# PID SCID panel

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| **versie** | v2-v3 (36 genen) | Centrum voor Medische Genetica Gent |

| Gene | OMIM gene ID | Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern |
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| *ADA* | 608958 | Adenosine deaminase deficiency, partial, 102700 (3), Autosomal recessive, Somatic mosaicism; Severe combined immunodeficiency due to ADA deficiency, 102700 (3), Autosomal recessive, Somatic mosaicism |
| *AK2* | 103020 | Reticular dysgenesis, 267500 (3), Autosomal recessive |
| *CARD11* | 607210 | B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive; Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant |
| *CD247* | 186780 | ?Immunodeficiency 25, 610163 (3), Autosomal recessive |
| *CD3D* | 186790 | Immunodeficiency 19, 615617 (3), Autosomal recessive |
| *CD3E* | 186830 | Immunodeficiency 18, 615615 (3), Autosomal recessive; Immunodeficiency 18, SCID variant, 615615 (3), Autosomal recessive |
| *CD3G* | 186740 | Immunodeficiency 17, CD3 gamma deficient, 615607 (3), Autosomal recessive |
| *CD8A* | 186910 | CD8 deficiency, familial, 608957 (3), Autosomal recessive |
| *CIITA* | 600005 | Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive; {Rheumatoid arthritis, susceptibility to}, 180300 (3) |
| *CORO1A* | 605000 | Immunodeficiency 8, 615401 (3), Autosomal recessive |
| *DCLRE1C* | 605988 | Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, Athabascan type, 602450 (3), Autosomal recessive |
| *DOCK8* | 611432 | Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive |
| *FOXN1* | 600838 | T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal recessive |
| *IL15RA* | 601070 | No OMIM phenotype |
| *IL2RG* | 308380 | Combined immunodeficiency, X-linked, moderate, 312863 (3), X-linked recessive; Severe combined immunodeficiency, X-linked, 300400 (3), X-linked recessive |
| *IL7R* | 146661 | Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971 (3), Autosomal recessive |
| *JAK3* | 600173 | SCID, autosomal recessive, T-negative/B-positive type, 600802 (3), Autosomal recessive |
| *LCK* | 153390 | ?Immunodeficiency 22, 615758 (3), Autosomal recessive |

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| *LIG4* | 601837 | LIG4 syndrome, 606593 (3); {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation |
| *NBN* | 602667 | Aplastic anemia, 609135 (3); Leukemia, acute lymphoblastic, 613065 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive |
| *NHEJ1* | 611290 | Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 (3) |
| *PGM3* | 172100 | Immunodeficiency 23, 615816 (3), Autosomal recessive |
| *PNP* | 164050 | Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive |
| *PRKDC* | 600899 | Immunodeficiency 26, with or without neurologic abnormalities, 615966 (3), Autosomal recessive |
| *PTPRC* | 151460 | {Hepatitis C virus, susceptibility to}, 609532 (3); Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 (3), Autosomal recessive |
| *RAG1* | 179615 | Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (3); Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive |
| *RAG2* | 179616 | Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive |
| *RFX5* | 601863 | Bare lymphocyte syndrome, type II, complementation group C, 209920 (3), Autosomal recessive; Bare lymphocyte syndrome, type II, complementation group E, 209920 (3), Autosomal recessive |
| *RFXANK* | 603200 | MHC class II deficiency, complementation group B, 209920 (3), Autosomal recessive |
| *RFXAP* | 601861 | Bare lymphocyte syndrome, type II, complementation group D, 209920 (3), Autosomal recessive |
| *RMRP* | 157660 | Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive |
| *STIM1* | 605921 | Immunodeficiency 10, 612783 (3), Autosomal recessive; Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant |
| *TBX1* | 602054 | Conotruncal anomaly face syndrome, 217095 (3); DiGeorge syndrome, 188400 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Velocardiofacial syndrome, 192430 (3), Autosomal dominant |
| *TTC7A* | 609332 | Gastrointestinal defects and immunodeficiency syndrome, 243150 (3), Autosomal recessive |

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| *WAS* | 300392 | Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive |
| *ZAP70* | 176947 | Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive; Immunodeficiency 48, 269840 (3), Autosomal recessive |

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: June 06, 2017

Possible phenotype mapping keys
(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
(2) the disorder has been placed on the map by linkage; no mutation has been found
(3) the molecular basis for the disorder is known; a mutation has been found in the gene
(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).
A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.