MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Autoimmune Lymphoproliferative Syndrome Gene Panel

The following applies to ALPSG / Autoimmune Lymphoproliferative Syndrome (ALPS) 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 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
ADA2	NM_001282225.2	-	-
CARD11	NM_032415.6	-	-
CASP10	NM_032977.4	-	-
CASP8	NM_001228.4	-	-
CTLA4	NM_005214.5	-	-
DEF6	NM_022047.4	-	-
FADD	NM_003824.3	-	-
FAS	NM_000043.6	-	-
FASLG	NM_000639.3	c.-261T>C	-
IL2RA	NM_000417.3	-	-
IL2RB	NM_000878.5	-	-
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	-	-
PIK3R1	NM_181523.3	-	-
PRKCD	NM_006254.4	-	-
RASGRP1	NM_005739.4	-	-
SH2D1A	NM_002351.4	-	-
STAT3	NM_139276.2	c.1282-89C>T	-
STK4	NM_006282.5	-	-
TET2	NM_001127208.2	-	-
TNFAIP3	NM_006290.4	-	-
TNFRSF9	NM_001561.6	-	-
TPP2	NM_003291.4	-	CNV analysis in exon 28 is not performed
XIAP	NM_001167.3	-	-