



## 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 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
<i>ACO1 (IRP1)</i>	NM_001278352.1	-	-
<i>ANKRD26</i>	NM_014915.2	c.-202 to c.-1	Analyses for sequence variants and CNV in exon 19 will not be performed.
<i>BHLHE41</i>	NM_030762.3	-	-
<i>BPGM</i>	NM_199186.2	-	-
<i>CYB5A</i>	NM_001914.4	-	-
<i>CYB5R3</i>	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.
<i>EGLN1 (PHD2)</i>	NM_022051.2	-	-
<i>EGLN2</i>	NM_080732.4	-	-
<i>EGLN3</i>	NM_022073.4	-	-
<i>EPAS1 (HIF2A)</i>	NM_001430.5	c.-550 to c.-383	CNV in exon 3 may not be detected or reported.
<i>EPO</i>	NM_000799.4	c.-136G>A	-
<i>EPOR</i>	NM_000121.4	-	-
<i>GFI1B</i>	NM_004188.7	-	-
<i>HIF1A</i>	NM_001530.4	NM_00243084.1 Exon 1	-
<i>HIF1AN</i>	NM_017902.3	-	-
<i>HIF3A</i>	NM_152795.4	-	-
<i>JAK2</i>	NM_004972.3	-	-
<i>KDM6A</i>	NM_021140.3	NM_001291415.1 Exon 14	Sequence variants and CNV in exons 13 and 19 may not be detected or reported.
<i>PFKM</i>	NM_000289.6	c.1413-64A>G	-
<i>PIEZO1</i>	NM_001142864.4	c.1848+31C>G	Sequence variants and CNV in exon 5 may not be detected or reported.
<i>PKLR</i>	NM_000298.6	+/-30 bp flanking, c.-98 to c.-1, NM_181871.3 Exon 1, c.1269+43T>C	-
<i>SH2B3</i>	NM_001291424.1	NM_005475.2 Exon 2	-
<i>SOCS3</i>	NM_003955.4	-	-
<i>VHL</i>	NM_000551.3	c.340+566 to c.340+844	-