MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for B-Cell and Antibody Deficiency Gene Panel

The following applies to BCELL / B-Cell and Antibody Deficiency 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 noncoding 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 to providers 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)	-
ADA2	NM_001282225.2	-	-
AICDA	NM_020661.4	-	-
ATP6AP1	NM_001183.6	-	-
BLNK	NM_013314.3	-	-
BTK	NM_000061.2	chrX:g.100641212T>C (c.-193A>G); chrX:g.100629827T>C (c.142-205A>G); chrX:g.100629416A>C (c.240+108T>G); chrX:g.100629415G>T (c.240+109C>A); chrX:g.100613695G>T (c.895-11C>A); chrX:g.100613290_100613300del (c.1102+2_1102+12del); chrX:g.100612471_100612472insTTTTTTCTACC (c.1177+28_1177+29insCACCTTTTTTCT); chrX:g.100609705T>G (c.1567-23A>C); chrX:g.100609705T>C (c.1567-23A>G); chrX:g.100609693_100609696del (c.1567-12_1567-9del)	-
CARD11	NM_032415.6	-	-
CD19	NM_001770.6	-	-
CD27	NM_001242.4	-	-
CD40	NM_001250.6	-	-
CD40LG	NM_000074.3	-	-
CD70	NM_001252.5	-	-
CD79A	NM_001783.4	-	-
CD79B	NM_000626.4	-	-
CD81	NM_004356.3	-	CNV analysis in exon 1 is not performed
CDCA7	NM_031942.5	-	-
CR2	NM_001006658.2	-	-
CTLA4	NM_005214.5	-	-
CXCR4	NM_003467.3	-	-

©2025 Mayo Foundation for Medical Education and Research

Targeted Genes and Methodology Details for B-Cell and Antibody Deficiency Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
DCLRE1C	NM_001033855.3	-	-
DNMT3B	NM_006892.4	chr20:g.31395557G>A (c.2421-11G>A)	-
GATA2	NM_032638.5	chr3:g.128202114 to chr3:g.128202177 (c.1017+526 through c.1017+589, corresponding to a highly conserved intronic region); chr3:g.128200787_128200806dup (c.1018-17_1020dup)	-
ICOS	NM_012092.4	-	-
IGHM	ENST00000390559.2	Whole gene deletion and duplication performed	CNV analysis in individual exons 1–4 is not performed
IGLL1	NM_020070.4	-	-
IKBKG	NM_001099857.3	chrX:g.153775960G>C (c.-16G>C); chrX:g.153788599A>T (c.519-23A>T); chrX:g.153787731C>T (c.518+866C>T); 11.7 kb deletion spanning IKBKG exons 4–10	CNV analysis in exons 3–10 is not performed with the exception of the common 11.7 kb deletion spanning exons 4–10
IKZF1	NM_001291845.2; NM_006060.6	-	CNV analysis in exons 4–8 is not performed
IKZF3	NM_012481.5	-	-
IL21	NM_021803.4	-	-
IL21R	NM_021798.4	-	-
IRF2BP2	NM_182972.2	-	-
KDM6A	NM_021140.3	-	-
KMT2A	NM_001197104.1	-	-
KMT2D	NM_003482.3	-	-
LIG1	NM_000234.3	-	-
LRBA	NM_006726.4	-	CNV analysis in exons 20 and 39 is not performed
MOGS	NM_006302.3	-	-
MS4A1	NM_152866.2	-	-
NFKB1	NM_003998.4	-	-
NFKB2	NM_001077494.3	-	-
PIK3CD	NM_005026.5	-	-
PIK3R1	NM_181523.3	-	-
PLCG2	NM_002661.5	-	-
PRKCD	NM_006254.4	-	-
RAC2	NM_002872.5	-	-
RAG1	NM_000448.2	-	-
RAG2	NM_000536.4	-	-
RNF168	NM_152617.4	-	-
SEC61A1	NM_013336.4	-	-
SH2D1A	NM_002351.4	-	-
SH3KBP1	NM_031892.3	-	-

Targeted Genes and Methodology Details for B-Cell and Antibody Deficiency Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
SLC39A7	NM_006979.3	-	-
TCF3	NM_001136139.4; NM_003200.5	-	-
TNFRSF13B	NM_012452.3	-	-
TNFRSF13C	NM_052945.3	-	-
TNFSF12	NM_003809.3	-	-
TNFSF13	NM_003808.3	-	-
TOP2B	NM_001068.3	-	-
TRNT1	NM_182916.3	-	-
UNG	NM_080911.3	-	-
XIAP	NM_001167.3	-	-

Effective Date	Version	Synopsis of Test Change
July 2025	V2	**Additional Evaluations** • *IGHM*: Whole gene deletion and duplication performed • *IKBKG*: 11.7 kb deletion spanning IKBKG exons 4–10