SCID GENE PANEL DG 3.2.0 (40 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
ADA	99,7	96,1	100	100	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
AK2	98,7	95,2	100	99,7	Reticular dysgenesis, 267500
B2M	100	100	100	100	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
CD247	100	99,4	100	100	?Immunodeficiency 25, 610163
CD3D	100	99,9	100	100	Immunodeficiency 19, 615617
CD3E	100	98,8	100	100	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	100	100	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD8A	100	100	100	100	CD8 deficiency, familial, 608957
CIITA	100	99,4	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920
CORO1A	99,9	98,9	100	99,9	Immunodeficiency 8, 615401
DCLRE1C	99,8	98,2	100	99,9	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DOCK2	99,8	98,8	100	100	Immunodeficiency 40, 616433
DOCK8	100	99	100	100	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
FCHO1	99,3	97,5	100	100	Immunodeficiency 76, 619164
FOXN1	100	99,1	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	92,8	100	100	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL7R	99,9	99,3	100	100	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
JAK3	99,5	97,6	100	100	SCID, autosomal recessive, T-negative/B-positive type, 600802
LAT	100	99,4	100	100	Immunodeficiency 52, 617514

LCK	98,2	96,1	100	100	?Immunodeficiency 22, 615758
LIG4	99,8	99,3	100	100	LIG4 syndrome, 606593
NHEJ1	99,8	97,2	100	100	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
PAX1	92,6	87,5	100	99,7	Otofaciocervical syndrome 2, 615560
PNP	99,8	98,7	100	100	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PRKDC	99,2	96,9	100	100	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PTPRC	98,8	93,9	100	99,9	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
RAC2	99,8	95,4	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	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	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	99,7	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	99,9	98,6	100	100	Bare lymphocyte syndrome, type II, complementation group D, 209920
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
SCIMP	80,4	79,9	97,8	89,1	No OMIM disease ID
STK4	99,9	99,7	100	100	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
TAP1	100	97,6	100	100	Bare lymphocyte syndrome, type I, 604571
TAP2	99,9	98,6	100	100	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,5	95,1	96,6	96,6	Bare lymphocyte syndrome, type I, 604571
TTC7A	99,6	97,1	100	100	Gastrointestinal defects and immunodeficiency syndrome, 243150
ZAP70	100	99,7	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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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-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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors