MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Primary Hemophagocytic Lymphohistiocytosis *(HLH)* Gene Panel

The following applies to HLHGP / Primary Hemophagocytic Lymphohistiocytosis Gene Panel. 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 test was updated July 2025. 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 on this version or prior versions of this test, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ADA	NM_000022.4	chr20:g.43249076C>T (c.976-34G>A); chr20:g.43248503A>T (c.1079-15T>A)	-
AP3B1	NM_003664.4	-	-
AP3D1	NM_001261826.3	-	-
BLOC1S6	NM_012388.3	-	-
CD27	NM_001242.4	-	-
CD70	NM_001252.5	-	-
CDC42	NM_001791.4	-	-
CORO1A	NM_007074.3	-	CNV analysis in exon 11 is not performed
CTPS1	NM_001905.4	-	-
IFNAR2	NM_207585.2	-	-
ITK	NM_005546.3	-	-
LYST	NM_000081.4	-	-
MAGT1	NM_032121.5	-	-
MVK	NM_000431.4	-	-
NLRC4	NM_021209.4	-	CNV analysis in exon 2 is not performed
PRF1	NM_001083116.3	-	-
RAB27A	NM_004580.5	-	-
SH2D1A	NM_002351.4	-	-
SLC7A7	NM_001126106.2	-	-
STX11	NM_003764.4	-	-
STXBP2	NM_006949.4	chr19:g.7705756_7705763del (c.326-30_326-23del); chr19:g.7705763_7705770del (c.326-23_326-16del)	-
UNC13D	NM_199242.2	chr17:g.73839908 to chr17:g.73839907 (c.118-308 to c.118-307); chr17:g.73827442C>T (c.2448-13G>A); chr17:g.73826245C>T (c.2831-13G>A); common 253kb inversion	-
XIAP	NM_001167.3	-	-

Effective Date	Version	Synopsis of Test Change
July 2025	V2	**Additional Evaluation** *UNC13D:* common 253kb inversion