Ambry Genetics®
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Lipid Menu

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Genetic testing can be important in screening, diagnosis, and potentially in treatment of lipid disorders. Knowing if your patient has a lipid disorder can help you determine their future coronary artery disease risk and guide your medical management recommendations. Genetic testing can confirm a diagnosis, particularly when clinical criteria are unclear or borderline in an individual. It can also help tailor medical treatment and clarify risks to family members, including the inheritance pattern.

INDICATIONS FOR TESTING	TEST NAME	GENES
Familial Chylomicronemia Syndrome (FCS)	FCSNext	*APOA5, APOC2, GPIHBP1, LMF1, LPL*
Sitosterolemia	Sitosterolemia	*ABCG5, ABCG8*
Personal or Family History of Hypercholesterolemia	FHNext	*APOB, LDLR, PCSK9, LDLRAP1*
Other Hereditary Lipid Disorders	CustomNext-*Cardio*	*ABCA1, ABCG5, ABCG8, APOA1, APOA5, APOB, APOC2, APOC3, APOE, CYP27A1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIPA, LMF1, LPL, PCSK9*

Familial Chylomicronemia Syndrome (FCS)

- Characterized by extremely high blood triglycerides, recurrent acute pancreatitis and eruptive xanthomas.
- Genetic testing is recommended for patients with high triglycerides (TG/TC ratio > 5 mg/dL), one or more episodes of pancreatitis prior to age 40

Sitosterolemia

- Characterized by inability to metabolize plant sterols and an increased risk of coronary artery disease and heart attack
- Genetic testing is recommended for patients with premature atherosclerosis, coronary artery disease, and/or heart attack who have negative FH testing and normal to mildly elevated LDL

Other Lipid Disorders

Choose from an 18 gene lipid menu to customize a test through CustomNext-*Cardio*. This customizable form will give you the ability to test for the following rare 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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