MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hypercholesterolemia Gene Panel

The following applies to HCHLG / Hypercholesterolemia Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants (CNV) in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from November 2022 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ABCG5	NM_022436.3	-	-
ABCG8	NM_022437.3	-	-
APOB	NM_000384.3	-	-
APOE	NM_000041.4	-	-
CETP	NM_000078.3	-	-
CYP27A1	NM_000784.4	-	-
LDLR	NM_000527.5	chr19:11200029_11200044 (c.-196_c.-181) (Repeat 1 Sp1 binding site) chr19:11200064_11200079 (c.-161_c.-146) (Repeat 2 SREBP binding site) chr19:11200080_11200095 (c.-145_c.-130) (Repeat 3 Sp1 binding site) chr19:11231284C>G (c.2140+86C>G) chr19:11231301G>T (c.2140+103G>T)	-
LDLRAP1	NM_015627.3	-	-
LIPA	NM_000235.4	-	-
LPL	NM_000237.3	-	CNV may not be detected in exon 10.
LRP6	NM_002336.3	-	-
PCSK9	NM_174936.4	-	-