PID Th17_CMC panel

versie v3 (15 genen) Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
AIRE	607358	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal recessive, Autosomal dominant
CARD9	607212	Candidiasis, familial, 2, autosomal recessive, 212050 (3), Autosomal recessive
CLEC7A	606264	{Aspergillosis, susceptibility to}, 614079 (3); Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive
DOCK8	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive
IL12B	161561	Immunodeficiency 29, mycobacteriosis, 614890 (3), Autosomal recessive
IL12RB1	601604	Immunodeficiency 30, 614891 (3), Autosomal recessive
IL17F	606496	?Candidiasis, familial, 6, autosomal dominant, 613956 (3)
IL17RA	605461	Immunodeficiency 51, 613953 (3), Autosomal recessive
IL17RC	610925	Candidiasis, familial, 9, 616445 (3), Autosomal recessive
PGM3	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
RORC	602943	Immunodeficiency 42, 616622 (3), Autosomal recessive
STAT1	600555	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 (3), Autosomal dominant; Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 (3), Autosomal recessive; Immunodeficiency 31C, autosomal dominant, 614162 (3), Autosomal dominant
STAT3	102582	Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant; Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant
TRAF3IP2	607043	?Candidiasis, familial, 8, 615527 (3), Autosomal recessive; {Psoriasis susceptibility 13}, 614070 (3)
TYK2	176941	Immunodeficiency 35, 611521 (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.