MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Severe Combined Immunodeficiency (SCID) Gene Panel

The following applies to SCIDP / Severe Combined Immunodeficiency (SCID) 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 (CNV). 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 July 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, or 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
ADA	NM_000022.4	chr20:g.43249076C>T (c.976-34G>A); chr20:g.43248503A>T (c.1079-15T>A)	-
AK2	NM_001625.4	-	-
ATM	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)	-
BCL11B	NM_138576.4	-	-
CARD11	NM_032415.6	-	-
CD247	NM_198053.2	-	-
CD3D	NM_000732.4	-	-
CD3E	NM_000733.4	-	CNV analysis in exon 4 is not performed
CD3G	NM_000073.2	-	-
CD8A	NM_001768.6	-	-
CHD7	NM_017780.4	chr8:g.61763035 to chr8:g.61763039 (c.5405-17 to c.5405-13)	-
CIITA	NM_000246.3	-	-
CORO1A	NM_007074.3	-	CNV analysis in exon 11 is not performed
DCLRE1C	NM_001033855.3	-	-
DOCK2	NM_004946.3	-	-
DOCK8	NM_203447.3	chr9:g.317025 to chr9:g.316892 (c.742-18 to c.742-151); chr9:g.372260_372348del (c.2083_2109+62del)	-
EXTL3	NM_001440.4	-	-
FOXN1	NM_003593.2	-	-
IKZF1	NM_001291845.2; NM_006060.6	-	CNV analysis in exons 4–8 is not performed
IL2RA	NM_000417.3	-	-
IL2RG	NM_000206.2	chrX:g.70331494G>A (c.-105C>T); chrX:g.70330553T>C (c.270-15A>G); chrX:g.70329246_70329257del (c.595-15_595-4del)	-

Targeted Genes and Methodology Details for Severe Combined Immunodeficiency (SCID) Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
IL7R	NM_002185.5	chr5:g.35867853G>A (c.379+288G>A)	-
JAK3	NM_000215.3	chr19:g.17946035C>T (c.1915-11G>A); chr19:g.17943239C>T (c.2680+89G>A)	-
LAT	NM_001014987.2	-	-
LCP2	NM_005565.5	-	CNV analysis in exon 8 is not performed
LIG4	NM_002312.3	-	-
MTHFD1	NM_005956.4	-	-
NBN	NM_002485.4	-	CNV analysis in exon 16 is not performed
NHEJ1	NM_024782.2	-	-
ORAI1	NM_032790.3	-	-
PAX1	NM_006192.5	-	-
PNP	NM_000270.3	chr14:g.20942914G>A (c.286-18G>A)	-
POLE2	NM_002692.4	-	CNV analysis in exons 8 and 19 is not performed
PRKDC	NM_006904.7	-	-
PTPRC	NM_002838.5	-	-
RAC2	NM_002872.5	-	-
RAG1	NM_000448.2	-	-
RAG2	NM_000536.4	-	-
RFX5	NM_000449.3	-	-
RFXANK	NM_003721.4	chr19:g.19308305_19308330del (c.338-25_338del); chr19:g.19308411_19308468del (c.419_438+38del)	-
RFXAP	NM_000538.3	-	-
RMRP (NME1)	NR_003051.3	5' UTR variants from n.-25 through the transcription initiation site	CNV analysis is not performed
SEMA3E	NM_012431.3	-	-
SMARCAL1	NM_014140.4	-	-
STIM1	NM_003156.3	-	-
TBX1	NM_080647.1	-	Sequence variants in exon 3 may not be reported
TTC7A	NM_020458.4	-	-
WAS	NM_000377.3	chrX:g.48547690_48547698delinsATCTGCAGACC (c.1339-19_1339-11delinsATCTGCAGACC)	-
WIPF1	NM_001077269.1	-	-
ZAP70	NM_001079.3	chr2:g.98349927G>A (c.838-80G>A); chr2:g.98349927G>A (c.1624-11G>A)	-