

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 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, 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
ATM	NM_000051.3	c.2639-384A>G; c.2639-19_2639-7del13; c.2839-579_2839-576delAAGT; c.4612-12A>G; c.5763-1050A>G; c.8585-13_8598del27	-
CARD11	NM_032415.6	-	-
CARMIL2	NM_001013838.3	-	-
CD27	NM_001242.4	-	-
CD70	NM_001252.5	-	-
CORO1A	NM_007074.3	-	CNV analysis in exon 11 is not performed
CTLA4	NM_005214.5	-	-
CTPS1	NM_001905.4	-	-
DEF6	NM_022047.4	-	-
GATA2	NM_032638.5	c.1017+526_1017+589 corresponding to a highly conserved intronic region; c.1018-17_1020	-
ITK	NM_005546.3	-	-
LRBA	NM_006726.4	-	CNV analysis in exons 20–39 is not performed
MAGT1	NM_032121.5	-	-
PIK3CD	NM_005026.5	-	-
PRF1	NM_001083116.3	-	-
PRKCD	NM_006254.4	-	-
RASGRP1	NM_005739.4	-	-
SH2D1A	NM_002351.4	-	-
STK4	NM_006282.5	-	-
STX11	NM_003764.4	-	-
STXBP2	NM_006949.4	c.326-30_326-23delGCCTGATG; c.326-23_326-16del	-
TET2	NM_001127208.2	-	-
TNFRSF9	NM_001561.6	-	-
UNC13D	NM_199242.2	c.118-308 to c.118-307; c.2448-13G>A; c.2831-13G>A	Analysis of 253kb inversion is not performed
XIAP	NM_001167.3	-	-