

WES SEVERE COMBINED IMMUNODEFICIENCY (SCID) DG

3.2

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ADA	96.7	99.7	96.1	102700
AK2	96.5	98.7	95.2	267500
B2M	207.3	100.0	100.0	241600;105200
CD247	98.5	100.0	99.4	610163
CD3D	160.1	100.0	99.9	615617
CD3E	130.5	100.0	98.8	615615
CD3G	145.6	100.0	100.0	615607
CD8A	126.0	100.0	100.0	608957
CIITA	165.0	100.0	99.4	209920
CORO1A	148.1	99.9	98.9	615401
DCLRE1C	137.2	99.8	98.2	602450;603554
DOCK2	121.5	99.8	98.8	616433
DOCK8	118.1	100.0	99.0	243700
FCHO1	107.1	99.3	97.5	619164
FOXP1	147.7	100.0	99.1	601705
IL2RG	58.3	99.8	92.8	312863;300400
IL7R	121.9	99.9	99.3	608971
JAK3	125.2	99.5	97.6	600802
LAT	116.4	100.0	99.4	617514
LCK	152.4	98.2	96.1	615758
LIG4	183.8	99.8	99.3	606593
NHEJ1	56.5	99.8	97.2	611291

PAX1	178.2	92.6	87.5	615545
PNP	109.8	99.8	98.7	613179
PRKDC	102.1	99.2	96.9	615966
PTPRC	104.3	98.8	93.9	608971
RAC2	97.5	99.8	95.4	608203
RAG1	145.7	100.0	100.0	603554;609889;601457;233650
RAG2	185.4	100.0	100.0	603554;601457;233650
RFX5	114.4	99.7	98.1	209920
RFXANK	131.8	100.0	99.7	209920
RFXAP	100.8	99.9	98.6	209920
RMRP				250250
SCIMP	95.2	80.4	79.9	-
STK4	118.7	99.9	99.7	614868
TAP1	107.8	100.0	97.6	604571
TAP2	94.2	99.9	98.6	604571
TAPBP	121.3	96.5	95.1	604571
TTC7A	111.6	99.6	97.1	243150
ZAP70	174.7	100.0	99.7	617006;269840

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.

Median Coverage describes the average number of reads seen across 50 exomes.

Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with no value for coverage are non protein coding genes.

Non protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions: September 1st, 2021.