# Hereditary Lipid Disorders and Associated Tests

<table>
<thead>
<tr>
<th>INDICATIONS FOR TESTING</th>
<th>TEST NAME</th>
<th>GENES</th>
<th>CHARACTERIZED BY:</th>
<th>GENETIC TESTING SUGGESTED FOR PATIENTS WHO HAVE:</th>
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</table>
| 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 atherosclerosis, 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

<table>
<thead>
<tr>
<th>GENES</th>
<th>Choose from up to 18 genes associated with hereditary lipid disorders: ABCA1, ABCG5, ABCG8, APOAI, APOA5, APOB, APOC2, APOC3, APOE, CYP27A1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIPA, LMF1, LPL, PCSK9</th>
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| 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 |