

*Identify children and adults at risk for  
premature coronary heart disease.*

## Hypercholesterolemia Evaluation

*Your patients' hypercholesterolemia may  
have a genetic cause.*

- Familial Hypercholesterolemia (FH) is due to autosomal dominant loss-of-function mutations in the gene *LDLR*, which codes for the low-density lipoprotein receptor (LDLR).<sup>1</sup>
  - FH-associated mutations in *LDLR* occur in about 1:500 individuals.<sup>2</sup>
- Familial Defective Apolipoprotein B-100 (FDB) is due to autosomal dominant loss-of-function mutations in the gene *APOB*, which codes for apolipoprotein B-100, the principal protein component of low-density lipoprotein (LDL).<sup>3,4</sup>
  - FDB-associated mutations in *APOB* occur in about 1:1000 individuals.<sup>2</sup>
- FH and FDB are characterized by elevated plasma levels of total and LDL-cholesterol and are clinically indistinguishable. Both are associated with atherosclerosis, which may begin in childhood, and premature coronary heart disease.<sup>5,6</sup>

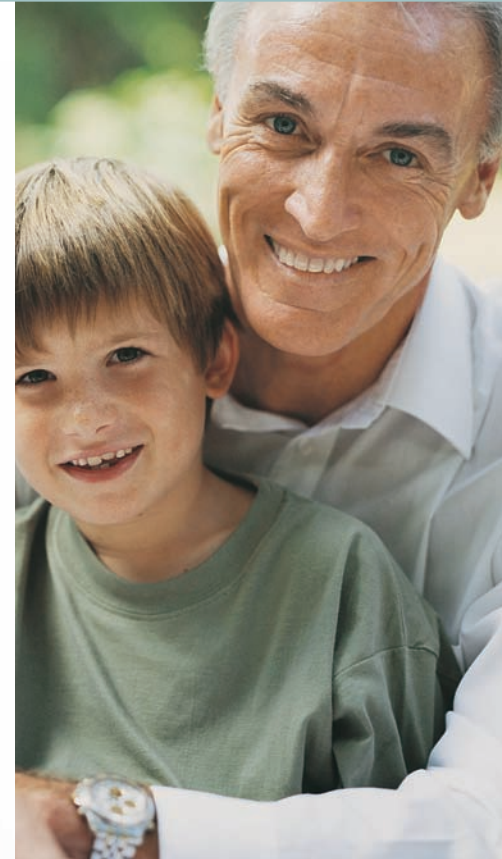
*Why genetic testing?*

### **Genetic testing for hypercholesterolemia-associated mutations in *LDLR* or *APOB***

- Can enable a diagnosis of FH or FDB, which may require more aggressive drug therapy than hypercholesterolemia due to other causes.<sup>7</sup>
  - Early intervention may prevent or repair atherosclerotic damage and lower the risk of coronary heart disease.
  - Recent clinical trials have indicated the safety and potential efficacy of statin use in children and adolescents.<sup>8,9</sup>
  - Knowledge of a predisposition for hypercholesterolemia and premature coronary heart disease may provide powerful motivation for lifestyle changes.
- Is the most reliable method for family screening for FH and FDB.<sup>10,11</sup>
  - FH and FDB are known to be underdiagnosed, and diagnosis typically does not occur until middle age.<sup>7</sup>
  - Family testing can allow early diagnosis and intervention which may prevent or delay onset of coronary heart disease.<sup>7</sup>

*Indications for testing:*

- Elevated plasma cholesterol levels
- Family history of premature coronary heart disease
- Family history of hypercholesterolemia



Testing performed under license from

Correlagen®

Correlagen Diagnostics, Inc.  
www.correlagen.com

athena diagnostics®

*Testing You Can Count On.*

**References:** 1. Brown MS, Goldstein JL (1986) *Science* 232:34-47. 2. Austin MA, et al., (2004) *Am J Epidemiol* 160:407-20. 3. Innerarity TL, et al., (1987) *Proc Natl Acad Sci USA* 84:6919-23. 4. Soria LF, et al., (1989) *Proc Natl Acad Sci USA* 86:587-91. 5. Wiegman A, et al., (2004) *Lancet* 363:369-70. 6. Austin MA, et al., (2004) *Am J Epidemiol* 160:421-9. 7. Marks D, et al., (2003) *Atherosclerosis* 168:1-14. 8. Hedman M, et al., (2005) *J Clin Endocrinol Metab* 90:1942-52. 9. Wiegman A, et al., (2004) *JAMA* 292:331-7. 10. Leren TP, (2004) *Clin Genet* 66:483-7. 11. Umans-Eckenhausen MAW, et al., (2001) *Lancet* 357:165-168.

For a brief review on FH/FDB, please visit [www.athenadiagnostics.com/DR](http://www.athenadiagnostics.com/DR)



www.athenadiagnostics.com

## Hypercholesterolemia Evaluation

**Typical Presentation:** Elevated plasma levels of total cholesterol and LDL-cholesterol; tendon xanthomata; premature coronary heart disease (CHD)

**Indications for Testing:**

- Elevated plasma cholesterol levels
- Family history of premature CHD
- Family history of hypercholesterolemia

### TEST DETAILS

**Test Code:** 895

**Test Includes:** *APOB* (Hypercholesterolemia) Mutation Analysis, #893  
*LDLR* (Hypercholesterolemia) DNA Sequencing Test, #894

**Test Turnaround:** 14-21 days

### TECHNICAL INFORMATION

**Methodology:** Polymerase Chain Reaction (PCR), DNA sequencing of:

- *LDLR*: entire protein coding region
- *APOB*: coding region where known FDB-associated mutations are located

### SHIPPING CONSIDERATIONS

**Specimen Type:** Whole blood

**Volume:** 10 mL (pediatric minimum: 2 mL)

**Collection Tube:** Yellow or lavender top

**Stability:** Hemolysis may compromise DNA recovery and integrity after 48 hrs

**Storage Conditions:** For short periods (until shipped) at 4°C

**Shipping Conditions:** Overnight at room temperature (specimen arrival must be less than 24 hrs after collection); ship Monday through Thursday only

To order the Hypercholesterolemia Evaluation (Test #895), call Athena Diagnostics Customer Service Representatives at:

**800-394-4493 x2**



*Testing You Can Count On.*

Four Biotech Park, 377 Plantation Street  
Worcester, MA 01605 • [www.AthenaDiagnostics.com](http://www.AthenaDiagnostics.com)