

Targeted Genes and Methodology Details for Epstein Barr Virus (EBV) Susceptibility and Lymphoproliferative Disorders Gene Panel

The following applies to EBLPD / Epstein Barr Virus (EBV) Susceptibility and Lymphoproliferative Disorders Gene Panel, Varies. 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 numbers 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
<i>ATM</i>	NM_000051.3	chr11:g.108138753A>G (c.2639-384A>G); chr11:g.108139118_108139130del (c.2639-19_2639-7del); chr11: g.108141212_108141215del (c.2839-579_2839-576del); chr11:g.108164028A>G (c.4612-12A>G); chr11:g.108179837A>G (c.5763-1050A>G); chr11:g.108217993_108218019del (c.8585-13_8598del)	-
<i>CARD11</i>	NM_032415.6	-	-
<i>CARMIL2</i>	NM_001013838.3	-	-
<i>CD27</i>	NM_001242.4	-	-
<i>CD70</i>	NM_001252.5	-	-
<i>CORO1A</i>	NM_007074.3	-	CNV analysis in exon 11 is not performed
<i>CTLA4</i>	NM_005214.5	-	-
<i>CTPS1</i>	NM_001905.4	-	-
<i>DEF6</i>	NM_022047.4	-	-
<i>GATA2</i>	NM_032638.5	chr3:g.128202114 to chr3:g.128202177 (c.1017+526 to c.1017+589 corresponding to a highly conserved intronic region); chr3: g.128200787_128200806dup (c.1018-17_1020dup)	-
<i>ITK</i>	NM_005546.3	-	-
<i>LRBA</i>	NM_006726.4	-	CNV analysis in exons 20 and 39 is not performed
<i>MAGT1</i>	NM_032121.5	-	-
<i>PIK3CD</i>	NM_005026.5	-	-
<i>PRF1</i>	NM_001083116.3	-	-
<i>PRKCD</i>	NM_006254.4	-	-
<i>RASGRP1</i>	NM_005739.4	-	-
<i>SH2D1A</i>	NM_002351.4	-	-
<i>STK4</i>	NM_006282.5	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>STX11</i>	NM_003764.4	-	-
<i>STXBP2</i>	NM_006949.4	chr19:g.7705756_7705763del (c.326-30_326-23del); chr19:g.7705763_7705770del (c.326-23_326-16del)	-
<i>TET2</i>	NM_001127208.2	-	-
<i>TNFRSF9</i>	NM_001561.6	-	-
<i>UNC13D</i>	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	-
<i>XIAP</i>	NM_001167.3	-	-

Effective Date	Version	Synopsis of Test Change
July 2025	V2	Additional Evaluations <ul style="list-style-type: none"> <i>UNC13D</i>: common 253kb inversion