SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL DG-5.2.0 (46 GENES)

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.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
ADA	83% | 83%	96% | 96%	99% | 99%	Adenosine deaminase deficiency, partial, 102700;Severe combined immunodeficiency due to ADA deficiency, 102700
AK2	90% | 89%	94% | 94%	99% | 99%	Reticular dysgenesis, 267500
B2M	100% | 100%	97% | 97%	100% | 100%	Amyloidosis, hereditary systemic 6, 620659;Immunodeficiency 43, 241600
CD247	67% | 66%	97% | 97%	99% | 99%	?Immunodeficiency 25, 610163
CD3D	100% | 100%	96% | 96%	99% | 99%	Immunodeficiency 19, severe combined, 615617
CD3E	100% | 100%	98% | 98%	99% | 99%	Immunodeficiency 18, 615615;Immunodeficiency 18, SCID variant, 615615
CD3G	100% | 100%	97% | 97%	98% | 98%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD8A	100% | 100%	91% | 91%	99% | 99%	Immunodeficiency 116, 608957
CIITA	99% | 99%	96% | 96%	99% | 99%	{Rheumatoid arthritis, susceptibility to}, 180300;MHC class II deficiency 1, 209920
CORO1A	92% | 92%	94% | 92%	98% | 95%	Immunodeficiency 8, 615401
DCLRE1C	97% | 97%	97% | 97%	99% | 99%	Severe combined immunodeficiency, Athabascan type, 602450;Omenn syndrome, 603554
DOCK2	100% | 100%	97% | 97%	99% | 99%	Immunodeficiency 40, 616433
DOCK8	99% | 99%	97% | 97%	99% | 99%	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700
FCHO1	97% | 96%	96% | 96%	98% | 98%	Immunodeficiency 76, 619164
FOXI3	98% | 98%	93% | 93%	98% | 98%	Craniofacial microsomia 2, 620444
FOXN1	100% | 100%	96% | 96%	99% | 99%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806;T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705;?T-cell immunodeficiency with thymic aplasia, 242700

IL2RG	99% | 99%	98% | 98%	99% | 99%	Combined immunodeficiency, X-linked, moderate, 312863;Severe combined immunodeficiency, X-linked, 300400
IL7R	100% | 100%	97% | 97%	99% | 99%	Immunodeficiency 104, severe combined, 608971
ITPKB	99% | 99%	96% | 96%	98% | 98%	
JAK3	100% | 100%	95% | 95%	99% | 99%	Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type, 600802
LAT	100% | 100%	96% | 96%	99% | 99%	Immunodeficiency 52, 617514
LCK	99% | 99%	97% | 97%	97% | 97%	Immunodeficiency 22, 615758
LCP2	100% | 100%	97% | 97%	99% | 99%	Immunodeficiency 81, 619374
LIG4	100% | 100%	98% | 98%	99% | 99%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
NHEJ1	93% | 93%	97% | 97%	100% | 100%	Microphthalmia/coloboma 13, 620968;Immunodeficiency 124, severe combined, 611291
NUDCD3	91% | 91%	97% | 97%	99% | 99%	
PAX1	100% | 100%	96% | 96%	99% | 99%	Otofaciocervical syndrome 2 with T-cell deficiency, 615560
PNP	100% | 100%	97% | 97%	99% | 99%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POLD3	100% | 100%	98% | 98%	99% | 99%	Immunodeficiency 122, 620869
PRKDC	100% | 100%	98% | 98%	99% | 99%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PSMB10	100% | 100%	97% | 97%	98% | 98%	Immunodeficiency 121 with autoinflammation, 620807;Proteasome-associated autoinflammatory syndrome 5, 619175
PTCRA	100% | 100%	97% | 97%	98% | 98%	Immunodeficiency 126, 620931
PTPRC	100% | 100%	98% | 98%	100% | 100%	Immunodeficiency 105, severe combined, 619924
RAC2	100% | 100%	96% | 96%	98% | 98%	Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203;?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987;Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986
RAG1	100% | 100%	97% | 97%	100% | 100%	Omenn syndrome, 603554;Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889
RAG2	100% | 100%	98% | 98%	100% | 100%	Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Omenn syndrome, 603554
RFX5	100% | 100%	97% | 97%	98% | 98%	?MHC class II deficiency 5, 620818;MHC class II deficiency 3, 620816
RFXANK	100% | 100%	96% | 96%	98% | 98%	MHC class II deficiency 2, 620815

RFXAP	100% | 100%	95% | 95%	99% | 99%	MHC class II deficiency 4, 620817
RMRP				Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
STK4	100% | 100%	98% | 98%	99% | 99%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
TAP1	97% | 96%	97% | 97%	99% | 99%	MHC class I deficiency 1, 604571
TAP2	98% | 98%	96% | 96%	99% | 99%	MHC class I deficiency 2, 620813
TAPBP	89% | 89%	95% | 95%	98% | 98%	?MHC class I deficiency 3, 620814
TTC7A	99% | 99%	97% | 97%	99% | 99%	Gastrointestinal defects and immunodeficiency syndrome, 243150
ZAP70	100% | 100%	97% | 97%	99% | 99%	Immunodeficiency 48, 269840;Autoimmune disease, multisystem, infantile-onset, 2, 617006