MAYO CLINIC LABORATORIES

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

The following applies to GNTHR / Thrombosis 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
ADAMTS13	NM_139025.4	-	-
F2	NM_000506.5	c.*1 to c.*115	-
F5	NM_000130.5	c.1296+268A>G	-
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	-	-
HRG	NM_000412.5	-	-
PIGA	NM_002641.3	-	-
PLAT	NM_000930.5	-	-
PLG	NM_000301.3	-	Sequence variants and CNV in exons 1–5, 16–17, and 19 may not be detected or reported
PROC	NM_000312.3	Intron 5, c.*1 to c.*80	-
PROCR	NM_006404.5	-	-
PROS1	NM_000313.4	-	Sequence variants and CNV in exons 2–15 may not be detected or reported Duplication analysis for CNV in exon 3 will not be performed
SERPINC1	NM_000488.3	c.-171C>G	-
SERPIND1	NM_000185.4	-	-
THBD	NM_000361.3	-	-