MAYO CLINIC LABORATORIES | Targeted Genes and Methodology Details for Inborn Errors of Immunity with Immune Dysregulation and Autoimmunity Gene Panel

The following applies to IMMAU / Inborn Errors of Immunity with Immune Dysregulation and Autoimmunity 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 number variants 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 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, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
AIRE	NM_000383.4	-	-
BACH2	NM_021813.4	-	-
CARD11	NM_032415.6	-	-
CASP10	NM_032977.4	-	-
CASP8	NM_001228.4	-	-
CD3G	NM_000073.2	-	-
CTLA4	NM_005214.5	-	-
DEF6	NM_022047.4	-	-
FADD	NM_003824.3	-	-
FASLG	NM_000639.3	c.-261T>C	-
FERMT1	NM_017671.4	c.-20A>G; c.1139+740G>A	CNV analysis in exon 9 is not performed
FOXP3	NM_014009.4	c.-105_-23+1303del1386 (also known as g.del-6247_-4859); c.-7G>T; c.*876 to c.*878	-
IL10	NM_000572.3	-	-
IL10RA	NM_001558.3	-	-
IL10RB	NM_000628.5	-	-
IL2RA	NM_000417.3	-	-
IL2RB	NM_000878.5	-	-
ITCH	NM_031483.7	-	-
JAK1	NM_002227.4	-	-
LRBA	NM_006726.4	-	CNV analysis in exons 20 and 39 is not performed
ORAI1	NM_032790.3	-	-
PEPD	NM_000285.4	-	-
PRKCD	NM_006254.4	-	-
RIPK1	NM_003804.6	c.688_688+20del	-
STAT3	NM_139276.2	c.1282-89C>T	-
STAT5B	NM_012448.4	-	CNV analysis in exons 6−8 is not performed
STIM1	NM_003156.3	-	-
TET2	NM_001127208.2	-	-
TGFB1	NM_000660.7	-	-
TPP2	NM_003291.4	-	CNV analysis in exon 28 is not performed