

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** Familial Hypercholesterolemia Genetic Panel
- **Test Code:** 3880

Related Diseases & Genes:

- HYPERCHOLESTEROLEMIA, FAMILIAL (OMIM #: 143890) || Gene: *LDLR* (OMIM #: 606945)
- Hypercholesterolemia, familial, 3 (OMIM #: 603776) || Gene: *PCSK9* (OMIM #: 607786)
- Hyperlipoproteinemia, type III (OMIM #:) || Gene: *APOE* (OMIM #: 107741)
- Hypercholesterolemia, due to ligand-defective apo B (OMIM #: 144010) || Gene: *APOB* (OMIM #: 107730)
- Hypobetalipoproteinemia (OMIM #: 615558)

Synonyms:

- FHC; FH
- LDL RECEPTOR DISORDER

Test Details:

- **Methodology:** Next Generation DNA Sequencing
- **Performed:** Everyday
- **Reported:** 4 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Cardiology

