MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Bleeding Disorders, Comprehensive Gene Panel

The following applies to GNBLC / Bleeding 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
F2	NM_000506.5	c.*1 to c.*115	-
F5	NM_000130.5	c.1296+268A>G	-
F7	NM_000131.4	c.-130 to c.-1, c.681+132G>T	-
F8	NM_000132.3	+/- 30 bp, c.-270 to c.-1, c.5587-93C>T, c.5998+182A>G, c.5999-277G>A, c.*1 to c.*70	-
F9	NM_000133.4	+/- 30 bp, c.-70 to c.-1, c.*980 to c.*1370	-
F10	NM_000504.4	-	Duplication analysis for CNV in exon 3 will not be performed
F11	NM_000128.4	c.485+122T>A	-
F13A1	NM_000129.4	c.-104 to c.-19+20	-
F13B	NM_001994.2	-	-
FGA	NM_000508.5	c.-202 to c.-1, c.1891+1 to c.1891+54	-
FGB	NM_005141.4	-	-
FGG	NM_000509.5	-	-
FGG	NM_021870.3	-	-
GGCX	NM_000821.7	-	-
GP1BA	NM_000173.7	-	Sequence variants in exon 2 may not be detected or reported
KLKB1	NM_000892.5	-	-
KNG1	NM_001102416.3	NM_000893.4 Exon 11	-
LMAN1	NM_005570.4	c.822+1 to c.822+70	Duplication analysis for CNV in exon 4 will not be performed
MCFD2	NM_139279.6	NM_001171511.2 Exon 1	-
PLAT	NM_000930.5	-	-
SERPINA1	NM_000295.5	c.1145T>G only	Analyses for sequence variants (other than c.1145T>G) and CNV will not be performed
SERPINE1	NM_000602.5	-	Analyses for sequence variants and CNV in exon 1 will not be performed
SERPINF2	NM_000934.3	-	-
THBD	NM_000361.3	-	-
VKORC1	NM_024006.6	-	Duplication analysis for CNV in exon 3 will not be performed
VWF	NM_000552.4	-	Sequence variants and CNV in exons 23–34 may not be detected or reported Duplication analysis for CNV in exon 26 will not be performed.