MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Red Blood Cell Membrane Disorders Gene Panel

The following applies to NMEM / Red Blood Cell Membrane Disorders 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 March 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, or 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
ABCB6	NM_005689.4	-	-
ANK1	NM_000037.4	c.-204C>G, c.-160 to c.-1, c.711+1 to c.711+20, c.1801-23 to c.1801-1, NM_001142446.1 Exons 23 and 43, c.5097-40 to c.5097-1, c.5544+1 to c.5544+96	-
EPB41	NM_001166005.1	-	-
EPB42	NM_000119.3	-	-
GYPC	NM_002101.5	-	-
KCNN4	NM_002250.3	c.1119+1 to c.1119+30	-
PIEZO1	NM_001142864.4	c.1848+31C>G	Sequence variants and CNV in exon 5 may not be detected or reported.
RHAG	NM_000324.3	-	-
SLC2A1	NM_006516.3	c.-107G>A, c.680-16 to c.680-1, c.1075-20 to c.1075-1	-
SLC4A1	NM_000342.4	c.-62G>A	-
SPTA1	NM_003126.4	+/-30 bp flanking, c.-145 to c.-1, c.4339-99C>T, c.*1 to c.*300	-
SPTB	NM_001355436.2	+/-30 bp flanking	-