SCID GENE PANEL DG 2.17 (37 genes)

Releasedate: 06-12-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ADA	111.3	100.0%	99.6%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
AK2	100.4	98.7%	94.5%	Reticular dysgenesis, 267500
B2M	194.6	100.0%	100.0%	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
CD247	99.1	99.9%	99.2%	?Immunodeficiency 25, 610163
CD3D	146.3	100.0%	99.9%	Immunodeficiency 19, 615617
CD3E	126.6	100.0%	99.6%	Immunodeficiency 18, SCID variant, 615615 Immunodeficiency 18, 615615
CD3G	137.2	100.0%	100.0%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD8A	166.1	100.0%	100.0%	CD8 deficiency, familial, 608957
CIITA	166.3	100.0%	100.0%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CORO1A	166.6	100.0%	99.2%	Immunodeficiency 8, 615401
DCLRE1C	139.2	99.9%	98.0%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DOCK2	123.0	100.0%	99.5%	Immunodeficiency 40, 616433
DOCK8	115.2	100.0%	99.7%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
FOXN1	149.7	100.0%	99.8%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
IL2RG	60.2	99.7%	94.3%	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL7R	114.7	100.0%	99.7%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
JAK3	134.3	98.7%	97.3%	SCID, autosomal recessive, T-negative/B-positive type, 600802
LAT	127.2	100.0%	99.8%	Immunodeficiency 52, 617514
LCK	163.1	99.7%	98.3%	?Immunodeficiency 22, 615758
LIG4	170.5	100.0%	99.9%	LIG4 syndrome, 606593
NHEJ1	60.2	99.4%	94.3%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
PNP	113.1	100.0%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PRKDC	98.5	99.4%	96.4%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PTPRC	98.4	98.3%	93.6%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971

RAC2	109.4	100.0%	99.4%	Neutrophil immunodeficiency syndrome, 608203
RAG1	158.7	100.0%	100.0%	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650
RAG2	188.7	100.0%	100.0%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RFX5	117.8	99.9%	98.2%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	133.2	100.0%	99.9%	MHC class II deficiency, complementation group B, 209920
RFXAP	127.3	100.0%	100.0%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
STK4	124.5	100.0%	99.7%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
TAP1	133.9	99.9%	97.7%	Bare lymphocyte syndrome, type I, 604571
TAP2	101.4	99.6%	98.7%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	130.6	96.6%	96.6%	Bare lymphocyte syndrome, type I, 604571
TTC7A	123.2	99.9%	98.9%	Gastrointestinal defects and immunodeficiency syndrome, 243150
ZAP70	206.3	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.
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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors