



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