SCID GENE PANEL DG 2.18 (37 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ADA	100%	99,70%	100%	100%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
AK2	98,80%	94,50%	100%	100%	Reticular dysgenesis, 267500
B2M	100%	100%	100%	100%	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
CD247	100%	100%	100%	100%	?Immunodeficiency 25, 610163
CD3D	100%	100%	100%	100%	Immunodeficiency 19, 615617
CD3E	100%	99,50%	100%	100%	Immunodeficiency 18, SCID variant, 615615 Immunodeficiency 18, 615615
CD3G	100%	100%	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD8A	100%	99,80%	100%	100%	CD8 deficiency, familial, 608957
CIITA	100%	99,50%	100%	100%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CORO1A	100%	98,60%	100%	100%	Immunodeficiency 8, 615401
DCLRE1C	100%	99,40%	100%	100%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DOCK2	100%	99,60%	100%	100%	Immunodeficiency 40, 616433
DOCK8	100%	99,60%	100%	100%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
FOXN1	100%	99,60%	100%	100%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
IL2RG	99,80%	97,10%	100%	100%	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL7R	100%	99,80%	100%	100%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
JAK3	99,90%	98,70%	100%	100%	SCID, autosomal recessive, T-negative/B-positive type, 600802
LAT	100%	99,20%	100%	100%	Immunodeficiency 52, 617514
LCK	98,90%	96,60%	100%	100%	?Immunodeficiency 22, 615758
LIG4	100%	99,90%	100%	100%	LIG4 syndrome, 606593
NHEJ1	100%	96,20%	100%	100%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
TAP1	100%	99,20%	100%	100%	Bare lymphocyte syndrome, type I, 604571

TAP2	99,90%	99,30%	100%	100%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,50%	95,50%	96,60%	96,60%	Bare lymphocyte syndrome, type I, 604571
PNP	99,80%	98,90%	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PRKDC	99,70%	98,00%	100%	100%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PTPRC	99,00%	95,10%	100%	100%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
RAC2	99,90%	98,30%	100%	100%	Neutrophil immunodeficiency syndrome, 608203
RAG1	100%	100%	100%	100%	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	100%	100%	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RFX5	99,70%	98,10%	100%	100%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	100%	99,50%	100%	100%	MHC class II deficiency, complementation group B, 209920
RFXAP	99,30%	97,00%	100%	99,90%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
STK4	100%	99,80%	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
TTC7A	99,30%	95,40%	100%	100%	Gastrointestinal defects and immunodeficiency syndrome, 243150
ZAP70	100%	99,30%	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID 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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors