PID_diagnostic panel

versie V5 (500 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 dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal recessive 7, 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	Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive; Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant
AGA	613228	Aspartylglucosaminuria, 208400 (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
AKT1	164730	Breast cancer, somatic, 114480 (3); Colorectal cancer, somatic, 114500 (3); Cowden syndrome 6, 615109 (3); Ovarian cancer, somatic, 167000 (3); Proteus syndrome, somatic, 176920 (3); {Schizophrenia, susceptibility to}, 181500 (2), Autosomal dominant
ALG13	300776	?Congenital disorder of glycosylation, type Is, 300884 (3), X-linked dominant; Epileptic encephalopathy, early infantile, 36, 300884 (3), X-linked dominant
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

AP4E1	607244	Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive; Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant
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	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718 (3), Autosomal recessive
ATM	607585	Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; Lymphoma, B-cell non-Hodgkin, somatic (3); Lymphoma, mantle cell, somatic (3); T-cell prolymphocytic leukemia, 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
ATP6V0A2	611716	Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive; Wrinkly skin syndrome, 278250 (3), Autosomal 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	?Immunodeficiency 37, 616098 (3), Autosomal recessive; Lymphoma, MALT, somatic, 137245 (3); {Lymphoma, follicular, somatic}, 605027 (3); {Male germ cell tumor, somatic}, 273300 (3); {Mesothelioma, somatic}, 156240 (3); {Sezary syndrome, somatic} (3)
BCL11B	606558	Immunodeficiency 49, 617237 (3), Autosomal dominant; Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (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
BTLA	607925	No OMIM phenotype
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)
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); [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)
CA2	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (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
CARD14	607211	Pityriasis rubra pilaris, 173200 (3), Autosomal dominant; Psoriasis 2, 602723 (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	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 (3), Autosomal recessive; {Breast cancer, protection against}, 114480 (3), Autosomal dominant, Somatic mutation; Hepatocellular carcinoma, somatic, 114550 (3); {Lung cancer, protection against}, 211980 (3), Autosomal dominant, Somatic mutation
CAVIN1	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
CBL	165360	?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation; Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant
CCBE1	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal 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	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
CD80	112203	No OMIM phenotype
CD81	186845	Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive
CD86	601020	No OMIM phenotype
CD8A	186910	CD8 deficiency, familial, 608957 (3), Autosomal recessive
CDCA7	609937	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910 (3), Autosomal recessive
CDKN2A	600160	Melanoma and neural system tumor syndrome, 155755 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 2}, 155601 (3), Autosomal dominant; Orolaryngeal cancer, multiple (3); Pancreatic cancer/melanoma syndrome, 606719 (3), Autosomal dominant
CDKN2B	600431	No OMIM phenotype
CEBPE	600749	Specific granule deficiency, 245480 (3), Autosomal recessive
CFB	138470	?Complement factor B deficiency, 615561 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3)
CFD	134350	Complement factor D deficiency, 613912 (3), Autosomal recessive

CFH	134370	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; {Macular degeneration, age-related, 4}, 610698 (3)
CFHR1	134371	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant
CFHR2	600889	No OMIM phenotype
CFHR3	605336	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant
CFHR4	605337	No OMIM phenotype
CFHR5	608593	Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant
CFI	217030	Complement factor I deficiency, 610984 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant
CFP	300383	Properdin deficiency, X-linked, 312060 (3), X-linked recessive
CFTR	602421	{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; Cystic fibrosis, 219700 (3), Autosomal recessive; {Hypertrypsinemia, neonatal} (3); {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; Sweat chloride elevation without CF (3)
CHD7	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
CIB1	602293	Epidermodysplasia verruciformis 3, 618267 (3), Autosomal recessive
CIITA	600005	Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive; {Rheumatoid arthritis, susceptibility to}, 180300 (3)
CLCN7	602727	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive
CLEC4D	609964	No OMIM phenotype
CLEC7A	606264	{Aspergillosis, susceptibility to}, 614079 (3); Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive
CLPB	616254	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 (3), Autosomal recessive
CNBP	116955	Myotonic dystrophy 2, 602668 (3), Autosomal dominant
COLEC11	612502	3MC syndrome 2, 265050 (3), Autosomal recessive

