MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Platelet Function Defect Gene Panel

The following applies to GNPFD / Platelet Function Defect 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
ANO6	NM_001025356.3	-	-
FERMT3	NM_031471.6	-	-
FLNA	NM_001456.3	NM_001110556.2 Exon 30	
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	-	-
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	-
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
PTGS1	NM_000962.4	-	-
RASGRP2	NM_153819.1	-	-
SRC	NM_005417.4	-	-
TBXA2R	NM_001060.6	-	-
TBXAS1	NM_001061.6	NM_001166253.3 Exon 6	-