MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hypobetalipoproteinemia Gene Panel

The following applies to HYPBG / Hypobetalipoproteinemia 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
ANGPTL3	NM_014495.4	-	-
APOB	NM_000384.3	-	-
MTTP	NM_000253.3	chr4:g.100522736A>G (c.1237-28A>G)	-
PCSK9	NM_174936.4	-	-
SAR1B	NM_001033503.3	-	-

Effective Date	Version	Synopsis of Test Change
6/4/2025	V2	Added Additional Evaluation for *MTTP*

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