MAYO CLINIC LABORATORIES | Targeted Genes and Methodology Details for Macro/Microthrombocytopenia Gene Panel

The following applies to GNMTC / Macro/Microthrombocytopenia Gene Panel, Next-Generation Sequencing. 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 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 June 2023 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	c.63+1 to c.63+53	-
ACTN1	NM_001130004.1	-	Sequence variants and CNV in exon 10 may not be detected or reported
ARPC1B	NM_005720.4	-	-
CDC42	NM_001791.4	-	Sequence variants and CNV in exons 2, 4, and 6 may not be detected or reported
DIAPH1	NM_005219.5	-	-
FLNA	NM_001456.3	NM_001110556.2 Exon 30	-
FYB1	NM_001465.6	-	Duplication analysis for CNV in exons 5 and 8 will not be performed
GATA1	NM_002049.4	c.-19-10 to c.-1	-
GNE	NM_001128227.3	-	-
GP1BA	NM_000173.7	-	Sequence variants in exon 2 may not be detected or reported
GP1BB	NM_000407.5	c.-160C>G	-
GP9	NM_000174.4	-	-
MPIG6B	NM_025260.3	c.*1 to c.*40	-
MYH9	NM_002473.5	-	-
PRKACG	NM_002732.3	-	-
SLFN14	NM_001129820.2	-	-
TPM4	NM_001145160.2	-	Sequence variants and CNV in exons 4 and 9 may not be detected or reported
TUBB1	NM_030773.4	-	-
WAS	NM_000377.3	c.1339-30 to c.1339-1	-