

WES SEVERE COMBINED IMMUNODEFICIENCY (SCID) DG

3.4

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ADA	165.7	100.0	100.0	102700
AK2	156.1	100.0	100.0	267500
B2M	253.5	100.0	100.0	241600;105200
CD247	163.4	100.0	100.0	610163
CD3D	158.8	100.0	100.0	615617
CD3E	161.9	100.0	100.0	615615
CD3G	155.7	100.0	100.0	615607
CD8A	204.3	100.0	100.0	608957
CIITA	217.7	100.0	100.0	209920
CORO1A	201.0	100.0	100.0	615401
DCLRE1C	167.9	100.0	100.0	602450;603554
DOCK2	153.8	100.0	100.0	616433
DOCK8	161.5	100.0	100.0	243700
FCHO1	189.1	100.0	100.0	619164
FOXP1	218.5	100.0	100.0	601705
IL2RG	165.1	100.0	100.0	312863;300400
IL7R	205.1	100.0	100.0	608971
ITPKB	218.3	100.0	100.0	No OMIM phenotype
JAK3	226.6	100.0	100.0	600802
LAT	203.8	100.0	100.0	617514
LCK	200.3	100.0	100.0	615758
LCP2	157.3	100.0	100.0	619374

LIG4	173.4	100.0	100.0	606593
NHEJ1	167.2	100.0	100.0	611291
PAX1	238.0	100.0	100.0	615545
PNP	177.3	100.0	100.0	613179
PRKDC	167.4	100.0	100.0	615966
PTPRC	163.6	100.0	100.0	608971
RAC2	166.9	100.0	100.0	608203
RAG1	212.7	100.0	100.0	603554;609889;601457;233650
RAG2	207.1	100.0	100.0	603554;601457;233650
RFX5	176.7	100.0	100.0	209920
RFXANK	170.1	100.0	100.0	209920
RFXAP	212.7	100.0	100.0	209920
RMRP				250250
STK4	155.1	100.0	100.0	614868
TAP1	304.4	100.0	100.0	604571
TAP2	272.9	100.0	100.0	604571
TAPBP	315.0	96.6	96.6	604571
TTC7A	175.0	100.0	100.0	243150
ZAP70	227.5	100.0	100.0	617006;269840

Gene symbols used follow HGNC 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.