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SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL DG-5.1.0 (46 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ADA	85.1%	84.4%	100%	99.9%	99.5%	Adenosine deaminase deficiency, partial, 102700;Severe combined immunodeficiency due to ADA deficiency, 102700
AK2	98.1%	93.2%	100%	100%	99.1%	Reticular dysgenesis, 267500
B2M	100%	100%	100%	100%	99.8%	Amyloidosis, hereditary systemic 6, 620659;Immunodeficiency 43, 241600
CD247	77.2%	71.4%	100%	100%	99.5%	?Immunodeficiency 25, 610163
CD3D	100%	100%	100%	100%	99.7%	Immunodeficiency 19, severe combined, 615617
CD3E	100%	100%	100%	100%	99.8%	Immunodeficiency 18, 615615;Immunodeficiency 18, SCID variant, 615615
CD3G	100%	100%	100%	100%	99.7%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD8A	100%	100%	100%	100%	99.5%	Immunodeficiency 116, 608957
CIITA	100%	100%	100%	100%	99.2%	{Rheumatoid arthritis, susceptibility to}, 180300;MHC class II deficiency 1, 209920
CORO1A	100%	100%	100%	100%	99.7%	Immunodeficiency 8, 615401

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DCLRE1C	97.1%	97.1%	100%	100%	99.9%	Severe combined immunodeficiency, Athabascan type, 602450;Omenn syndrome, 603554
DOCK2	100%	100%	100%	100%	99.7%	Immunodeficiency 40, 616433
DOCK8	98.6%	98.6%	100%	100%	99.7%	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700
FCHO1	98.6%	96.4%	100%	99.9%	99.2%	Immunodeficiency 76, 619164
FOXI3	100%	99.9%	100%	99.5%	97.6%	Craniofacial microsomia 2, 620444
FOXN1	100%	100%	100%	100%	99.8%	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	100%	100%	98.8%	90.9%	72.5%	Combined immunodeficiency, X-linked, moderate, 312863;Severe combined immunodeficiency, X-linked, 300400
IL7R	100%	100%	100%	100%	99.7%	Immunodeficiency 104, severe combined, 608971
ITPKB	100%	100%	100%	100%	99.5%	
JAK3	100%	100%	100%	99.9%	98.9%	Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type, 600802
LAT	100%	100%	100%	100%	99.7%	Immunodeficiency 52, 617514

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LCK	98%	98%	100%	100%	99.7%	Immunodeficiency 22, 615758
LCP2	100%	100%	100%	100%	99.6%	Immunodeficiency 81, 619374
LIG4	100%	100%	100%	100%	99.8%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
NHEJ1	92.9%	92.9%	100%	100%	99.7%	Microphthalmia/coloboma 13, 620968;Immunodeficiency 124, severe combined, 611291
NUDCD3	91%	91%	100%	99.9%	99.2%	
PAX1	100%	100%	100%	100%	98.6%	Otofaciocervical syndrome 2 with T-cell deficiency, 615560
PNP	100%	100%	100%	100%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POLD3	100%	100%	100%	100%	99.5%	Immunodeficiency 122, 620869
PRKDC	100%	100%	100%	100%	99.7%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PSMB10	100%	100%	100%	100%	99%	Immunodeficiency 121 with autoinflammation, 620807;Proteasome-associated autoinflammatory syndrome 5, 619175
PTCRA	100%	100%	100%	100%	99.8%	Immunodeficiency 126, 620931
PTPRC	100%	100%	100%	100%	99.8%	Immunodeficiency 105, severe combined, 619924

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RAC2	100%	100%	100%	99.9%	99.4%	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%	100%	100%	99.7%	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%	100%	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Omenn syndrome, 603554
RFX5	100%	100%	100%	100%	99.8%	?MHC class II deficiency 5, 620818;MHC class II deficiency 3, 620816
RFXANK	100%	100%	100%	100%	99.4%	MHC class II deficiency 2, 620815
RFXAP	100%	100%	100%	99.8%	98.8%	MHC class II deficiency 4, 620817

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RMRP						Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
STK4	100%	100%	100%	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
TAP1	99.7%	97.9%	100%	100%	99.7%	MHC class I deficiency 1, 604571
TAP2	97.9%	97.9%	100%	99.9%	99%	MHC class I deficiency 2, 620813
TAPBP	88.8%	88.8%	100%	99.8%	98%	?MHC class I deficiency 3, 620814
TTC7A	100%	99.8%	100%	100%	99.6%	Gastrointestinal defects and immunodeficiency syndrome, 243150
ZAP70	100%	100%	100%	100%	99.4%	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 mapped against GRCh38.
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 mapped against GRCh38.
srWGS 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 covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS 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 : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors