MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hereditary Erythrocytosis Gene Panel

The following applies to NHEP / Hereditary Erythrocytosis 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 August 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
ACO1 (IRP1)	NM_001278352.1	-	-
ANKRD26	NM_014915.2	c.-202 to c.-1	Analyses for sequence variants and CNV in exon 19 will not be performed
BHLHE41	NM_030762.3	-	-
BPGM	NM_199186.2	-	-
CYB5A	NM_001914.4	-	-
CYB5R3	NM_000398.7	NM_001171660.1 Exon 1, c.548-18 to c.548-1	Duplication analysis for CNV in exon 1 will not be performed
EGLN1 (PHD2)	NM_022051.2	-	-
EGLN2	NM_080732.4	-	-
EGLN3	NM_022073.4	-	-
EPAS1 (HIF2A)	NM_001430.5	c.-550 to c.-383	CNV in exon 3 may not be detected or reported
EPO	NM_000799.4	c.-252C>T, c.-136G>A, c.14-28 to c.14-1	-
EPOR	NM_000121.4	-	-
GFI1B	NM_004188.7	-	-
HIF1A	NM_001530.4	NM_00243084.1 Exon 1	-
HIF1AN	NM_017902.3	-	-
HIF3A	NM_152795.4	-	-
JAK2	NM_004972.3	-	-
KDM6A	NM_021140.3	NM_001291415.1 Exon 14	Sequence variants and CNV in exons 13 and 19 may not be detected or reported
PFKM	NM_000289.6	c.1413-64A>G	-
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	-
SH2B3	NM_001291424.1	NM_005475.2 Exon 2	-
SOCS3	NM_003955.4	-	-
VHL	NM_000551.3	c.340+566 to c.340+844	-

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