MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Platelet Disorders Comprehensive Gene Panel

The following applies to GNPLT / Platelet Disorders, Comprehensive 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
ABCC4	NM_005845.4	-	-
ABCG5	NM_022436.3	-	-
ABCG8	NM_022437.3	c.63+1 to c.63+53	-
ACTB	NM_001101.5	-	Sequence variants and CNV in exons 3, 4, and 6 may not be detected or reported
ACTN1	NM_001130004.1	-	Sequence variants and CNV in exon 10 may not be detected or reported
ANKRD26	NM_014915.2	c.-200 to c.-1	Analyses for sequence variants and CNV in exon 19 will not be performed
ANO6	NM_001025356.3	-	-
AP3B1	NM_003664.4	-	-
AP3D1	NM_001261826.3	-	-
ARPC1B	NM_005720.4	-	-
BLOC1S3	NM_212550.5	-	-
BLOC1S5	NM_201280.3	-	-
BLOC1S6	NM_012388.3	-	-
CDC42	NM_001791.4	-	Sequence variants and CNV in exons 2, 4, and 6 may not be detected or reported
CYCS	NM_018947.6	-	Sequence variants and CNV in exons 2 and 3 may not be detected or reported
DIAPH1	NM_005219.5	-	-
DTNBP1	NM_032122.4	-	-
ETV6	NM_001987.5	-	-
FERMT3	NM_031471.6	-	-
FLI1	NM_002017.5	-	-
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	-
GATA2	NM_032638.5	c.1017+532T>A, c.1017+572C>T, c.1017+582G>T	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
GFI1B	NM_004188.7	c.-20 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	-
GP6	NM_001083899.2	-	Duplication analysis for CNV in exon 7 will not be performed
GP9	NM_000174.4	-	-
HOXA11	NM_005523.6	-	-
HPS1	NM_000195.5	c.988-50 to c.988-1	-
HPS3	NM_032383.5	-	-
HPS4	NM_022081.5	-	-
HPS5	NM_181507.2	-	-
HPS6	NM_024747.6	-	-
IKZF5	NM_001271840.1	-	-
ITGA2B	NM_000419.5	c.1210+105A>G, c.1211-78A>G, c.*165T>C	-
ITGB3	NM_000212.2	c.1125+1 to c.1125+40	-
KDSR	NM_002035.4	-	-
LYST	NM_000081.4	c.9107-30 to c.9107-1	Sequence variants and CNV in exon 30 may not be detected or reported
MASTL	NM_032844.5	NM_0013205757.2 Exon 10	-
MECOM	NM_004991.4	-	-
MPIG6B	NM_025260.3	c.*1 to c.*40	-
MPL	NM_005373.3	-	-
MYH9	NM_002473.5	-	-
NBEA	NM_015678.4	-	Sequence variants and CNV in exons 2, 6-13, and 16 may not be detected or reported
NBEAL2	NM_015175.3	-	Analyses for sequence variants and CNV in exon 1 will not be performed
ORAI1	NM_032790.3	-	-
P2RY1	NM_002563.5	-	-
P2RY12	NM_022788.4	-	-
PLA2G4A	NM_024420.2	-	Duplication analysis for CNV in exons 2 and 6 will not be performed
PLAU	NM_002658.5	NM_001145031.2 Exon 1	-
PRKACG	NM_002732.3	-	-
PTGS1	NM_000962.4	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
RASGRP2	NM_153819.1	-	-
RBM8A	NM_005105.4	c.67+1 to c.67+40	Sequence variants and CNV in exons 1–6 may not be detected or reported
RUNX1	NM_001754.4	NM_001122607.1 Exon 5	Analyses for sequence variants and CNV in exon 1 will not be performed
SLFN14	NM_001129820.2	-	-
SRC	NM_005417.4	-	-
STIM1	NM_003156.3	NM_001382567.1 Exon 11, NM_001277961.1 Exon 11, NM_001277962.1 Exon 12	-
STXBP2	NM_006949.4	c.326-40 to c.326-1	-
TBXA2R	NM_001060.6	-	-
TBXAS1	NM_001061.6	NM_001166253.3 Exon 6	-
THPO	NM_001289998.1	c.-52 to c.-1	-
TPM4	NM_001145160.2	-	Sequence variants and CNV in exons 4 and 9 may not be detected or reported
TUBB1	NM_030773.4	-	-
VIPAS39	NM_022067.4	-	-
VPS33B	NM_018668.4	-	-
WAS	NM_000377.3	c.1339-30 to c.1339-1	-