

WES SEVERE COMBINED IMMUNODEFICIENCY (SCID) DG

3.5

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ADA	125.3	100.0	100.0	102700
AK2	133.8	100.0	100.0	267500
B2M	165.0	100.0	100.0	241600;105200
CD247	130.8	100.0	100.0	610163
CD3D	123.8	100.0	100.0	615617
CD3E	135.9	100.0	100.0	615615
CD3G	144.2	100.0	100.0	615607
CD8A	129.7	100.0	100.0	608957
CIITA	121.7	100.0	100.0	209920
CORO1A	130.9	100.0	100.0	615401
DCLRE1C	154.6	100.0	100.0	602450;603554
DOCK2	123.8	99.9	99.5	616433
DOCK8	126.5	100.0	100.0	243700
FCHO1	117.7	100.0	100.0	619164
FOXI3	117.0	99.8	99.0	-
FOXN1	142.3	100.0	100.0	601705
IL2RG	99.6	100.0	100.0	312863;300400
IL7R	148.6	100.0	100.0	608971
ITPKB	115.2	100.0	100.0	No OMIM phenotype
JAK3	125.3	100.0	100.0	600802
LAT	142.8	100.0	100.0	617514
LCK	139.1	100.0	100.0	615758

LCP2	135.3	100.0	100.0	619374
LIG4	156.5	100.0	100.0	606593
NHEJ1	132.1	100.0	100.0	611291
PAX1	156.3	100.0	100.0	615545
PNP	144.5	100.0	100.0	613179
PRKDC	135.0	100.0	100.0	615966
PTPRC	144.6	100.0	99.8	608971
RAC2	131.4	100.0	100.0	608203
RAG1	140.9	100.0	100.0	603554;609889;601457;233650
RAG2	151.3	100.0	100.0	603554;601457;233650
RFX5	119.0	100.0	100.0	209920
RFXANK	123.9	100.0	100.0	209920
RFXAP	132.2	100.0	100.0	209920
RMRP				250250
STK4	138.6	100.0	100.0	614868
TAP1	137.0	100.0	100.0	604571
TAP2	130.3	100.0	100.0	604571
TAPBP	116.9	95.9	95.9	604571
TTC7A	116.2	100.0	100.0	243150
ZAP70	139.3	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.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors