SCID GENE PANEL DG 3.00 (37 genes)

Releasedate: 02-12-2020

Gene	Agilent V5 covered > 10x	Agilent V5 covered > 20x	TWIST covered > 10x	TWIST covered 20x	Associated Phenotype description and OMIM disease ID
ADA	100	99,7	100	100	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
AK2	98,9	94,9	100	99,9	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	99,5	100	100	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	100	100	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD8A	100	99,8	100	100	CD8 deficiency, familial, 608957
CIITA	100	99,5	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CORO1A	100	98,6	100	100	Immunodeficiency 8, 615401
DCLRE1C	100	99,4	100	100	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DOCK2	100	99,6	100	100	Immunodeficiency 40, 616433
DOCK8	100	99,6	100	100	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
FOXN1	100	99,6	100	100	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
IL2RG	99,8	97,1	100	100	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL7R	100	99,8	100	100	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
JAK3	99,9	98,7	100	100	SCID, autosomal recessive, T-negative/B-positive type, 600802

LAT	100	99,2	100	100	Immunodeficiency 52, 617514
LCK	98,9	96,6	100	100	?Immunodeficiency 22, 615758
LIG4	100	99,9	100	100	{Multiple myeloma, resistance to}, 254500 LIG4 syndrome, 606593
NHEJ1	100	96,2	100	100	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
PNP	99,8	98,9	100	100	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PRKDC	99,7	98	100	100	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PTPRC	99	95,1	100	100	{Hepatitis C virus, susceptibility to}, 609532 Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
RAC2	99,9	98,3	100	100	?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986 Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 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	99,7	98,1	100	100	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	100	99,5	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	99,3	97	100	99,9	Bare lymphocyte syndrome, type II, complementation group D, 209920
RMRP					Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
STK4	100	99,8	100	100	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
TAP1	100	99,2	100	100	Bare lymphocyte syndrome, type I, 604571
TAP2	99,9	99,3	100	100	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,5	95,5	96,6	96,6	Bare lymphocyte syndrome, type I, 604571

TTC7A	99,3	95,4	100	100	Gastrointestinal defects and immunodeficiency syndrome, 243150
ZAP70	100	99,3	100	100	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840

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 : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors