SEVERE COMBINED IMMUNODEFICIENCY (SCID) GENE PANEL DG 3.3.0 (41 genes)

Releasedate: 13-01-2022

| Gene | TWIST covered >10x | TWIST covered >20x | Associated Phenotype Description and OMIM disease ID |
|---------|--------------------------|--------------------------|--|
| ADA | 100% | 100% | Adenosine deaminase deficiency, partial, 102700 |
| | | | Severe combined immunodeficiency due to ADA deficiency, 102700 |
| AK2 | 100% | 100% | Reticular dysgenesis, 267500 |
| B2M | 100% | 100% | ?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600 |
| CD247 | 100% | 100% | ?Immunodeficiency 25, 610163 |
| CD3D | 100% | 100% | Immunodeficiency 19, 615617 |
| CD3E | 100% | 100% | Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615 |
| CD3G | 100% | 100% | Immunodeficiency 17, CD3 gamma deficient, 615607 |
| CD8A | 100% | 100% | CD8 deficiency, familial, 608957 |
| CIITA | 100% | 100% | Bare lymphocyte syndrome, type II, complementation group A, 209920 |
| CORO1A | 100% | 100% | Immunodeficiency 8, 615401 |
| DCLRE1C | 100% | 100% | Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554 |
| DOCK2 | 100% | 100% | Immunodeficiency 40, 616433 |
| DOCK8 | 100% | 100% | Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 |
| FCHO1 | 100% | 100% | Immunodeficiency 76, 619164 |
| FOXN1 | 100% | 100% | T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 |
| IL2RG | 100% | 100% | Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400 |
| IL7R | 100% | 100% | Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971 |
| ITPKB | 100% | 100% | No OMIM disease ID |
| JAK3 | 100% | 100% | SCID, autosomal recessive, T-negative/B-positive type, 600802 |
| LAT | 100% | 100% | Immunodeficiency 52, 617514 |

| LCK 100% 100% ? Immunodeficiency 22, 615758 LCP2 100% 100% 100% LIG4 syndrome, 606593 LIG4 100% 100% Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 PAX1 100% 100% Otdoorcevical syndrome 2, 615560 PNP 100% 100% Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 PRKDC 100% 100% Immunodeficiency 26, with or without neurologic abnormalities, 615966 PTRC 100% 100% Immunodeficiency 26, with or without neurologic abnormalities, 615966 RAC2 100% 100% Immunodeficiency 73 with defective neutrophil chemotaxis and leukocytosis, 608203 RRAC2 100% 100% Severe combined immunodeficiency, 71, retain-equity, enemotaxis and hypogammaglobulinemia, 618987 RAG2 100% 100% Omenn syndrome, 603554 Severe combined immunodeficiency, 8 cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 RFX5 100% 100% Severe combined immunodeficiency, 6, B cell-negative, 601457 Combined cellular | | | | |
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| NHEI1 100% 100% Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 PAXI 100% 100% Otofaciocervical syndrome 2, 615560 PRND 100% 100% Immunodeficiency 26, with or without neurologic abnormalities, 615966 PTPRC 100% 100% Severe combined immunodeficiency, 7 cell-negative, 8-cell/natural killer-cell positive, 608971 RAC2 100% 100% Immunodeficiency 73A with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 immunodeficiency 73S with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 immunodeficiency 73S with defective neutrophil chemotaxis and lymphopenia, 618986 RAG1 100% 100% Omen syndrome, 603554 Severe combined ellular and humoral immunodeficiency, 8 cell-negative, 601457 Severe combined ellular and humoral immunodeficiency, 8 cell-negative, 601457 Combined cellular and humoral immunodeficiency, 8 cell-negative, 601457 Combined cellular and humoral immunodeficiency, 8 cell-negative, 601457 RFX5 100% 100% Bare lymphocyte syndrome, type II, complementation group C, 209920 RFXANK 100% 100% Bare lymphocyte syndrome, type II, complementation group B, 209920 RFXAP 100% 100% | LCP2 | 100% | 100% | ?Immunodeficiency 81, 619374 |
| PAX1 100% 100% Otofaciocervical syndrome 2, 615560 PNP 100% 100% Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 PRKDC 100% 100% Immunodeficiency 26, with or without neurologic abnormalities, 615966 PTRRC 100% 100% Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 RAC2 100% 100% Immunodeficiency 73A with defective neutrophil chemotaxis and lytopagmmaglobulinemia, 618987 immunodeficiency, 73B with defective neutrophil chemotaxis and lytopagmmaglobulinemia, 618986 RAG1 100% 100% Omenn syndrome, 603554 Severe combined ellular and humoral immune defects with granulomas, 233650 RAG2 100% 100% Severe combined ellular and humoral immune defects with granulomas, 233650 RFX5 100 100% Bare lymphocyte syndrome, type II, complementation group E, 209920 RFXANK 100 MHC class II deficiency, surforme, type II, complementation group E, 209920 RFXANK 100% Bare lymphocyte syndrome, type II, complementation group D, 209920 RFXAP NC NC Anauxetic dysplasia vithout hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250 | LIG4 | 100% | 100% | LIG4 syndrome, 606593 |
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| Cartilage-hair hypoplasia, 250250 STK4 100% 100% T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 TAP1 100% 100% Bare lymphocyte syndrome, type I, 604571 TAP2 100% 100% Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 TAPBP 96% 96% Bare lymphocyte syndrome, type I, 604571 TTC7A 100% 100% Gastrointestinal defects and immunodeficiency syndrome, 243150 ZAP70 100% 100% Immunodeficiency 48, 269840 | RMRP | NC | NC | Anauxetic dysplasia 1, 607095 |
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| TTC7A 100% 100% Gastrointestinal defects and immunodeficiency syndrome, 243150 ZAP70 100% 100% Immunodeficiency 48, 269840 | TAP2 | 100% | 100% | Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 |
| ZAP70 100% 100% Immunodeficiency 48, 269840 | TAPBP | 96% | 96% | Bare lymphocyte syndrome, type I, 604571 |
| ' ' | TTC7A | 100% | 100% | Gastrointestinal defects and immunodeficiency syndrome, 243150 |
| Autoimmune disease, multisystem, infantile-onset, 2, 617006 | ZAP70 | 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. 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 is the chemistry used for 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: January 13th, 2022.

This list is accurate for panel version DG 3.3.0

Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors