

Using Genetic Testing to Detect Familial Hypercholesterolemia and Increased Risk of Coronary Heart Disease

Indications:

- Presence or family history of early-onset coronary heart disease (CHD) (before 45 years in males and before 55 years in females)
- Presence or family history of tendon xanthomas
- Presence or family history of (untreated) total plasma cholesterol levels above 290 mg/dL (7.5 mmol/L) in adults or above 260 mg/dL (6.7 mmol/L) in children under 16 (plasma LDL cholesterol levels above 190 mg/dL (4.9 mmol/L) in adults or above 155 mg/dL (4 mmol/L) in children under 16)

Benefits:

Genetic testing can facilitate

- diagnosis of Familial Hypercholesterolemia (FH) in relatives of patients.
- timely treatment to prevent coronary heart disease.

Background:

- FH has been linked to atherosclerosis and a dramatically increased risk of early-onset CHD.¹
- FH is associated with autosomal dominant loss-of-function mutations in *LDLR* or *APOB*, which occur at a prevalence of 1:500 and 1:1000, respectively.²
- Much of FH-associated CHD can be delayed or prevented through timely treatment.³⁻⁷
- FH is both underdiagnosed and undertreated, especially in the young.^{8,9}
- Family testing can increase diagnosis and treatment levels.¹⁰⁻¹²
- Genetic testing is more sensitive and more specific than biochemical testing for identifying affected relatives of patients.^{10,13}

References: 1. Austin MA et al (2004) *Am J Epidemiol* 160:407-20. 2. Austin MA et al (2004) *Am J Epidemiol* 160:421-9. 3. de Sauvage Nolting PR, et al (2002) *Am J Cardiol* 90:181-4. 4. Smilde TJ et al (2001) *Lancet* 357:577-81. 5. Wiegman A et al (2004) *JAMA* 292:331-7. 6. Hedman M et al (2005) *J Clin Endocrinol Metab* 90:1942-52. 7. Austin MA et al (2002) *Genet Med* 4:275-8. 8. WHO-Human Genetics Programme. Report of a second WHO consultation. Geneva, 1999. 9. Neil HAW et al (2000) *BMJ* 321:148. 10. Umans-Eckenhausen MAW et al (2001) *Lancet* 357:165. 11. Fouchier et al (2005) *Hum Mutat* 26:550. 12. Leren et al (2004) *Semin Vasc Med* 4:75. 13. Damgaard D et al (2005) *Atherosclerosis* 180:155-60.

Ordering Information: please see other side



Ordering Information for the *LDLR* and *APOB* DNA Sequencing Tests (Early-Onset CHD)

Indications for Testing

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Ordering Information

Gene(s)	CPT Codes	Test Code
<i>LDLR</i>	83891(1) 83892(1) 83898(18) 83904(36) 83909(36) 83912(1)	190101
<i>APOB</i>	83891(1) 83892(1) 83898(1) 83904(2) 83909(2) 83912(1)	190102
Multi-Gene Panel*		
<i>LDLR + APOB</i>	83891(1) 83892(1) 83898(19) 83904(38) 83909(38) 83912(3)	190199

* For the multi-gene panel, a summary report will be issued in addition to an abbreviated report for each individual gene.

Family Testing (single amplicon)

<i>LDLR</i>	83891(1) 83892(1) 83898(1) 83904(2) 83909(2) 83912(1)	190101
<i>APOB</i>	83891(1) 83892(1) 83898(1) 83904(2) 83909(2) 83912(1)	190102

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of protein-coding region

Sample Requirements

- For blood samples:
 - 2 mL whole blood in EDTA tube (lavender top)
 - Samples can be stored briefly at 4°C, but should be shipped on day of collection.
- For buccal swab samples:
 - Please contact client services at 1-866-647-0735 for instructions.
- All sample types should be shipped overnight at room temperature.
- To request a sample shipping kit, please call 1-866-647-0735.

Turn-around Times

Turn-around times typically range from 7 to 21 days of receipt of sample and all required forms, but may vary depending on test volume and test-specific technical difficulties. Current TATs are posted on our website. Please schedule patient follow-up appointments for discussion of test results conservatively at 6 weeks.

For more information, please contact Correlagen Diagnostics, Inc., at 1-866-647-0735 or visit us on the web at www.correlagen.com.