PID_diagnostic panel

versie	v6 (483 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ACD	609377	?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant
ACP5	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
ACTB	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant
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
ADA2	607575	Sneddon syndrome, 182410 (3), Autosomal recessive; Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 (3), Autosomal recessive
ADAM17	603639	?Inflammatory skin and bowel disease, neonatal, 1, 614328 (3), Autosomal recessive
ADAR	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
AICDA	605257	Immunodeficiency with hyper-IgM, type 2, 605258 (3), Autosomal recessive
AIRE	607358	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal recessive, Autosomal dominant
AK2	103020	Reticular dysgenesis, 267500 (3), Autosomal recessive
ALPI	171740	No OMIM phenotype
AP1S3	615781	{Psoriasis 15, pustular, susceptibility to}, 616106 (3), Autosomal dominant
AP3B1	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP3D1	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
APOL1	603743	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 (3); {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 (3)
ARHGEF1	601855	?Immunodeficiency 62, 618459 (3), Autosomal recessive
ARPC1B	604223	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718 (3), Autosomal recessive
ATG16L1	610767	{Inflammatory bowel disease (Crohn disease) 10}, 611081 (3)
ATG4A	300663	No OMIM phenotype

ATM	607585	Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3)
ATP2A2	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
ATP6AP1	300197	Immunodeficiency 47, 300972 (3), X-linked recessive
B2M	109700	?Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Immunodeficiency 43, 241600 (3), Autosomal recessive
BACH2	605394	Immunodeficiency 60, 618394 (3), Autosomal dominant
BCL10	603517	{Lymphoma, follicular, somatic}, 605027 (3); ?Immunodeficiency 37, 616098 (3), Autosomal recessive; {Sezary syndrome, somatic} (3); {Male germ cell tumor, somatic}, 273300 (3); Lymphoma, MALT, somatic, 137245 (3); {Mesothelioma, somatic}, 156240 (3)
BCL11B	606558	Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (3), Autosomal dominant; Immunodeficiency 49, 617237 (3), Autosomal dominant
BLK	191305	Maturity-onset diabetes of the young, type 11, 613375 (3), Autosomal dominant
BLM	604610	Bloom syndrome, 210900 (3), Autosomal recessive
BLNK	604515	?Agammaglobulinemia 4, 613502 (3), Autosomal recessive
BLOC1S3	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
BLOC1S6	604310	?Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive
BTK	300300	Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive; Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 (3), X-linked recessive
C1QA	120550	C1q deficiency, 613652 (3), Autosomal recessive
C1QB	120570	C1q deficiency, 613652 (3), Autosomal recessive
C1QC	120575	C1q deficiency, 613652 (3), Autosomal recessive
C1R	613785	Ehlers-Danlos syndrome, periodontal type, 1, 130080 (3), Autosomal dominant
C1S	120580	C1s deficiency, 613783 (3); Ehlers-Danlos syndrome, periodontal type, 2, 617174 (3), Autosomal dominant
C2	613927	C2 deficiency, 217000 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant
C2orf69	619219	Combined oxidative phosphorylation deficiency 53, 619423 (3), Autosomal recessive
C3	120700	C3 deficiency, 613779 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3), Autosomal dominant; {Macular degeneration, age-related, 9}, 611378 (3)
C4A	120810	[Blood group, Rodgers], 614374 (3); C4a deficiency, 614380 (3), Autosomal recessive
C4B	120820	C4B deficiency, 614379 (3)
C4BPA	120830	No OMIM phenotype

C5	120900	C5 deficiency, 609536 (3), Autosomal recessive; [Eculizumab, poor response to], 615749 (3), Autosomal dominant
C6	217050	C6 deficiency, 612446 (3); Combined C6/C7 deficiency (3)
C7	217070	C7 deficiency, 610102 (3)
C8A	120950	C8 deficiency, type I, 613790 (3), Autosomal recessive
C8B	120960	C8 deficiency, type II, 613789 (3), Autosomal recessive
C8G	120930	No OMIM phenotype
C9	120940	C9 deficiency, 613825 (3); {Macular degeneration, age-related, 15, susceptibility to}, 615591 (3)
CARD11	607210	B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive
CARD14	607211	Psoriasis 2, 602723 (3), Autosomal dominant; Pityriasis rubra pilaris, 173200 (3), Autosomal dominant
CARD9	607212	Candidiasis, familial, 2, autosomal recessive, 212050 (3), Autosomal recessive
CARMIL2	610859	Immunodeficiency 58, 618131 (3), Autosomal recessive
CASP10	601762	Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
CASP8	601763	{Breast cancer, protection against}, 114480 (3), Somatic mutation, Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 (3), Autosomal recessive; {Lung cancer, protection against}, 211980 (3), Somatic mutation, Autosomal dominant
CBL	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant
CCBE1	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
CCDC28B	610162	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive
CD19	107265	Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive
CD247	186780	?Immunodeficiency 25, 610163 (3), Autosomal recessive
CD27	186711	Lymphoproliferative syndrome 2, 615122 (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
CD4	186940	Immunodeficiency 79, 619238 (3), Autosomal recessive; OKT4 epitope deficiency, 613949 (3)

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
CD46	120920	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 (3), Autosomal recessive, Autosomal dominant
CD55	125240	[Blood group Cromer], 613793 (3), Autosomal recessive; Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 (3), Autosomal recessive
CD59	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive
CD70	602840	Lymphoproliferative syndrome 3, 618261 (3), Autosomal 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
CD8A	186910	CD8 deficiency, familial, 608957 (3), Autosomal recessive
CDC42	116952	Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant
CDCA7	609937	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910 (3), Autosomal recessive
CEBPE	600749	Specific granule deficiency, 245480 (3), Autosomal recessive
CFB	138470	?Complement factor B deficiency, 615561 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant
CFD	134350	Complement factor D deficiency, 613912 (3), Autosomal recessive
CFH	134370	{Macular degeneration, age-related, 4}, 610698 (3); Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal recessive, Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal recessive, Autosomal dominant
CFHR1	134371	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant
CFHR2	600889	No OMIM phenotype
CFHR3	605336	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant
CFHR4	605337	No OMIM phenotype
CFHR5	608593	Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant
CFI	217030	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant; Complement factor I deficiency, 610984 (3), Autosomal recessive
CFP	300383	Properdin deficiency, X-linked, 312060 (3), X-linked recessive

CFTR	602421	Cystic fibrosis, 219700 (3), Autosomal recessive; Sweat chloride elevation without CF (3); Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; {Hypertrypsinemia, neonatal} (3)
CHD7	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CIB1	602293	Epidermodysplasia verruciformis 3, 618267 (3), Autosomal recessive
CIITA	600005	{Rheumatoid arthritis, susceptibility to}, 180300 (3); Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive
CLCN7	602727	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant
CLEC7A	606264	Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive; {Aspergillosis, susceptibility to}, 614079 (3)
CLPB	616254	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 (3), Autosomal recessive
COL7A1	120120	EBD, localisata variant (3); Epidermolysis bullosa, pretibial, 131850 (3), Autosomal recessive, Autosomal dominant; Transient bullous of the newborn, 131705 (3), Autosomal recessive, Autosomal dominant; EBD, Bart type, 132000 (3), Autosomal dominant; Epidermolysis bullosa dystrophica, AD, 131750 (3), Autosomal dominant; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal recessive, Autosomal dominant; EBD inversa, 226600 (3), Autosomal recessive; Epidermolysis bullosa dystrophica, AR, 226600 (3), Autosomal recessive; Toenail dystrophy, isolated, 607523 (3), Autosomal dominant
COPA	601924	{Autoimmune interstitial lung, joint, and kidney disease}, 616414 (3), Autosomal dominant
CORO1A	605000	Immunodeficiency 8, 615401 (3), Autosomal recessive
CPT2	600650	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant; CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant
CR2	120650	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3); Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive
CREBBP	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CSF2RA	306250	Surfactant metabolism dysfunction, pulmonary, 4, 300770 (3)

CSF2RB	138981	Surfactant metabolism dysfunction, pulmonary, 5, 614370 (3), Autosomal recessive
CSF3R	138971	Neutropenia, severe congenital, 7, autosomal recessive, 617014 (3), Autosomal recessive
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTLA4	123890	Autoimmune lymphoproliferative syndrome, type V, 616100 (3), Autosomal dominant; {Diabetes mellitus, insulin-dependent, 12}, 601388 (3); {Celiac disease, susceptibility to, 3}, 609755 (3); {Hashimoto thyroiditis}, 140300 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
CTNNBL1	611537	No OMIM phenotype
CTPS1	123860	Immunodeficiency 24, 615897 (3), Autosomal recessive
CTSC	602365	Periodontitis 1, juvenile, 170650 (3), Autosomal recessive; Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive
CXCR4	162643	WHIM syndrome 1, 193670 (3), Autosomal dominant; Myelokathexis, isolated, 193670 (3), Autosomal dominant
CYBA	608508	Chronic granulomatous disease 4, autosomal recessive, 233690 (3), Autosomal recessive
CYBB	300481	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive; Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive
CYBC1	618334	Chronic granulomatous disease 5, autosomal recessive, 618935 (3), Autosomal recessive
DBR1	607024	{Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441 (3), Autosomal recessive
DCLRE1B	609683	No OMIM phenotype
DCLRE1C	605988	Severe combined immunodeficiency, Athabascan type, 602450 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive
DDX58	609631	Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant
DEF6	610094	No OMIM phenotype
DGKE	601440	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (3), Autosomal recessive
DHFR	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
DKC1	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DNAJC21	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
DNASE1	125505	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
DNASE1L3	602244	Systemic lupus erythematosus 16, 614420 (3), Autosomal recessive
DNASE2	126350	No OMIM phenotype

