/dL and >1 first-degree relative similarly affected or LDL-C levels >250 mg/dL even in the absence of family history
Sitosterolemia	Sitosterolemia	*ABCG5, ABCG8*	• Inability to metabolize plant sterols • Increased risk of coronary artery disease and heart attack	• Premature artherosclerosis, coronary artery disease, and/or heart attack • Negative FH testing AND • Normal to mildly elevated LDL-C levels
Familial chylomicronemia syndrome	FCSNext	*APOA5, APOC2, GPIHBP1, LMF1, LPL*	• Extremely high blood triglycerides • Eruptive xanthomas • Recurrent acute pancreatitis	• High triglycerides (TG/TC ratio > 5 mg/dL) • Have ≥1 episode of pancreatitis prior to age 40
Other hereditary lipid disorders	CustomNext-*Cardio*	Choose from up to 18 genes associated with hereditary lipid disorders: *ABCA1, ABCG5, ABCG8, APOA1, APOA5, APOB, APOC2, APOC3, APOE, CYP27A1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIPA, LMF1, LPL, PCSK9*	**TEST FOR THE FOLLOWING LIPID DISORDERS** • Familial HDL deficiency • Lysosomal acid lipase deficiency • LCAT deficiency/Fish-eye disease • Hyperlipoproteinemia type III • Cerebrotendinous xanthomastosis (CTX) • Apolipoprotein C-III deficiency	

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