



1-800-533-1710

Hypercholesterolemia Gene Panel

HCHLG

PATIENT NAME NORMAL, HCHLG				ORDER NUMBER M213000210
PATIENT ID SA00154421	DATE OF BIRTH 11/25/1985	AGE 36 Y	SEX Female	REQUESTED BY CLIENT CLIENT
COLLECTED 9/12/2022, 8:00 AM	RECEIVED 9/20/2022, 2:56 PM	REPORTED 9/28/2022, 9:12 AM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location. 7028846 MCL RochesterCampus Rochester MN 55901				CLIENT ORDER NUMBER SA00154421 CLIENT MRN SA00154421

TEST DESCRIPTION

Evaluation of 12 genes associated with familial hypercholesterolemia (FH), sitosterolemia, and other monogenic forms of inherited hypercholesterolemia, including targeted reporting of APOE E2/E2 homozygosity

SPECIMEN

DNA (ul)

RESULT SUMMARY

Negative

RESULT

No reportable variants were detected.

INTERPRETATION

This result decreases the likelihood but does not rule out the involvement of the genes evaluated in this panel.

Individuals may have a pathogenic variant in one of the interrogated genes that is not detectable by the methods utilized. Additionally, the clinical phenotype that is observed in this individual and/or family may be due to a pathogenic variant or variants in another gene not targeted by this test.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

Homozygosity for the APOE E2 allele (E2/E2) was NOT detected.

Homozygosity for the E2 allele may be considered a mild risk factor for type III hyperlipoproteinemia, however it is not considered a major contributor to disease risk. Therefore, the absence of this genotype should be interpreted in conjunction with other clinical and laboratory findings (including other genetic variants detected by this test). Other genetic, hormonal, and environmental factors also contribute to disease expression. See OMIM for additional information regarding APOE genotype. Other high frequency APOE alleles (E3, E4), which have not been firmly associated with hyperlipoproteinemia, are not reported.

METHOD

Next generation sequencing (NGS) and/or Sanger sequencing was performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed, as well as some other regions that have known

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pathogenic variants. The human genome reference GRCh37/hg19 build was used for sequence read alignment. At least 99% of the bases are covered at a read depth over 30X. Sensitivity is estimated at above 99% for single nucleotide variants, above 94% for indels less than 40 base pairs (bp), above 95% for deletions up to 75 bp and insertions up to 47 bp. NGS, and/or a polymerase chain reaction (PCR)-based quantitative method was performed to test for the presence of deletions and duplications in the genes analyzed.

Reporting of the homozygous APOE (NM_000041.3) E2 genotype is based on the presence of the homozygous c.388T (reference allele) and c.526C>T (rs7412) variants.

See the Genes Analyzed field for a list of genes tested.

There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria. See www.mayocliniclabs.com (TEST ID HCHLG) for details regarding genes with regions not routinely covered.

GENES ANALYZED

ABCG5, ABCG8, APOB, APOE, CETP, CYP27A1, LDLR, LDLRAP1, LIPA, LPL, LRP6 and PCSK9

DISCLAIMER

Clinical Correlations

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

If testing was performed because of a clinically significant family history it is often useful to first test an affected family member. Detection of a reportable variant(s) in an affected family member would allow for more informative testing of at risk individuals.

To discuss the availability of further testing options or for assistance in the interpretation of these results, Mayo Clinic Laboratory genetic counselors can be contacted at 1-800-533-1710.

Technical Limitations

Next generation sequencing may not detect all types of genomic variants. In rare cases, false negative or false positive results may occur. The depth of coverage may be variable for some target regions, but assay performance below the minimum acceptable criteria or for failed regions will be noted. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

The Hypercholesterolemia Gene Panel does NOT include assessment or interpretation of the common APOE alleles E3, or E4. Additionally, heterozygous E2 alleles are not reported.

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There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria.

Additionally, low level mosaic variants may not be detected.

This test is not designed to differentiate between somatic and germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the significance of results.

Genes may be added or removed based on updated clinical relevance. Please refer to the Targeted Genes and Methodology Details for the Hypercholesterolemia Gene Panel in the Special Instructions section of the Test Catalog for the most up to date list of genes included in this test.

Reclassification of Variants Policy

See www.mayocliniclabs.com (TEST ID HCHLG) for information regarding the laboratory's policy for reclassification of variants.

Variant Evaluation

Variant curation is performed using published ACMG-AMP recommendations as a guideline. Other gene-specific guidelines may also be considered. Variants classified as benign or likely benign are not reported.

Results from in silico evaluation tools may change over time and should be interpreted with caution and professional clinical judgment.

TEST CLASSIFICATION

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

RELEASED BY

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