MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hypertriglyceridemia Gene Panel

The following applies to HYPTG / Hypertriglyceridemia 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 (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
APOA5	NM_052968.5	-	-
APOC2	NM_000483.5	-	-
APOE	NM_000041.4	-	-
CREB3L3	NM_032607.3	-	-
GPD1	NM_005276.4	-	CNV may not be detected in exon 8
GPIHBP1	NM_178172.6	-	-
LCAT	NM_000229.2	-	-
LIPA	NM_000235.4	-	-
LIPC	NM_000236.3	-	-
LMF1	NM_022773.4	-	-
LPL	NM_000237.3	-	CNV may not be detected in exon 10
LRP6	NM_002336.3	-	-