



Cardiology Genetics Testing Solutions

Familial Hypercholesterolemia (FH) Test Offering Code

Familial Hypercholesterolemia Panel 94877

LDLR, APOB, PCSK9 sequencing and deletion/duplication

Familial Hypercholesterolemia, Single Site 94878

Specific variant testing for a known familial variant or variants in *LDLR, APOB, and/or PCSK9*

Records documenting the variant must be provided.

Testing and Services Overview

Quest Diagnostics offers a portfolio of cardiac testing, including genetic testing for familial hypercholesterolemia (FH). It gives practices the flexibility of multiple test options for focused or comprehensive risk testing, a lab that's simple to work with, and access to expert-guided education tools. The value to you: improved actionability, efficiency, and patient engagement. Because a test result is only as good as the action it inspires.

Ordering and Reporting

Ordering and reporting can be done by paper or electronically, using Quantum eLabs® or other EMR interfaces. Required sample volume is 2–5mL. Specimens can be collected with standard 4mL tubes, or at one of our more than 2,200 Patient Service Centers.

Turnaround Time

Two to 3 weeks upon receipt of sample in the laboratory if the clinical history form and order are complete, and the health plan does not require prior authorization. Turnaround time may vary based on insurance prior authorizations or on delays caused by incomplete orders.

Health Plan Coverage

We are in-network with most national plans and many local plans.

Financial Assistance

Uninsured patients in financial need may be eligible for no fee or reduced-fee laboratory services, if qualified based on U.S. Department of Health and Human Services poverty guidelines. We also offer **payment plans**, which allow patients to pay in monthly installments until their balance is paid in full.

Prior Authorization Services

Our team is committed to helping you and your patients navigate the complexities of insurance reimbursement.

- **Insurance verification**

Our team will verify coverage and obtain prior authorization as required by your patient's health plan. This is done upon receipt of completed paperwork. The insurance verification may be done with or without receipt of a patient specimen. We will manage this process from beginning to end.

- **Estimated patient responsibility**

If your patient's out-of-pocket expense is estimated to be over \$350, we will notify you and/or your patient prior to performing the test.

For additional information, please visit [QuestCardioGenetics.com](https://www.questdiagnostics.com/cardio-genetics). For additional testing information or to speak with a genetic counselor, please call **1.866.GENE.INFO**.

Technical Specifications

Next-generation sequencing (NGS) with confirmation as needed by array CGH.

Technology and Design

- NGS method that interrogates all coding regions and splice junctions to detect sequence variations, deletions, and duplications in the genes. Certain deep intronic regions are queried for known pathogenic variants
- Random shearing of DNA is followed by bait tile capture of target sequences. This strategy creates unique clones and an enriched population of target sequences. Our methodology significantly reduces potential errors caused by Sanger sequencing and PCR-based methods, such as allele dropout, oversampling of an individual clone, and polymorphisms occurring under the PCR primer
- Alignment using proprietary software in combination with the Illumina NextSeq 500 platform enables extremely accurate allele identification
- Deletion and duplication are assessed by bioinformatic analysis of sequencing reads and confirmed as necessary by a custom targeted microarray

Analytic Sensitivity and Specificity

- >99.9% sensitivity and specificity in the validation study

Base Pair Coverage

- Our quality metric requires each base of every exon to be sequenced from at least 20 different clones. With this stringent quality metric, there has not been a false positive or false negative result in our validation series

Variant Classification

Classification Methods

- Variant classification using the American College of Medical Genetics 5-tier classification system
- Initial variant assessment performed by a team of scientists with expertise in variant analysis
- Analysis integrates data from published literature, private and public mutation databases, in silico prediction tools, and evolutionary conservation data in the context of patient phenotypes, disease mechanisms, and cellular and molecular biology
- The final interpretation is performed by board-certified directors at Quest Diagnostics Nichols Institute, and multiple reviews are performed for every positive and variant of unknown significance (VUS) case
- When a VUS is identified, the relatives of the affected family member may qualify for a family study program at no cost

Reclassification

- Ordering providers will be contacted if reclassification occurs

Genetic Counseling

Our team of genetic counselors is available from 8:30am to 8:00pm EST by calling Quest Genomics Client Services at **1.866.GENE.INFO** (1.866.436.3463) to help medical professionals with genetics-related questions, including appropriate test selection and interpretation of results. We also connect patients with external genetic counselors as requested.