COPA	601924	{Autoimmune interstitial lung, joint, and kidney disease}, 616414 (3), Autosomal dominant
CORO1A	605000	Immunodeficiency 8, 615401 (3), Autosomal recessive
CPT2	600650	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; {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant
CR2	120650	Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive; {Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3)
CREBBP	600140	Menke-Hennekam syndrome 1, 618332 (3); Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CSF2RA	425000	No OMIM phenotype
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; {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
CTPS1	123860	Immunodeficiency 24, 615897 (3), Autosomal recessive
CTSC	602365	Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive; Periodontitis 1, juvenile, 170650 (3), Autosomal recessive
CXCL12	600835	{AIDS, resistance to}, 609423 (3)
CXCR4	162643	Myelokathexis, isolated (3); WHIM syndrome, 193670 (3), Autosomal dominant
CYBA	608508	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690 (3), Autosomal recessive
CYBB	300481	Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive; Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive
CYBC1 (C17orf62)	No OMIM gene	No OMIM phenotype
DBR1	607024	No OMIM phenotype
DCLRE1B	609683	No OMIM phenotype
DCLRE1C	605988	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, Athabascan type, 602450 (3), Autosomal recessive

DDX58	609631	Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant
DEF6	610094	No OMIM phenotype
DGCR6	601279	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
ERCC2	126340	?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
ERCC3	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
ERCC6L2	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
EXTL3	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
EZR	123900	No OMIM phenotype
F12	610619	Angioedema, hereditary, type III, 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	Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant; Squamous cell carcinoma, burn scar-related, somatic (3)
FASLG	134638	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation
FASN	600212	No OMIM phenotype
FAT4	612411	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive; Van Maldergem syndrome 2, 615546 (3), Autosomal recessive
FCGR1A	146760	[IgG receptor I, phagocytic, familial deficiency of] (3)
FCGR2A	146790	{Lupus nephritis, susceptibility to}, 152700 (3), Autosomal dominant; {Malaria, severe, susceptibility to}, 611162 (3); {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700 (3), Autosomal recessive
FCGR2B	604590	{Malaria, resistance to}, 611162 (3); {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
FCGR3A	146740	Immunodeficiency 20, 615707 (3), Autosomal recessive
FCGR3B	610665	Neutropenia, alloimmune neonatal (3)
FCGRT	601437	No OMIM phenotype
FCHO1	613437	No OMIM phenotype
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
FOXN1	600838	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
G6PC	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
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	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3), X-linked recessive; Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 (3); Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3), X-linked recessive; Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3), X-linked recessive

GATA2	137295	Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Leukemia, acute myeloid, susceptibility to}, 601626 (3), Autosomal dominant, Somatic mutation; {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
GINS1	610608	Immunodeficiency 55, 617827 (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)
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
IFNGR1	107470	{H. pylori infection, susceptibility to}, 600263 (3); {Hepatitis B virus infection, susceptibility to}, 610424 (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)
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 15A, 618204 (3), Autosomal dominant; Immunodeficiency 15B, 615592 (3), Autosomal recessive
IKBKG	300248	Ectodermal dysplasia and immunodeficiency 1, 300291 (3); Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (3); Immunodeficiency 33, 300636 (3), X-linked recessive; Immunodeficiency, isolated, 300584 (3); Incontinentia pigmenti, 308300 (3), X-linked dominant; Invasive pneumococcal disease, recurrent isolated, 2, 300640 (3)

IKZF1	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
IL10	124092	{Graft-versus-host disease, protection against}, 614395 (3); {HIV-1, susceptibility to}, 609423 (3); {Rheumatoid arthritis, progression of}, 180300 (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)
IL1RL1	601203	No OMIM phenotype
IL1RN	147679	{Gastric cancer risk after H. pylori infection}, 137215 (3), Autosomal dominant; Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive; {Microvascular complications of diabetes 4}, 612628 (3)
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	{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3); Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive
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); [Interleukin-6 receptor, soluble, serum level of, QTL], 614689 (3)
IL6ST	600694	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523 (3)

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
INPP5D	601582	No OMIM phenotype
INSR	147670	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (3); Hyperinsulinemic hypoglycemia, familial, 5, 609968 (3), Autosomal dominant; Leprechaunism, 246200 (3), Autosomal recessive; Rabson-Mendenhall syndrome, 262190 (3), Autosomal recessive
IRAK1	300283	No OMIM phenotype
IRAK2	603304	No OMIM phenotype
IRAK4	606883	IRAK4 deficiency, 607676 (3); Invasive pneumococcal disease, recurrent isolated, 1, 610799 (3)
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	No OMIM phenotype
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
JAGN1	616012	Neutropenia, severe congenital, 6, autosomal recessive, 616022 (3), Autosomal recessive
JAK1	147795	No OMIM phenotype
JAK2	147796	{Budd-Chiari syndrome, somatic}, 600880 (3); Erythrocytosis, somatic, 133100 (3); Leukemia, acute myeloid, somatic, 601626 (3); Myelofibrosis, somatic, 254450 (3); Polycythemia vera, somatic, 263300 (3); Thrombocythemia 3, 614521 (3), Autosomal dominant, Somatic mutation
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	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 (2), Autosomal dominant; Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant

KMT2D	602113	Kabuki syndrome 1, 147920 (3), Autosomal dominant
KRAS	190070	Arteriovenous malformation of the brain, somatic, 108010 (3); Bladder cancer, somatic, 109800 (3); Breast cancer, somatic, 114480 (3); Cardiofaciocutaneous syndrome 2, 615278 (3); Gastric cancer, somatic, 137215 (3); Leukemia, acute myeloid, 601626 (3), Autosomal dominant, Somatic mutation; Lung cancer, somatic, 211980 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; Oculoectodermal syndrome, somatic, 600268 (3); Pancreatic carcinoma, somatic, 260350 (3); RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3)
LACC1	613409	No OMIM phenotype
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
LIG1	126391	No OMIM phenotype
LIG4	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
LIPA	613497	Cholesteryl ester storage disease, 278000 (3), Autosomal recessive; Wolman 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
LTBP3	602090	Dental anomalies and short stature, 601216 (3), Autosomal recessive; Geleophysic dysplasia 3, 617809 (3), Autosomal dominant
LYST	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
MAGT1	300715	Congenital disorder of glycosylation, type Icc, 301031 (3), X-linked recessive; Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3)
MAL2	No OMIM gene	No OMIM phenotype
MALT1	604860	Immunodeficiency 12, 615468 (3), Autosomal recessive
MAN2B1	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MANBA	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
MAP3K14	604655	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
MC2R	607397	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3), Autosomal recessive
MCM4	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive

MEFV	608107	Familial Mediterranean fever, AD, 134610 (3), Autosomal dominant; Familial Mediterranean fever, AR, 249100 (3), Autosomal recessive
MLPH	606526	Griscelli syndrome, type 3, 609227 (3), Autosomal recessive
MOGS	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
MPO	606989	{Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; {Lung cancer, protection against, in smokers} (3); Myeloperoxidase deficiency, 254600 (3), Autosomal recessive
MRE11	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
MRTFA (MKL1)	606078	Megakaryoblastic leukemia, acute (3)
MS4A1	112210	Immunodeficiency, common variable, 5, 613495 (3), Autosomal recessive
MSH6	600678	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 (3), Autosomal dominant; {Endometrial cancer, familial}, 608089 (3), Autosomal dominant, Somatic mutation; Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive
MSN	309845	Immunodeficiency 50, 300988 (3), X-linked recessive
MST1	142408	No OMIM phenotype
MTHFD1	172460	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MVK	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Mevalonic aciduria, 610377 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant
MYD88	602170	Macroglobulinemia, Waldenstrom, somatic, 153600 (3); Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 (3)
MYO5A	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive
MYSM1	612176	Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive
NBAS	608025	Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive; Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive
NBN	602667	Aplastic anemia, 609135 (3); Leukemia, acute lymphoblastic, 613065 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NCF1	608512	Chronic granulomatous disease due to deficiency of NCF-1, 233700 (3), Autosomal recessive
NCF2	608515	Chronic granulomatous disease due to deficiency of NCF-2, 233710 (3), Autosomal recessive
NCF4	601488	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960 (3), Autosomal recessive
NCSTN	605254	Acne inversa, familial, 1, 142690 (3), Autosomal dominant
NEIL2	608933	No OMIM phenotype
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	Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Ventricular septal defect 3, 614432 (3), Autosomal dominant
NLRC4	606831	Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant; ?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant
NLRP1	606636	Autoinflammation with arthritis and dyskeratosis, 617388 (3), Autosomal recessive, Autosomal dominant; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant; {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 (3)
NLRP12	609648	Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant
NLRP3	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (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; {Inflammatory bowel disease 1, Crohn disease}, 266600 (3), Multifactorial; {Psoriatic arthritis, susceptibility to}, 607507 (2); {Yao syndrome}, 617321 (3), Multifactorial
NOP10	606471	Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive

NRAS	164790	Colorectal cancer, somatic, 114500 (3); Epidermal nevus, somatic, 162900 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Neurocutaneous melanosis, somatic, 249400 (3); Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3)
NSMCE3	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive
OAS1	164350	No OMIM phenotype
OFD1	300170	Joubert syndrome 10, 300804 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
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
PAX5	167414	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3)
PBX1	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant
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
PIGA	311770	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3)
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
PLEKHM1	611466	Osteopetrosis, autosomal dominant 3, 618107 (3), Autosomal dominant; ?Osteopetrosis, autosomal recessive 6, 611497 (3), Autosomal recessive

PLG	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
PMM2	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PMS2	600259	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3); Mismatch repair cancer syndrome, 276300 (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	{Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant; Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (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	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive; Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive
POLR3C	617454	No OMIM phenotype
POLR3F	617455	No OMIM phenotype
POMP	613386	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive; Proteasome-associated autoinflammatory syndrome 2, 618048 (3), Autosomal dominant
POT1	606478	{Glioma susceptibility 9}, 616568 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848 (3), Autosomal dominant
PRF1	170280	Aplastic anemia, 609135 (3); Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Lymphoma, non-Hodgkin, 605027 (3)
PRKCD	176977	Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive
PRKDC	600899	Immunodeficiency 26, with or without neurologic abnormalities, 615966 (3), Autosomal recessive
PRPS1	311850	Arts syndrome, 301835 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive

PSEN1	104311	?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; Pick disease, 172700 (3), Autosomal dominant
PSENEN	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant
PSMA3	176843	No OMIM phenotype
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	No OMIM phenotype
PSTPIP1	606347	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 (3), Autosomal dominant
PSTPIP2	616046	No OMIM phenotype
PTEN	601728	Cowden syndrome 1, 158350 (3), Autosomal dominant; {Glioma susceptibility 2}, 613028 (3); Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3)
PTPN11	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3); Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), Autosomal dominant
PTPN22	600716	{Diabetes, type 1, susceptibility to}, 222100 (3), Autosomal recessive; {Rheumatoid arthritis, susceptibility to}, 180300 (3); {Systemic lupus erythematosus susceptibility to}, 152700 (3), Autosomal dominant
PTPN6	176883	No OMIM phenotype
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
RAB27A	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
RAC2	602049	Neutrophil immunodeficiency syndrome, 608203 (3)
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
RANBP2	601181	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033 (3), Autosomal dominant
RASGRP1	603962	Immunodeficiency 64, 618534 (3)
RBCK1	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
RC3H1	609424	No OMIM phenotype
RECQL4	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive; Rothmund-Thomson syndrome, 268400 (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, 618108 (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
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
RNU4ATAC	601428	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive; Roifman syndrome, 616651 (3), Autosomal recessive
RORC	602943	Immunodeficiency 42, 616622 (3), Autosomal recessive
RPSA	150370	Asplenia, isolated congenital, 271400 (3), Autosomal dominant
RRAS2	600098	Ovarian carcinoma (3)
RSPH9	612648	Ciliary dyskinesia, primary, 12, 612650 (3)

RTEL1	608833	Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant
RTL1	611896	No OMIM phenotype
SAMD9	610456	MIRAGE syndrome, 617053 (3), Autosomal dominant; Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive
SAMD9L	611170	Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant
SAMHD1	606754	Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive; ?Chilblain lupus 2, 614415 (3), Autosomal dominant
SBDS	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
SDHA	600857	Cardiomyopathy, dilated, 1GG, 613642 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive; Paragangliomas 5, 614165 (3), Autosomal dominant
SEC61A1	609213	Hyperuricemic nephropathy, familial juvenile, 4, 617056 (3), Autosomal dominant
SEMA3E	608166	?CHARGE syndrome, 214800 (3), Autosomal dominant
SERAC1	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
SERPING1	606860	Angioedema, hereditary, types I and II, 106100 (3), Autosomal recessive, Autosomal dominant; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant
SH2B3	605093	Erythrocytosis, somatic, 133100 (3); Myelofibrosis, somatic, 254450 (3); Thrombocythemia, somatic, 187950 (3)
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	{Buruli ulcer, susceptibility to}, 610446 (3); {Mycobacterium tuberculosis, susceptibility to infection by}, 607948 (3)
SLC29A3	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
SLC35A1	605634	Congenital disorder of glycosylation, type IIf, 603585 (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
SLC39A4	607059	Acrodermatitis enteropathica, 201100 (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
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
SOCS4	616337	No OMIM phenotype
SP110	604457	Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive; {Mycobacterium tuberculosis, susceptibility to}, 607948 (3)
SPINK5	605010	Netherton syndrome, 256500 (3), Autosomal recessive
SPPL2A	608238	No OMIM phenotype
SRP54	604857	No OMIM phenotype
SRP72	602122	Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant
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
STAT2	600556	Immunodeficiency 44, 616636 (3), Autosomal recessive
STAT3	102582	Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant; Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant
STAT4	600558	{Systemic lupus erythematosus, susceptibility to, 11}, 612253 (3)
STAT5B	604260	Growth hormone insensitivity with immunodeficiency, 245590 (3); Leukemia, acute promyelocytic, somatic, 102578 (3)
STAT6	601512	No OMIM phenotype
STIM1	605921	Immunodeficiency 10, 612783 (3), Autosomal recessive; Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant
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, 613101 (3)
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
TAZ	300394	Barth syndrome, 302060 (3), X-linked 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	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
TCF3	147141	Agammaglobulinemia 8, autosomal dominant, 616941 (3), Autosomal dominant
TCF7L1	604652	No OMIM phenotype
TCIRG1	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive
TCN2	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
TERC	602322	{Aplastic anemia}, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant; {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743 (3), Autosomal dominant
TERT	187270	{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), Autosomal dominant, Somatic mutation; {Melanoma, cutaneous malignant, 9}, 615134 (3); {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 (3), Autosomal dominant
TFRC	190010	Immunodeficiency 46, 616740 (3), Autosomal recessive
TGFB1	190180	Camurati-Engelmann disease, 131300 (3), Autosomal dominant; {Cystic fibrosis lung disease, modifier of}, 219700 (3), Autosomal recessive; Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 (3), Autosomal recessive
TGFBR1	190181	Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant; {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant
TGFBR2	190182	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3); Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant
THBD	188040	{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant; Thrombophilia due to thrombomodulin defect, 614486 (3)
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	{Bacteremia, protection against}, 614382 (3); {Malaria, protection against}, 611162 (3); {Pneumococcal disease, invasive, protection against}, 610799 (3); {Tuberculosis, protection against}, 607948 (3)
TLR3	603029	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002 (3), Autosomal recessive, Autosomal dominant; {HIV1 infection, resistance to}, 609423 (3)
TLR4	603030	No OMIM phenotype
TMC6	605828	Epidermodysplasia verruciformis, 226400 (3), Autosomal recessive
TMC8	605829	Epidermodysplasia verruciformis 2, 618231 (3)
TNFAIP3	191163	Autoinflammatory syndrome, familial, Behcet-like, 616744 (3), Autosomal dominant
TNFRSF11A	603499	Osteolysis, familial expansile, 174810 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive; {Paget disease of bone 2, early-onset}, 602080 (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
TOP2B	126431	No OMIM phenotype
TP53	191170	{Adrenocortical carcinoma, pediatric}, 202300 (3), Autosomal dominant; {Basal cell carcinoma 7}, 614740 (3), Autosomal dominant; Bone marrow failure syndrome 5, 618165 (3), Autosomal dominant; Breast cancer, somatic, 114480 (3); {Choroid plexus papilloma}, 260500 (3), Autosomal dominant; {Colorectal cancer}, 114500 (3), Autosomal dominant, Somatic mutation; {Glioma susceptibility 1}, 137800 (3), Autosomal dominant, Somatic mutation; Hepatocellular carcinoma, somatic, 114550 (3); Li-Fraumeni syndrome, 151623 (3), Autosomal dominant; Nasopharyngeal carcinoma, somatic, 607107 (3); {Osteosarcoma}, 259500 (3), Somatic mutation; Pancreatic cancer, somatic, 260350 (3)
TPP1	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
TPP2	190470	No OMIM phenotype
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	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant
TRIM21	109092	No OMIM phenotype
TRIM22	606559	No OMIM phenotype
TRNT1	612907	Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive; Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (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
UNC119	604011	?Cone-rod dystrophy (3); ?Immunodeficiency 13, 615518 (3), Autosomal dominant
UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3)
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
VAV2	600428	No OMIM phenotype
VIPAS39	613401	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
VPS13B	607817	Cohen syndrome, 216550 (3), Autosomal recessive
VPS33B	608552	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
VPS45	610035	Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive
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
WDR1	604734	No OMIM phenotype
WIPF1	602357	?Wiskott-Aldrich syndrome 2, 614493 (3)

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
XRCC4	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
ZAP70	176947	Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive; Immunodeficiency 48, 269840 (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

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: Sept 30, 2019

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.