MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hereditary Hemolytic Anemia Gene Panel

The following applies to NHHA / Hereditary Hemolytic Anemia 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 copy number variants (CNV). 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		
AHSP	NM_016633.4	-	-
AK1	NM_000476.2	NM_001318122.2 Exon 1	-
ALDOA	NM_184041.4	-	-
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	-
BCL11A	NM_022893.4	NM_138559.1 Exon 5	-
CDIN1 (C15orf41)	NM_001130010.3	-	-
CD59	NM_203330.2	-	-
CDAN1	NM_138477.4	-	-
EPB41	NM_001166005.1	-	-
EPB42	NM_000119.3	-	-
G6PD	NM_001042351.3	-	-
GATA1	NM_002049.4	c.-19-10 to c.-1, c.831-30 to c.831-1	-
GCLC	NM_001498.4	-	-
GPI	NM_000175.5	-	-
GSR	NM_000637.5	-	Duplication analysis for CNV in exon 2 will not be performed.
GSS	NM_000178.4	c.-9+5G>A, c.129+1663A>G	-
GYPC	NM_002101.5	-	-
HK1	NM_033496.2	c.-193A>G	-
HMOX1	NM_002133.3	-	-
KCNN4	NM_002250.3	c.1119+1 to c.1119+30	-
KIF23	NM_138555.4	NM_001367805.3 Exon 8	-
KLF1	NM_006563.5	c.-154C>T, c.-124T>C	-

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Targeted Genes and Methodology Details for Hereditary Hemolytic Anemia Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
NT5C3A	NM_001002010.4	c.-71 to c.-1, NM_001002009.2 Exon 2	Duplication analysis for CNV in exons 4–5 will not be performed. Sequence variants in exon 5 may not be detected or reported.
PFKM	NM_000289.6	c.1413-64A>G	-
PGK1	NM_000291.4	c.1214-30 to c.1214-1	Sequence variants and CNV in exon 11 may not be detected or reported.
PGLS	NM_012088.3	-	-
PIEZO1	NM_001142864.4	c.1848+31C>G	Sequence variants and CNV in exon 5 may not be detected or reported.
PKLR	NM_000298.6	+/-30 bp flanking, c.-98 to c.-1, NM_181871.3 Exon 1, c.1269+43T>C	-
RHAG	NM_000324.3	-	-
SEC23B	NM_001172745.3	c.221+31A>G	-
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	-
TMPRSS6	NM_153609.3	-	-
TPI1	NM_000365.6	-	Sequence variants and CNV in exon 7 may not be detected or reported.