DNMT3B	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive
DOCK2	603122	Immunodeficiency 40, 616433 (3), Autosomal recessive
DOCK8	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive
DOK3	611435	No OMIM phenotype
DTNBP1	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
EFL1	617538	Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive
ELANE	130130	Neutropenia, cyclic, 162800 (3), Autosomal dominant; Neutropenia, severe congenital 1, autosomal dominant, 202700 (3), Autosomal dominant
ELF4	300775	No OMIM phenotype
EPG5	615068	Vici syndrome, 242840 (3), Autosomal recessive
ERBIN	606944	No OMIM phenotype
ERCC6L2	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
EXTL3	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
F12	610619	Angioedema, hereditary, 3, 610618 (3), Autosomal dominant; Factor XII deficiency, 234000 (3), Autosomal recessive
FAAP24	610884	No OMIM phenotype
FADD	602457	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759 (3), Autosomal recessive
FAS	134637	Squamous cell carcinoma, burn scar-related, somatic (3); Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant
FASLG	134638	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant
FAT4	612411	Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive
FCGR2B	604590	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3)
FCGR3A	146740	Immunodeficiency 20, 615707 (3), Autosomal recessive
FCGR3B	610665	No OMIM phenotype
FCHO1	613437	Immunodeficiency 76, 619164 (3), Autosomal recessive
FCN3	604973	Immunodeficiency due to ficolin 3 deficiency, 613860 (3), Autosomal recessive
FERMT1	607900	Kindler syndrome, 173650 (3), Autosomal recessive
FERMT3	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive
FNIP1	610594	No OMIM phenotype
FOXN1	600838	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 (3); T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal recessive

FOXP3	300292	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive
FPR1	136537	No OMIM phenotype
G6PC3	611045	Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive
G6PD	305900	Hemolytic anemia, G6PD deficient (favism), 300908 (3), X-linked dominant; {Resistance to malaria due to G6PD deficiency}, 611162 (3)
GATA1	305371	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 (3); Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3), X-linked recessive; Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3), X-linked recessive; Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3), X-linked recessive
GATA2	137295	{Leukemia, acute myeloid, susceptibility to}, 601626 (3), Somatic mutation, Autosomal dominant; Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3)
GFI1	600871	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 (3), Autosomal dominant; Neutropenia, severe congenital 2, autosomal dominant, 613107 (3), Autosomal dominant
GIMAP5	608086	No OMIM phenotype
GINS1	610608	Immunodeficiency 55, 617827 (3), Autosomal recessive
GUCY2C	601330	Diarrhea 6, 614616 (3), Autosomal dominant; Meconium ileus, 614665 (3), Autosomal recessive
HAVCR2	606652	T-cell lymphoma, subcutaneous panniculitis-like, 618398 (3), Autosomal recessive
HAX1	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
HELLS	603946	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911 (3), Autosomal recessive
HMOX1	141250	Heme oxygenase-1 deficiency, 614034 (3); {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (3)
HPS1	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
HPS4	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
HPS6	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
HTRA2	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
HYOU1	601746	?Immunodeficiency 59 and hypoglycemia, 233600 (3), Autosomal recessive
ICOS	604558	Immunodeficiency, common variable, 1, 607594 (3), Autosomal recessive
ICOSLG	605717	No OMIM phenotype
IFIH1	606951	Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant

IFNAR1	107450	No OMIM phenotype
IFNAR2	602376	{Hepatitis B virus, susceptibility to}, 610424 (3); ?Immunodeficiency 45, 616669 (3), Autosomal recessive
IFNG	147570	{Hepatitis C virus, response to therapy of}, 609532 (3); {TSC2 angiomyolipomas, renal, modifier of}, 613254 (3), Autosomal dominant; {Aplastic anemia}, 609135 (3); ?Immunodeficiency 69, mycobacteriosis, 618963 (3), Autosomal recessive; {Tuberculosis, protection against}, 607948 (3); {AIDS, rapid progression to}, 609423 (3)
IFNGR1	107470	{H. pylori infection, susceptibility to}, 600263 (3); Immunodeficiency 27A, mycobacteriosis, AR, 209950 (3), Autosomal recessive; Immunodeficiency 27B, mycobacteriosis, AD, 615978 (3), Autosomal dominant; {Tuberculosis infection, protection against}, 607948 (3); {Tuberculosis, susceptibility to}, 607948 (3); {Hepatitis B virus infection, susceptibility to}, 610424 (3)
IFNGR2	147569	Immunodeficiency 28, mycobacteriosis, 614889 (3), Autosomal recessive
IGHM	147020	Agammaglobulinemia 1, 601495 (3), Autosomal recessive
IGKC	147200	Kappa light chain deficiency, 614102 (3), Autosomal recessive
IGLL1	146770	Agammaglobulinemia 2, 613500 (3), Autosomal recessive
IKBKB	603258	Immunodeficiency 15B, 615592 (3), Autosomal recessive; Immunodeficiency 15A, 618204 (3), Autosomal dominant
IKBKG	300248	Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive
IKZF1	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
IL10	124092	{Rheumatoid arthritis, progression of}, 180300 (3); {Graft-versus-host disease, protection against}, 614395 (3); {HIV-1, susceptibility to}, 609423 (3)
IL10RA	146933	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148 (3), Autosomal recessive
IL10RB	123889	{Hepatitis B virus, susceptibility to}, 610424 (3); Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 (3), Autosomal recessive
IL12B	161561	Immunodeficiency 29, mycobacteriosis, 614890 (3), Autosomal recessive
IL12RB1	601604	Immunodeficiency 30, 614891 (3), Autosomal recessive
IL12RB2	601642	No OMIM phenotype
IL15RA	601070	No OMIM phenotype
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
IL18	600953	No OMIM phenotype
IL18BP	604113	{?Hepatitis, fulminant viral, susceptibility to}, 618549 (3), Autosomal recessive
IL1RL1	601203	No OMIM phenotype

IL1RN	147679	{Gastric cancer risk after H. pylori infection}, 137215 (3), Autosomal dominant; {Microvascular complications of diabetes 4}, 612628 (3); Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive
IL21	605384	?Immunodeficiency, common variable, 11, 615767 (3), Autosomal recessive
IL21R	605383	[IgE, elevated level of], 147050 (3), Autosomal dominant; Immunodeficiency 56, 615207 (3), Autosomal recessive
IL23R	607562	{Inflammatory bowel disease 17, protection against}, 612261 (3); {Psoriasis, protection against}, 605606 (3)
IL2RA	147730	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive; {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3)
IL2RB	146710	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495 (3), Autosomal recessive
IL2RG	308380	Combined immunodeficiency, X-linked, moderate, 312863 (3), X-linked recessive; Severe combined immunodeficiency, X-linked, 300400 (3), X-linked recessive
IL36RN	605507	Psoriasis 14, pustular, 614204 (3), Autosomal recessive
IL6R	147880	[Interleukin 6, serum level of, QTL], 614752 (3); Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944 (3), Autosomal recessive; [Interleukin-6 receptor, soluble, serum level of, QTL], 614689 (3)
IL6ST	600694	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523 (3), Autosomal recessive
IL7R	146661	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971 (3), Autosomal recessive
ILRUN (C6orf106)	612217	No OMIM phenotype
INO80	610169	No OMIM phenotype
IRAK1	300283	No OMIM phenotype
IRAK4	606883	Immunodeficiency 67, 607676 (3), Autosomal recessive
IRF2BP2	615332	?Immunodeficiency, common variable, 14, 617765 (3), Autosomal dominant
IRF3	603734	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532 (3), Autosomal dominant
IRF4	601900	[Skin/hair/eye pigmentation, variation in, 8], 611724 (3)
IRF7	605047	?Immunodeficiency 39, 616345 (3), Autosomal recessive
IRF8	601565	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 (3), Autosomal dominant; Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 (3), Autosomal recessive
IRF9	147574	Immunodeficiency 65, susceptibility to viral infections, 618648 (3), Autosomal recessive
ISG15	147571	Immunodeficiency 38, 616126 (3), Autosomal recessive

ITCH	606409	Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3), Autosomal recessive
ITGB2	600065	Leukocyte adhesion deficiency, 116920 (3), Autosomal recessive
ITK	186973	Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive
ITPKB	147522	No OMIM phenotype
IVNS1ABP	609209	Immunodeficiency 70, 618969 (3), Autosomal dominant
JAGN1	616012	Neutropenia, severe congenital, 6, autosomal recessive, 616022 (3), Autosomal recessive
JAK1	147795	Autoinflammation, immune dysregulation, and eosinophilia, 618999 (3), Autosomal dominant
JAK3	600173	SCID, autosomal recessive, T-negative/B-positive type, 600802 (3), Autosomal recessive
KDM6A	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KMT2A	159555	Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
KMT2D	602113	Kabuki syndrome 1, 147920 (3), Autosomal dominant
KRAS	190070	Gastric cancer, somatic, 137215 (3); Oculoectodermal syndrome, somatic, 600268 (3); Breast cancer, somatic, 114480 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Lung cancer, somatic, 211980 (3); Pancreatic carcinoma, somatic, 260350 (3); Leukemia, acute myeloid, somatic, 601626 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3)
LACC1	613409	Juvenile arthritis, 618795 (3), Autosomal recessive
LAMTOR2	610389	:Immunodeficiency due to defect in MAPBP-interacting protein, 610798 (3), Autosomal recessive
LAT	602354	Immunodeficiency 52, 617514 (3), Autosomal recessive
LCK	153390	?Immunodeficiency 22, 615758 (3), Autosomal recessive
LCP2	601603	?Immunodeficiency 81, 619374 (3), Autosomal recessive
LIG1	126391	No OMIM phenotype
LIG4	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
LIPA	613497	Wolman disease, 278000 (3), Autosomal recessive; Cholesteryl ester storage disease, 278000 (3), Autosomal recessive
LPIN2	605519	Majeed syndrome, 609628 (3)
LRBA	606453	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive
LRRC8A	608360	?Agammaglobulinemia 5, 613506 (3), Autosomal dominant
LSM11	617910	No OMIM phenotype
LYST	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
MAGT1	300715	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3), X-linked recessive; Congenital disorder of glycosylation, type Icc, 301031 (3), X-linked recessive

MALT1	604860	Immunodeficiency 12, 615468 (3), Autosomal recessive
MAN2B1	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MAP1LC3B2	No OMIM gene	No OMIM phenotype
MAP3K14	604655	No OMIM phenotype
MAPK8	601158	No OMIM phenotype
MASP1	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
MASP2	605102	MASP2 deficiency, 613791 (3), Autosomal recessive
MBL2	154545	{Chronic infections, due to MBL deficiency}, 614372 (3), Autosomal dominant
MCM10	609357	Immunodeficiency 80 with or without cardiomyopathy, 619313 (3), Autosomal recessive
MCM4	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive
MEFV	608107	Neutrophilic dermatosis, acute febrile, 608068 (3), Autosomal dominant; Familial Mediterranean fever, AR, 249100 (3), Autosomal recessive; Familial Mediterranean fever, AD, 134610 (3), Autosomal dominant
MOGS	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
MPEG1	610390	Immunodeficiency 77, 619223 (3), Autosomal dominant
MPO	606989	{Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; Myeloperoxidase deficiency, 254600 (3), Autosomal recessive; {Lung cancer, protection against, in smokers} (3)
MRE11	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
MRTFA	606078	?Immunodeficiency 66, 618847 (3), Autosomal recessive
MS4A1	112210	?Immunodeficiency, common variable, 5, 613495 (3), Autosomal recessive
MSH6	600678	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 (3), Autosomal dominant; Mismatch repair cancer syndrome 3, 619097 (3), Autosomal recessive; {Endometrial cancer, familial}, 608089 (3), Somatic mutation, Autosomal dominant
MSN	309845	Immunodeficiency 50, 300988 (3), X-linked recessive
MTHFD1	172460	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 (3), Autosomal recessive
MVK	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
MYD88	602170	Macroglobulinemia, Waldenstrom, somatic, 153600 (3); Immunodeficiency 68, 612260 (3), Autosomal recessive
MYO5B	606540	Diarrhea 2, with microvillus atrophy, 251850 (3), Autosomal recessive
MYSM1	612176	Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive
NBAS	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive

NBN	602667	Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NCF1	608512	Chronic granulomatous disease 1, autosomal recessive, 233700 (3), Autosomal recessive
NCF2	608515	Chronic granulomatous disease 2, autosomal recessive, 233710 (3), Autosomal recessive
NCF4	601488	Chronic granulomatous disease 3, autosomal recessive, 613960 (3), Autosomal recessive
NCKAP1L	141180	Immunodeficiency 72 with autoinflammation, 618982 (3), Autosomal recessive
NCSTN	605254	Acne inversa, familial, 1, 142690 (3), Autosomal dominant
NFAT5	604708	No OMIM phenotype
NFE2L2	600492	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 (3), Autosomal dominant
NFKB1	164011	Immunodeficiency, common variable, 12, 616576 (3), Autosomal dominant
NFKB2	164012	Immunodeficiency, common variable, 10, 615577 (3), Autosomal dominant
NFKBIA	164008	Ectodermal dysplasia and immunodeficiency 2, 612132 (3), Autosomal dominant
NHEJ1	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 (3)
NHP2	606470	Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive
NKX2-5	600584	Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Ventricular septal defect 3, 614432 (3), Autosomal dominant; Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant
NLRC4	606831	?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant; Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant
NLRP1	606636	{Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 (3); ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 (3), Autosomal recessive; Autoinflammation with arthritis and dyskeratosis, 617388 (3), Autosomal recessive, Autosomal dominant; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant
NLRP12	609648	Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant

NLRP3	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NLRP7	609661	Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive
NOD2	605956	Blau syndrome, 186580 (3), Autosomal dominant; {Yao syndrome}, 617321 (3), Multifactorial; {Inflammatory bowel disease 1, Crohn disease}, 266600 (3), Multifactorial
NOP10	606471	Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
NOS2	163730	{Malaria, resistance to}, 611162 (3)
NPC1	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NRAS	164790	Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3)
NSMCE3	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive
OAS1	164350	No OMIM phenotype
ORAI1	610277	Immunodeficiency 9, 612782 (3), Autosomal recessive; Myopathy, tubular aggregate, 2, 615883 (3), Autosomal dominant
OSTM1	607649	Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive
OTULIN	615712	Autoinflammation, panniculitis, and dermatosis syndrome, 617099 (3), Autosomal recessive
PARN	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 (3), Autosomal dominant
PAX1	167411	Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive
PCCA	232000	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PEPD	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
PGM3	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PIK3CD	602839	Immunodeficiency 14A, autosomal dominant, 615513 (3), Autosomal dominant; Immunodeficiency 14B, autosomal recessive, 619281 (3), Autosomal recessive; ?Roifman-Chitayat syndrome, digenic, 613328 (3), Digenic recessive
PIK3CG	601232	No OMIM phenotype

PIK3R1	171833	Immunodeficiency 36, 616005 (3), Autosomal dominant; ?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; 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
PLEKHM1	611466	?Osteopetrosis, autosomal recessive 6, 611497 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 3, 618107 (3), Autosomal dominant
PLG	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Angioedema, hereditary, 4, 619360 (3), Autosomal dominant; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
PMS2	600259	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3); Mismatch repair cancer syndrome 4, 619101 (3), Autosomal recessive
PNP	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive
POLA1	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive
POLD1	174761	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant
POLD2	600815	No OMIM phenotype
POLE	174762	{Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive; IMAGE-I syndrome, 618336 (3), Autosomal recessive
POLE2	602670	No OMIM phenotype
POLR3A	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
POLR3C	617454	No OMIM phenotype
POLR3F	617455	No OMIM phenotype
POMP	613386	Proteasome-associated autoinflammatory syndrome 2, 618048 (3), Autosomal dominant; Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive
PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
PRIM1	176635	No OMIM phenotype
PRKCD	176977	Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive
PRKDC	600899	Immunodeficiency 26, with or without neurologic abnormalities, 615966 (3), Autosomal recessive

PSEN1	104311	Pick disease, 172700 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; ?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant
PSENEN	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant
PSMA3	176843	No OMIM phenotype
PSMB10	176847	Proteasome-associated autoinflammatory syndrome 5, 619175 (3), Autosomal recessive
PSMB4	602177	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591 (3), Autosomal recessive
PSMB8	177046	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040 (3), Autosomal recessive
PSMB9	177045	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591 (3), Autosomal recessive
PSMG2	609702	?Proteasome-associated autoinflammatory syndrome 4, 619183 (3), Autosomal recessive
PSTPIP1	606347	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 (3), Autosomal dominant
PSTPIP2	616046	No OMIM phenotype
PTEN	601728	{Glioma susceptibility 2}, 613028 (3); {Meningioma}, 607174 (3), Autosomal dominant; Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3); Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant
PTPN11	176876	Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
PTPN2	176887	No OMIM phenotype
PTPN6	176883	No OMIM phenotype
PTPRC	151460	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 (3), Autosomal recessive; {Hepatitis C virus, susceptibility to}, 609532 (3)
RAB27A	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
RAC2	602049	Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203 (3), Autosomal dominant; ?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 (3); Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986 (3)

RAG1	179615	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (3)
RAG2	179616	Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive
RANBP2	601181	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033 (3), Autosomal dominant
RASGRP1	603962	Immunodeficiency 64, 618534 (3), Autosomal recessive
RBCK1	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
RC3H1	609424	?Immune dysregulation and systemic hyperinflammation syndrome, 618998 (3)
RECQL4	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive
REL	164910	No OMIM phenotype
RELA	164014	?Mucocutaneous ulceration, chronic, 618287 (3), Autosomal dominant
RELB	604758	?Immunodeficiency 53, 617585 (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
RHOH	602037	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307 (3), Autosomal recessive
RIPK1	603453	Immunodeficiency 57 with autoinflammation, 618108 (3), Autosomal recessive; Autoinflammation with episodic fever and lymphadenopathy, 618852 (3), Autosomal dominant
RMRP	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive
RNASEH2A	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
RNASEH2B	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
RNASEH2C	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
RNF168	612688	RIDDLE syndrome, 611943 (3), Autosomal recessive
RNF31	612487	No OMIM phenotype
RORC	602943	Immunodeficiency 42, 616622 (3), Autosomal recessive

