SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL DG 3.8.1 (42 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
ADA	100.0%	100.0%	100.0%	99.8%	Adenosine deaminase deficiency, partial, 102700;Severe combined immunodeficiency due to ADA deficiency, 102700
AK2	100.0%	100.0%	100.0%	99.3%	Reticular dysgenesis, 267500
B2M	100.0%	100.0%	100.0%	99.5%	?Amyloidosis, familial visceral, 105200;Immunodeficiency 43, 241600
CD247	100.0%	100.0%	100.0%	99.9%	?Immunodeficiency 25, 610163
CD3D	100.0%	100.0%	100.0%	99.9%	Immunodeficiency 19, severe combined, 615617
CD3E	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 18, 615615;Immunodeficiency 18, SCID variant, 615615
CD3G	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 17, CD3 gamma deficient, 615607

CD8A	100.0%	100.0%	100.0%	99.9%	Immunodeficiency 116, 608957
CIITA	100.0%	100.0%	100.0%	99.5%	{Rheumatoid arthritis, susceptibility to}, 180300;Bare lymphocyte syndrome, type II, complementation group A, 209920
CORO1A	100.0%	100.0%	100.0%	98.7%	Immunodeficiency 8, 615401
DCLRE1C	100.0%	100.0%	100.0%	99.0%	Severe combined immunodeficiency, Athabascan type, 602450;Omenn syndrome, 603554
DOCK2	99.9%	99.5%	100.0%	99.6%	Immunodeficiency 40, 616433
DOCK8	100.0%	100.0%	100.0%	99.4%	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700
FCHO1	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 76, 619164
FOXI3	99.8%	99.0%	100.0%	98.7%	Craniofacial microsomia 2, 620444
FOXN1	100.0%	100.0%	100.0%	99.7%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806;T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705

IL2RG	100.0%	100.0%	98.6%	73.9%	Combined immunodeficiency, X-linked, moderate, 312863;Severe combined immunodeficiency, X-linked, 300400
IL7R	100.0%	100.0%	100.0%	99.7%	Immunodeficiency 104, severe combined, 608971
ITPKB	100.0%	100.0%	100.0%	99.9%	
JAK3	100.0%	100.0%	100.0%	99.6%	SCID, autosomal recessive, T-negative/B-positive type, 600802
LAT	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 52, 617514
LCK	100.0%	100.0%	100.0%	99.8%	?Immunodeficiency 22, 615758
LCP2	100.0%	100.0%	100.0%	99.4%	?Immunodeficiency 81, 619374
LIG4	100.0%	100.0%	100.0%	99.4%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
NHEJ1	100.0%	100.0%	100.0%	99.7%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
PAX1	100.0%	100.0%	100.0%	99.2%	Otofaciocervical syndrome 2, 615560

PNP	100.0%	100.0%	100.0%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PRKDC	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PTPRC	100.0%	99.8%	100.0%	99.3%	Immunodeficiency 105, severe combined, 619924
RAC2	100.0%	100.0%	100.0%	99.7%	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	100.0%	100.0%	100.0%	99.6%	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	100.0%	100.0%	100.0%	99.3%	Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Omenn syndrome, 603554
RFX5	100.0%	100.0%	100.0%	99.4%	Bare lymphocyte syndrome, type II, complementation group C, 209920;Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	100.0%	100.0%	100.0%	100.0%	Bare lymphocyte syndrome, type II, complementation group B, 209920
RFXAP	100.0%	100.0%	100.0%	99.4%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RMRP					Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
STK4	100.0%	100.0%	100.0%	99.4%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
TAP1	100.0%	100.0%	100.0%	99.4%	Bare lymphocyte syndrome, type I, 604571

TAP2	100.0%	100.0%	100.0%	98.9%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	95.9%	95.9%	100.0%	99.3%	Bare lymphocyte syndrome, type I, 604571
TTC7A	100.0%	100.0%	100.0%	99.7%	Gastrointestinal defects and immunodeficiency syndrome, 243150
ZAP70	100.0%	100.0%	100.0%	99.9%	Immunodeficiency 48, 269840;Autoimmune disease, multisystem, infantile-onset, 2, 617006

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 X2 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry.
TWIST X2 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry.
srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS GRCh38 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : March 17th, 2023.
This list is accurate for panel version DG 3.8.1

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