PID CVID panel

versie	v3-v4-v5 (33 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ADA2 (CECR1)	607575	Polyarteritis nodosa, childhood-onset, 615688 (3), Autosomal recessive; ?Sneddon syndrome, 182410 (3), Autosomal recessive
AICDA	605257	Immunodeficiency with hyper-IgM, type 2, 605258 (3), Autosomal recessive
BLK	191305	Maturity-onset diabetes of the young, type 11, 613375 (3), Autosomal dominant
BLNK	604515	Agammaglobulinemia 4, 613502 (3), Autosomal recessive
BTK	300300	Agammaglobulinemia and isolated hormone deficiency, 307200 (3), X-linked recessive; Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive
CD19	107265	Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive
CD40	109535	Immunodeficiency with hyper-IgM, type 3, 606843 (3), Autosomal recessive
CD40LG	300386	Immunodeficiency, X-linked, with hyper-IgM, 308230 (3), X-linked recessive
CD79A	112205	Agammaglobulinemia 3, 613501 (3), Autosomal recessive
CD79B	147245	Agammaglobulinemia 6, 612692 (3), Autosomal recessive
CD81	186845	Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive
CR2	120650	Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive; {Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3)
CTLA4	123890	Autoimmune lymphoproliferative syndrome, type V, 616100 (3), Autosomal dominant; {Celiac disease, susceptibility to, 3}, 609755 (3); {Diabetes mellitus, insulin-dependent, 12}, 601388 (3); {Hashimoto thyroiditis}, 140300 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
ICOS	604558	Immunodeficiency, common variable, 1, 607594 (3), Autosomal recessive
IGHM	147020	Agammaglobulinemia 1, 601495 (3), Autosomal recessive
IGLL1	146770	Agammaglobulinemia 2, 613500 (3), Autosomal recessive
IKZF1	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant

IL21	605384	?Immunodeficiency, common variable, 11, 615767 (3), Autosomal recessive
IL21R	605383	[IgE, elevated level of], 147050 (3), Autosomal dominant; Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 (3), Autosomal recessive
LRBA	606453	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive
MS4A1	112210	Immunodeficiency, common variable, 5, 613495 (3), Autosomal recessive
NFKB1	164011	Immunodeficiency, common variable, 12, 616576 (3), Autosomal dominant
NFKB2	164012	Immunodeficiency, common variable, 10, 615577 (3), Autosomal dominant
PIK3CD	602839	Immunodeficiency 14, 615513 (3), Autosomal dominant
PIK3R1	171833	?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; Immunodeficiency 36, 616005 (3), Autosomal dominant; SHORT syndrome, 269880 (3), Autosomal dominant
PLCG2	600220	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 (3), Autosomal dominant; Familial cold autoinflammatory syndrome 3, 614468 (3), Autosomal dominant
PRKCD	176977	Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive
RAC2	602049	Neutrophil immunodeficiency syndrome, 608203 (3)
TCF7L1	604652	No OMIM phenotype
TNFRSF13B	604907	Immunodeficiency, common variable, 2, 240500 (3), Autosomal recessive, Autosomal dominant; Immunoglobulin A deficiency 2, 609529 (3)
TNFRSF13C	606269	Immunodeficiency, common variable, 4, 613494 (3), Autosomal recessive
UNG	191525	Immunodeficiency with hyper IgM, type 5, 608106 (3), Autosomal recessive
VAV1	164875	No OMIM phenotype

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.