RPSA	150370	Asplenia, isolated congenital, 271400 (3), Autosomal dominant
RTEL1	608833	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant
SAMD9	610456	Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive; Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 (3), Autosomal dominant; MIRAGE syndrome, 617053 (3), Autosomal dominant
SAMD9L	611170	Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant; Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 (3), Autosomal dominant
SAMHD1	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
SASH3	300441	No OMIM phenotype
SBDS	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
SDHA	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Paragangliomas 5, 614165 (3), Autosomal dominant
SEC61A1	609213	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056 (3), Autosomal dominant
SEMA3E	608166	?CHARGE syndrome, 214800 (3), Autosomal dominant
SERPING1	606860	Angioedema, hereditary, 1 and 2, 106100 (3), Autosomal recessive, Autosomal dominant; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant
SGPL1	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive
SH2D1A	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
SH3BP2	602104	Cherubism, 118400 (3), Autosomal dominant
SH3KBP1	300374	?Immunodeficiency 61, 300310 (3), X-linked recessive
SKIV2L	600478	Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive
SLC11A1	600266	{Mycobacterium tuberculosis, susceptibility to infection by}, 607948 (3); {Buruli ulcer, susceptibility to}, 610446 (3)
SLC29A3	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
SLC35C1	605881	Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive
SLC37A4	602671	Glycogen storage disease Ib, 232220 (3), Autosomal recessive; Glycogen storage disease Ic, 232240 (3), Autosomal recessive
SLC39A7	601416	No OMIM phenotype

SLC46A1	611672	Folate malabsorption, hereditary, 229050 (3), Autosomal recessive
SLC7A7	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
SLC9A3	182307	Diarrhea 8, secretory sodium, congenital, 616868 (3), Autosomal recessive
SMARCAL1	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
SMARCD2	601736	Specific granule deficiency 2, 617475 (3), Autosomal recessive
SNX10	614780	Osteopetrosis, autosomal recessive 8, 615085 (3), Autosomal recessive
SOCS1	603597	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375 (3), Autosomal dominant
SOCS4	616337	No OMIM phenotype
SP110	604457	{Mycobacterium tuberculosis, susceptibility to}, 607948 (3); Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive
SPI1	165170	No OMIM phenotype
SPINK5	605010	Netherton syndrome, 256500 (3), Autosomal recessive
SPPL2A	608238	No OMIM phenotype
SRP54	604857	Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant
SRP72	602122	Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant
STAT1	600555	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 (3), Autosomal dominant; Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 (3), Autosomal dominant; Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 (3), Autosomal recessive
STAT2	600556	Pseudo-TORCH syndrome 3, 618886 (3), Autosomal recessive; Immunodeficiency 44, 616636 (3), Autosomal recessive
STAT3	102582	Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant
STAT4	600558	{Systemic lupus erythematosus, susceptibility to, 11}, 612253 (3)
STAT5B	604260	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 (3), Autosomal recessive; Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 (3), Autosomal dominant; Leukemia, acute promyelocytic, somatic, 102578 (3)
STAT6	601512	No OMIM phenotype
STIM1	605921	Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive
STING1 (TMEM173)	612374	STING-associated vasculopathy, infantile-onset, 615934 (3), Autosomal dominant
STK4	604965	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 (3)
STN1	613128	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive

STX11	605014	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive
STXBP2	601717	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101 (3)
STXBP3	No OMIM gene	No OMIM phenotype
SYK	600085	Immunodeficiency 82 with systemic inflammation, 619381 (3), Autosomal dominant
TAFAZZIN (TAZ)	300394	Barth syndrome, 302060 (3), X-linked recessive
TAP1	170260	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
TAP2	170261	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 (3), Autosomal recessive
TAPBP	601962	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
TBK1	604834	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 (3), Autosomal dominant
TBX1	602054	Tetralogy of Fallot, 187500 (3), Autosomal dominant; DiGeorge syndrome, 188400 (3), Autosomal dominant; Conotruncal anomaly face syndrome, 217095 (3); Velocardiofacial syndrome, 192430 (3), Autosomal dominant
TBX21	604895	Asthma and nasal polyps, 208550 (3), Autosomal recessive; {Asthma, aspirin-induced, susceptibility to}, 208550 (3), Autosomal recessive
TCF3	147141	Agammaglobulinemia 8, autosomal dominant, 616941 (3), Autosomal dominant
TCIRG1	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive
TCN2	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
TERC	602322	{Pulmonary fibrosis, idiopathic, susceptibility to}, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant; {Aplastic anemia}, 614743 (3), Autosomal dominant
TERT	187270	{Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 9}, 615134 (3); {Dyskeratosis congenita, autosomal dominant 2}, 613989 (3), Autosomal recessive, Autosomal dominant; {Dyskeratosis congenita, autosomal recessive 4}, 613989 (3), Autosomal recessive, Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Somatic mutation, Autosomal dominant
TET2	612839	Myelodysplastic syndrome, somatic, 614286 (3); Immunodeficiency 75, 619126 (3), Autosomal recessive
TFRC	190010	Immunodeficiency 46, 616740 (3), Autosomal recessive
TGFB1	190180	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 (3), Autosomal recessive; Camurati-Engelmann disease, 131300 (3), Autosomal dominant; {Cystic fibrosis lung disease, modifier of}, 219700 (3), Autosomal recessive

TGFBR1	190181	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant
TGFBR2	190182	Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3)
THBD	188040	Thrombophilia due to thrombomodulin defect, 614486 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant
TICAM1	607601	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850 (3), Autosomal recessive, Autosomal dominant
TINF2	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TIRAP	606252	{Malaria, protection against}, 611162 (3); {Tuberculosis, protection against}, 607948 (3); {Bacteremia, protection against}, 614382 (3)
TLR3	603029	{HIV1 infection, resistance to}, 609423 (3); {Immunodeficiency 83, susceptibility to viral infections}, 613002 (3), Autosomal recessive, Autosomal dominant
TLR4	603030/611488	-/Macular degeneration, age-related, 10, 611488 (2)
TLR7	300365	Immunodeficiency 74, COVID19-related, X-linked, 301051 (3), X-linked recessive
TMC6	605828	Epidermodysplasia verruciformis, 226400 (3), Autosomal recessive
TMC8	605829	Epidermodysplasia verruciformis 2, 618231 (3), Autosomal recessive
TNFAIP3	191163	Autoinflammatory syndrome, familial, Behcet-like, 616744 (3), Autosomal dominant
TNFRSF11A	603499	Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive; {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant; Osteolysis, familial expansile, 174810 (3), Autosomal dominant
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
TNFRSF1A	191190	{Multiple sclerosis, susceptibility to, 5}, 614810 (3); Periodic fever, familial, 142680 (3), Autosomal dominant
TNFRSF4	600315	?Immunodeficiency 16, 615593 (3), Autosomal recessive
TNFRSF9	602250	No OMIM phenotype
TNFSF11	602642	Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive
TNFSF12	602695	No OMIM phenotype
TNFSF13	604472	No OMIM phenotype
TOP2B	126431	No OMIM phenotype
TPP2	190470	Immunodeficiency 78 with autoimmunity and developmental delay, 619220 (3), Autosomal recessive

TRAC	186880	Immunodeficiency 7, TCR-alpha/beta deficient, 615387 (3), Autosomal recessive
TRAF3	601896	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849 (3)
TRAF3IP2	607043	?Candidiasis, familial, 8, 615527 (3), Autosomal recessive; {Psoriasis susceptibility 13}, 614070 (3)
TREX1	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
TRIM22	606559	No OMIM phenotype
TRNT1	612907	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive; Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive
TTC37	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
TTC7A	609332	Gastrointestinal defects and immunodeficiency syndrome, 243150 (3), Autosomal recessive
TYK2	176941	Immunodeficiency 35, 611521 (3), Autosomal recessive
UBA1	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive; VEXAS syndrome, somatic, 301054 (3)
UNC119	604011	?Immunodeficiency 13, 615518 (3), Autosomal dominant; ?Cone-rod dystrophy (3)
UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3), Autosomal recessive
UNC93B1	608204	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551 (3)
UNG	191525	Immunodeficiency with hyper IgM, type 5, 608106 (3), Autosomal recessive
USB1	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive
USP18	607057	Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive
VAV1	164875	No OMIM phenotype
VPS13B	607817	Cohen syndrome, 216550 (3), Autosomal recessive
VPS45	610035	Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive
WAS	300392	Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive; Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive; Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive
WDR1	604734	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550 (3), Autosomal recessive
WIPF1	602357	Wiskott-Aldrich syndrome 2, 614493 (3), Autosomal recessive

WRAP53	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
XBP1	194355	{Major affective disorder-7, susceptibility to}, 612371 (3)
XIAP	300079	Lymphoproliferative syndrome, X-linked, 2, 300635 (3), X-linked recessive
ZAP70	176947	Immunodeficiency 48, 269840 (3), Autosomal recessive; Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive
ZBTB24	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
ZNF341	618269	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282 (3), Autosomal recessive
ZNFX1	618931	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: July 26, 2021

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.