SEVERE COMBINED IMMUNODEFICIENCY (SCID) GENE PANEL DG 3.6.0 (42 GENES)

Releasedate: 05-04-2023

Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*Associated Phenotype description and OMIM disease ID*
ADA	100%	100%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
AK2	100%	100%	Reticular dysgenesis, 267500
B2M	100%	100%	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
CD247	100%	100%	?Immunodeficiency 25, 610163
CD3D	100%	100%	Immunodeficiency 19, severe combined, 615617
CD3E	100%	100%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD8A	100%	100%	CD8 deficiency, familial, 608957
CIITA	100%	100%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CORO1A	100%	100%	Immunodeficiency 8, 615401
DCLRE1C	100%	100%	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DOCK2	99%	99%	Immunodeficiency 40, 616433
DOCK8	100%	100%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
FCHO1	100%	100%	Immunodeficiency 76, 619164
FOXI3	99%	99%	No OMIM disease ID
FOXN1	100%	100%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
IL2RG	100%	100%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL7R	100%	100%	Immunodeficiency 104, severe combined, 608971
ITPKB	100%	100%	No OMIM disease ID
JAK3	100%	100%	SCID, autosomal recessive, T-negative/B-positive type, 600802

LAT	100%	100%	Immunodeficiency 52, 617514
LCK	100%	100%	?Immunodeficiency 22, 615758
LCP2	100%	100%	?Immunodeficiency 81, 619374
LIG4	100%	100%	LIG4 syndrome, 606593
NHEJ1	100%	100%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
PAX1	100%	100%	Otofaciocervical syndrome 2, 615560
PNP	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PRKDC	100%	100%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PTPRC	100%	99%	Immunodeficiency 105, severe combined, 619924
RAC2	100%	100%	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%	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%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RFX5	100%	100%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	100%	100%	Bare lymphocyte syndrome, type II, complementation group B, 209920
RFXAP	100%	100%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RMRP	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
STK4	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
TAP1	100%	100%	Bare lymphocyte syndrome, type I, 604571
TAP2	100%	100%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	95%	95%	Bare lymphocyte syndrome, type I, 604571
TTC7A	100%	100%	Gastrointestinal defects and immunodeficiency syndrome, 243150
ZAP70	100%	100%	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.
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TWIST X2 is the chemistry used for WES analysis.

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 coverage denoting NC 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 : March 17th, 2023
This list is accurate for panel version DG 3.6.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors