

PID diagnostic panel		
versie	v7 (512 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ACD</i>	609377	?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal dominant, Autosomal recessive; ?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal dominant, Autosomal recessive
<i>ACP5</i>	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
<i>ACTB</i>	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant
<i>ADA</i>	608958	Adenosine deaminase deficiency, partial, 102700 (3), Somatic mosaicism, Autosomal recessive; Severe combined immunodeficiency due to ADA deficiency, 102700 (3), Somatic mosaicism, Autosomal recessive
<i>ADA2</i>	607575	Sneddon syndrome, 182410 (3), Autosomal recessive; Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 (3), Autosomal recessive
<i>ADAM17</i>	603639	?Inflammatory skin and bowel disease, neonatal, 1, 614328 (3), Autosomal recessive
<i>ADAR</i>	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
<i>AGR2</i>	606358	No OMIM phenotype
<i>AICDA</i>	605257	Immunodeficiency with hyper-IgM, type 2, 605258 (3), Autosomal recessive
<i>AIRE</i>	607358	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal dominant, Autosomal recessive
<i>AK2</i>	103020	Reticular dysgenesis, 267500 (3), Autosomal recessive
<i>ALPI</i>	171740	No OMIM phenotype
<i>ALPK1</i>	607347	ROSAH syndrome, 614979 (3), Autosomal dominant
<i>AP1S3</i>	615781	{Psoriasis 15, pustular, susceptibility to}, 616106 (3), Autosomal dominant
<i>AP3B1</i>	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
<i>AP3D1</i>	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
<i>APOL1</i>	603743	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 (3); {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 (3)
<i>ARHGEF1</i>	601855	?Immunodeficiency 62, 618459 (3), Autosomal recessive
<i>ARPC1B</i>	604223	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718 (3), Autosomal recessive

<i>ATAD3A</i>	612316	Harel-Yoon syndrome, 617183 (3), Autosomal dominant, Autosomal recessive; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
<i>ATG16L1</i>	610767	{Inflammatory bowel disease (Crohn disease) 10}, 611081 (3)
<i>ATG4A</i>	300663	No OMIM phenotype
<i>ATM</i>	607585	Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3)
<i>ATP2A2</i>	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
<i>ATP6AP1</i>	300197	Immunodeficiency 47, 300972 (3), X-linked recessive
<i>B2M</i>	109700	?Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Immunodeficiency 43, 241600 (3), Autosomal recessive
<i>BACH2</i>	605394	Immunodeficiency 60 and autoimmunity, 618394 (3), Autosomal dominant
<i>BCL10</i>	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)
<i>BCL11B</i>	606558	Immunodeficiency 49, severe combined, 617237 (3), Autosomal dominant; Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (3), Autosomal dominant
<i>BLK</i>	191305	Maturity-onset diabetes of the young, type 11, 613375 (3), Autosomal dominant
<i>BLM</i>	604610	Bloom syndrome, 210900 (3), Autosomal recessive
<i>BLNK</i>	604515	?Agammaglobulinemia 4, 613502 (3), Autosomal recessive
<i>BLOC1S3</i>	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
<i>BLOC1S6</i>	604310	?Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive
<i>BTK</i>	300300	Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive; Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 (3), X-linked recessive
<i>C1QA</i>	120550	C1q deficiency, 613652 (3), Autosomal recessive
<i>C1QB</i>	120570	C1q deficiency, 613652 (3), Autosomal recessive
<i>C1QC</i>	120575	C1q deficiency, 613652 (3), Autosomal recessive
<i>C1R</i>	613785	Ehlers-Danlos syndrome, periodontal type, 1, 130080 (3), Autosomal dominant
<i>C1S</i>	120580	C1s deficiency, 613783 (3); Ehlers-Danlos syndrome, periodontal type, 2, 617174 (3), Autosomal dominant
<i>C2</i>	613927	C2 deficiency, 217000 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant

<i>C2orf69</i>	619219	Combined oxidative phosphorylation deficiency 53, 619423 (3), Autosomal recessive
<i>C3</i>	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)
<i>C4A</i>	120810	[Blood group, Rodgers], 614374 (3); C4a deficiency, 614380 (3), Autosomal recessive
<i>C4B</i>	120820	C4B deficiency, 614379 (3)
<i>C4BPA</i>	120830	No OMIM phenotype
<i>C5</i>	120900	C5 deficiency, 609536 (3), Autosomal recessive; [Eculizumab, poor response to], 615749 (3), Autosomal dominant
<i>C6</i>	217050	C6 deficiency, 612446 (3), Autosomal recessive; Combined C6/C7 deficiency (3)
<i>C7</i>	217070	C7 deficiency, 610102 (3)
<i>C8A</i>	120950	C8 deficiency, type I, 613790 (3), Autosomal recessive
<i>C8B</i>	120960	C8 deficiency, type II, 613789 (3), Autosomal recessive
<i>C8G</i>	120930	No OMIM phenotype
<i>C9</i>	120940	C9 deficiency, 613825 (3); {Macular degeneration, age-related, 15, susceptibility to}, 615591 (3), Autosomal dominant
<i>CARD11</i>	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
<i>CARD14</i>	607211	Psoriasis 2, 602723 (3), Autosomal dominant; Pityriasis rubra pilaris, 173200 (3), Autosomal dominant
<i>CARD9</i>	607212	Immunodeficiency 103, susceptibility to fungal infection, 212050 (3), Autosomal recessive
<i>CARMIL2</i>	610859	Immunodeficiency 58, 618131 (3), Autosomal recessive
<i>CASP10</i>	601762	Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
<i>CASP8</i>	601763	{Breast cancer, protection against}, 114480 (3), Autosomal dominant, Somatic mutation; Hepatocellular carcinoma, somatic, 114550 (3); ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 (3), Autosomal recessive; {Lung cancer, protection against}, 211980 (3), Autosomal dominant, Somatic mutation
<i>CBL</i>	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation
<i>CCBE1</i>	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
<i>CCDC28B</i>	610162	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Digenic recessive, Autosomal recessive

<i>CD19</i>	107265	Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive
<i>CD247</i>	186780	?Immunodeficiency 25, 610163 (3), Autosomal recessive
<i>CD27</i>	186711	Lymphoproliferative syndrome 2, 615122 (3), Autosomal recessive
<i>CD28</i>	186760	No OMIM phenotype
<i>CD3D</i>	186790	Immunodeficiency 19, severe combined, 615617 (3), Autosomal recessive
<i>CD3E</i>	186830	Immunodeficiency 18, 615615 (3), Autosomal recessive; Immunodeficiency 18, SCID variant, 615615 (3), Autosomal recessive
<i>CD3G</i>	186740	Immunodeficiency 17, CD3 gamma deficient, 615607 (3), Autosomal recessive
<i>CD4</i>	186940	Immunodeficiency 79, 619238 (3), Autosomal recessive; OKT4 epitope deficiency, 613949 (3)
<i>CD40</i>	109535	Immunodeficiency with hyper-IgM, type 3, 606843 (3), Autosomal recessive
<i>CD40LG</i>	300386	Immunodeficiency, X-linked, with hyper-IgM, 308230 (3), X-linked recessive
<i>CD46</i>	120920	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 (3), Autosomal dominant, Autosomal recessive
<i>CD55</i>	125240	[Blood group Cromer], 613793 (3), Autosomal recessive; Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 (3), Autosomal recessive
<i>CD59</i>	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive
<i>CD70</i>	602840	Lymphoproliferative syndrome 3, 618261 (3), Autosomal recessive
<i>CD79A</i>	112205	Agammaglobulinemia 3, 613501 (3), Autosomal recessive
<i>CD79B</i>	147245	Agammaglobulinemia 6, 612692 (3), Autosomal recessive
<i>CD81</i>	186845	Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive
<i>CD8A</i>	186910	CD8 deficiency, familial, 608957 (3), Autosomal recessive
<i>CDC42</i>	116952	Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant
<i>CDCA7</i>	609937	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910 (3), Autosomal recessive
<i>CDSN</i>	602593	Hypotrichosis 2, 146520 (3), Autosomal dominant; Peeling skin syndrome 1, 270300 (3), Autosomal recessive
<i>CEBPE</i>	600749	Specific granule deficiency, 245480 (3), Autosomal recessive
<i>CFB</i>	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
<i>CFD</i>	134350	Complement factor D deficiency, 613912 (3), Autosomal recessive

<i>CFH</i>	134370	{Macular degeneration, age-related, 4}, 610698 (3); Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal dominant, Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal dominant, Autosomal recessive
<i>CFHR1</i>	134371	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal dominant, Autosomal recessive
<i>CFHR2</i>	600889	No OMIM phenotype
<i>CFHR3</i>	605336	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal dominant, Autosomal recessive
<i>CFHR4</i>	605337	No OMIM phenotype
<i>CFHR5</i>	608593	Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant
<i>CFI</i>	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
<i>CFP</i>	300383	Properdin deficiency, X-linked, 312060 (3), X-linked recessive
<i>CFTR</i>	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)
<i>CHD7</i>	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
<i>CHUK</i>	600664	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 (3), Autosomal recessive; ?Cocoon syndrome, 613630 (3), Autosomal recessive
<i>CIB1</i>	602293	Epidermodysplasia verruciformis 3, 618267 (3), Autosomal recessive
<i>CIITA</i>	600005	{Rheumatoid arthritis, susceptibility to}, 180300 (3); Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive
<i>CLCN7</i>	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
<i>CLEC7A</i>	606264	Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive; {Aspergillosis, susceptibility to}, 614079 (3)

<i>CLPB</i>	616254	Neutropenia, severe congenital, 9, autosomal dominant, 619813 (3), Autosomal dominant; 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 (3), Autosomal recessive; 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 (3), Autosomal dominant
<i>COL7A1</i>	120120	EBD, localisata variant (3); Epidermolysis bullosa, pretibial, 131850 (3), Autosomal dominant, Autosomal recessive; Transient bullous of the newborn, 131705 (3), Autosomal dominant, Autosomal recessive; EBD, Bart type, 132000 (3), Autosomal dominant; Epidermolysis bullosa dystrophica, AD, 131750 (3), Autosomal dominant; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal dominant, Autosomal recessive; EBD inversa, 226600 (3), Autosomal recessive; Epidermolysis bullosa dystrophica, AR, 226600 (3), Autosomal recessive; Toenail dystrophy, isolated, 607523 (3), Autosomal dominant
<i>COPA</i>	601924	{Autoimmune interstitial lung, joint, and kidney disease}, 616414 (3), Autosomal dominant
<i>COPG1</i>	615525	No OMIM phenotype
<i>CORO1A</i>	605000	Immunodeficiency 8, 615401 (3), Autosomal recessive
<i>CPT2</i>	600650	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal dominant, Autosomal recessive; 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 dominant, Autosomal recessive
<i>CR2</i>	120650	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3); Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive
<i>CRACR2A</i>	614178	No OMIM phenotype
<i>CREBBP</i>	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
<i>CSF2RA</i>	306250	Surfactant metabolism dysfunction, pulmonary, 4, 300770 (3)
<i>CSF2RB</i>	138981	Surfactant metabolism dysfunction, pulmonary, 5, 614370 (3), Autosomal recessive
<i>CSF3R</i>	138971	Neutropenia, severe congenital, 7, autosomal recessive, 617014 (3), Autosomal recessive; ?Neutrophilia, hereditary, 162830 (3), Autosomal dominant
<i>CTC1</i>	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
<i>CTLA4</i>	123890	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 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

<i>CTNBL1</i>	611537	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846 (3), Autosomal recessive
<i>CTPS1</i>	123860	Immunodeficiency 24, 615897 (3), Autosomal recessive
<i>CTSC</i>	602365	Periodontitis 1, juvenile, 170650 (3), Autosomal recessive; Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive
<i>CXCR2</i>	146928	?WHIM syndrome 2, 619407 (3), Autosomal recessive
<i>CXCR4</i>	162643	WHIM syndrome 1, 193670 (3), Autosomal dominant; Myelokathexis, isolated, 193670 (3), Autosomal dominant
<i>CYBA</i>	608508	Chronic granulomatous disease 4, autosomal recessive, 233690 (3), Autosomal recessive
<i>CYBB</i>	300481	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive; Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive
<i>CYBC1</i>	618334	Chronic granulomatous disease 5, autosomal recessive, 618935 (3), Autosomal recessive
<i>DBR1</i>	607024	{Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441 (3), Autosomal recessive
<i>DCLRE1B</i>	609683	No OMIM phenotype
<i>DCLRE1C</i>	605988	Severe combined immunodeficiency, Athabaskan type, 602450 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive
<i>DEF6</i>	610094	Immunodeficiency 87 and autoimmunity, 619573 (3), Autosomal recessive
<i>DGKE</i>	601440	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (3), Autosomal recessive
<i>DHFR</i>	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
<i>DIAPH1</i>	602121	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive
<i>DKC1</i>	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
<i>DNAJC21</i>	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
<i>DNASE1</i>	125505	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
<i>DNASE1L3</i>	602244	Systemic lupus erythematosus 16, 614420 (3), Autosomal recessive
<i>DNASE2</i>	126350	Autoinflammatory-pancytopenia syndrome, 619858 (3), Autosomal recessive
<i>DNMT3B</i>	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive; Facioscapulohumeral muscular dystrophy 4, digenic, 619478 (3), Digenic dominant
<i>DOCK2</i>	603122	Immunodeficiency 40, 616433 (3), Autosomal recessive
<i>DOCK8</i>	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive

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<i>DOK3</i>	611435	No OMIM phenotype
<i>DPP9</i>	608258	No OMIM phenotype
<i>DSG1</i>	125670	Keratosis palmoplantaris striata I, AD, 148700 (3), Autosomal dominant; Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3), Autosomal recessive
<i>DSP</i>	125647	Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Skin fragility-woolly hair syndrome, 607655 (3), Autosomal recessive; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Keratosis palmoplantaris striata II, 612908 (3), Autosomal dominant; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive
<i>DTNBP1</i>	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
<i>EFL1</i>	617538	Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive
<i>ELANE</i>	130130	Neutropenia, cyclic, 162800 (3), Autosomal dominant; Neutropenia, severe congenital 1, autosomal dominant, 202700 (3), Autosomal dominant
<i>ELF4</i>	300775	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074 (3), X-linked recessive
<i>EPG5</i>	615068	Vici syndrome, 242840 (3), Autosomal recessive
<i>ERBIN</i>	606944	No OMIM phenotype
<i>ERCC6L2</i>	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
<i>EXTL3</i>	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
<i>F12</i>	610619	Angioedema, hereditary, 3, 610618 (3), Autosomal dominant; Factor XII deficiency, 234000 (3), Autosomal recessive
<i>FAAP24</i>	610884	No OMIM phenotype
<i>FADD</i>	602457	Immunodeficiency 90 with encephalopathy, functional hyposplenism, and hepatic dysfunction, 613759 (3), Autosomal recessive
<i>FAS</i>	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
<i>FASLG</i>	134638	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation
<i>FAT4</i>	612411	Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive
<i>FCGR2B</i>	604590	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3)
<i>FCGR3A</i>	146740	Immunodeficiency 20, 615707 (3), Autosomal recessive
<i>FCGR3B</i>	610665	No OMIM phenotype
<i>FCHO1</i>	613437	Immunodeficiency 76, 619164 (3), Autosomal recessive

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<i>FCN3</i>	604973	Immunodeficiency due to ficolin 3 deficiency, 613860 (3), Autosomal recessive
<i>FERMT1</i>	607900	Kindler syndrome, 173650 (3), Autosomal recessive
<i>FERMT3</i>	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive
<i>FGL2</i>	605351	No OMIM phenotype
<i>FLG</i>	135940	Ichthyosis vulgaris, 146700 (3), Autosomal dominant, Autosomal recessive; {Dermatitis, atopic, susceptibility to, 2}, 605803 (3)
<i>FNIP1</i>	610594	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705 (3), Autosomal recessive
<i>FOXN1</i>	600838	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 (3), Autosomal dominant; T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal recessive
<i>FOXP3</i>	300292	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive
<i>FPR1</i>	136537	No OMIM phenotype
<i>G6PC3</i>	611045	Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive
<i>G6PD</i>	305900	Hemolytic anemia, G6PD deficient (favism), 300908 (3), X-linked dominant; {Resistance to malaria due to G6PD deficiency}, 611162 (3)
<i>GATA1</i>	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; Hemolytic anemia due to elevated adenosine deaminase, 301083 (3), X-linked recessive
<i>GATA2</i>	137295	{Leukemia, acute myeloid, susceptibility to}, 601626 (3), Autosomal dominant, Somatic mutation; Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3)
<i>GFI1</i>	600871	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 (3), Autosomal dominant; Neutropenia, severe congenital 2, autosomal dominant, 613107 (3), Autosomal dominant
<i>GIMAP5</i>	608086	Portal hypertension, noncirrhotic, 2, 619463 (3), Autosomal recessive
<i>GINS1</i>	610608	Immunodeficiency 55, 617827 (3), Autosomal recessive
<i>GTF3A</i>	600860	No OMIM phenotype
<i>GUCY2C</i>	601330	Diarrhea 6, 614616 (3), Autosomal dominant; Meconium ileus, 614665 (3), Autosomal recessive
<i>HAVCR2</i>	606652	T-cell lymphoma, subcutaneous panniculitis-like, 618398 (3), Autosomal recessive

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<i>HAX1</i>	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
<i>HCK</i>	142370	No OMIM phenotype
<i>HELLS</i>	603946	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911 (3), Autosomal recessive
<i>HMOX1</i>	141250	Heme oxygenase-1 deficiency, 614034 (3), Autosomal recessive; {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (3)
<i>HPS1</i>	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
<i>HPS4</i>	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
<i>HPS6</i>	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
<i>HTRA2</i>	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
<i>HYOU1</i>	601746	?Immunodeficiency 59 and hypoglycemia, 233600 (3), Autosomal recessive
<i>ICOS</i>	604558	Immunodeficiency, common variable, 1, 607594 (3), Autosomal recessive
<i>ICOSLG</i>	605717	No OMIM phenotype
<i>IFIH1</i>	606951	Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi- Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton- Merten syndrome 1, 182250 (3), Autosomal dominant
<i>IFNAR1</i>	107450	Immunodeficiency 106, susceptibility to viral infections, 619935 (3), Autosomal recessive
<i>IFNAR2</i>	602376	{Hepatitis B virus, susceptibility to}, 610424 (3); Immunodeficiency 45, 616669 (3), Autosomal recessive
<i>IFNG</i>	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)
<i>IFNGR1</i>	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)
<i>IFNGR2</i>	147569	Immunodeficiency 28, mycobacteriosis, 614889 (3), Autosomal recessive
<i>IGHM</i>	147020	Agammaglobulinemia 1, 601495 (3), Autosomal recessive
<i>IGKC</i>	147200	Kappa light chain deficiency, 614102 (3), Autosomal recessive
<i>IGLL1</i>	146770	Agammaglobulinemia 2, 613500 (3), Autosomal recessive
<i>IKBKB</i>	603258	Immunodeficiency 15B, 615592 (3), Autosomal recessive; Immunodeficiency 15A, 618204 (3), Autosomal dominant

<i>IKBKG</i>	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; Autoinflammatory disease, systemic, X-linked, 301081 (3), X-linked
<i>IKZF1</i>	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
<i>IKZF2</i>	606234	No OMIM phenotype
<i>IKZF3</i>	606221	?Immunodeficiency 84, 619437 (3)
<i>IL10</i>	124092	{Rheumatoid arthritis, progression of}, 180300 (3); {Graft-versus-host disease, protection against}, 614395 (3); {HIV-1, susceptibility to}, 609423 (3)
<i>IL10RA</i>	146933	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148 (3), Autosomal recessive
<i>IL10RB</i>	123889	{Hepatitis B virus, susceptibility to}, 610424 (3); Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 (3), Autosomal recessive
<i>IL12B</i>	161561	Immunodeficiency 29, mycobacteriosis, 614890 (3), Autosomal recessive
<i>IL12RB1</i>	601604	Immunodeficiency 30, 614891 (3), Autosomal recessive
<i>IL12RB2</i>	601642	No OMIM phenotype
<i>IL15RA</i>	601070	No OMIM phenotype
<i>IL17F</i>	606496	?Candidiasis, familial, 6, autosomal dominant, 613956 (3)
<i>IL17RA</i>	605461	Immunodeficiency 51, 613953 (3), Autosomal recessive
<i>IL17RC</i>	610925	Candidiasis, familial, 9, 616445 (3), Autosomal recessive
<i>IL18</i>	600953	No OMIM phenotype
<i>IL18BP</i>	604113	{?Hepatitis, fulminant viral, susceptibility to}, 618549 (3), Autosomal recessive
<i>IL1RL1</i>	601203	No OMIM phenotype
<i>IL1RN</i>	147679	{Gastric cancer risk after H. pylori infection}, 613659 (3); {Microvascular complications of diabetes 4}, 612628 (3); Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive
<i>IL21</i>	605384	?Immunodeficiency, common variable, 11, 615767 (3), Autosomal recessive
<i>IL21R</i>	605383	[IgE, elevated level of], 147050 (3), Autosomal dominant; Immunodeficiency 56, 615207 (3), Autosomal recessive
<i>IL23R</i>	607562	{Inflammatory bowel disease 17, protection against}, 612261 (3); {Psoriasis, protection against}, 605606 (3)
<i>IL2RA</i>	147730	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive; {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3)
<i>IL2RB</i>	146710	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495 (3), Autosomal recessive
<i>IL2RG</i>	308380	Combined immunodeficiency, X-linked, moderate, 312863 (3), X-linked recessive; Severe combined immunodeficiency, X-linked, 300400 (3), X-linked recessive

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<i>IL36RN</i>	605507	Psoriasis 14, pustular, 614204 (3), Autosomal recessive
<i>IL6R</i>	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)
<i>IL6ST</i>	600694	Stuve-Wiedemann syndrome 2, 619751 (3), Autosomal recessive; Hyper-IgE recurrent infection syndrome 4A, autosomal dominant, 619752 (3), Autosomal dominant; ?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750 (3), Autosomal dominant; Hyper-IgE recurrent infection syndrome 4B, autosomal recessive, 618523 (3), Autosomal recessive
<i>IL7R</i>	146661	Immunodeficiency 104, severe combined, 608971 (3), Autosomal recessive
<i>ILRUN</i>	612217	No OMIM phenotype
<i>INO80</i>	610169	No OMIM phenotype
<i>IPO8</i>	605600	VISS syndrome, 619472 (3), Autosomal recessive
<i>IRAK1</i>	300283	No OMIM phenotype
<i>IRAK4</i>	606883	Immunodeficiency 67, 607676 (3), Autosomal recessive
<i>IRF2BP2</i>	615332	?Immunodeficiency, common variable, 14, 617765 (3), Autosomal dominant
<i>IRF3</i>	603734	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532 (3), Autosomal dominant
<i>IRF4</i>	601900	[Skin/hair/eye pigmentation, variation in, 8], 611724 (3)
<i>IRF7</i>	605047	?Immunodeficiency 39, 616345 (3), Autosomal recessive
<i>IRF8</i>	601565	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 (3), Autosomal dominant; Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 (3), Autosomal recessive
<i>IRF9</i>	147574	Immunodeficiency 65, susceptibility to viral infections, 618648 (3), Autosomal recessive
<i>ISG15</i>	147571	Immunodeficiency 38, 616126 (3), Autosomal recessive
<i>ITCH</i>	606409	Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3), Autosomal recessive
<i>ITGB2</i>	600065	Leukocyte adhesion deficiency, 116920 (3), Autosomal recessive
<i>ITK</i>	186973	Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive
<i>ITPKB</i>	147522	No OMIM phenotype
<i>IVNS1ABP</i>	609209	Immunodeficiency 70, 618969 (3), Autosomal dominant
<i>JAGN1</i>	616012	Neutropenia, severe congenital, 6, autosomal recessive, 616022 (3), Autosomal recessive
<i>JAK1</i>	147795	Autoinflammation, immune dysregulation, and eosinophilia, 618999 (3), Autosomal dominant
<i>JAK3</i>	600173	SCID, autosomal recessive, T-negative/B-positive type, 600802 (3), Autosomal recessive
<i>KDM6A</i>	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
<i>KLF2</i>	602016	No OMIM phenotype

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<i>KMT2A</i>	159555	Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
<i>KMT2D</i>	602113	Kabuki syndrome 1, 147920 (3), Autosomal dominant
<i>KRAS</i>	190070	Gastric cancer, somatic, 613659 (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)
<i>LACC1</i>	613409	Juvenile arthritis, 618795 (3), Autosomal recessive
<i>LAMTOR2</i>	610389	Immunodeficiency due to defect in MAPBP-interacting protein, 610798 (3), Autosomal recessive
<i>LAT</i>	602354	Immunodeficiency 52, 617514 (3), Autosomal recessive
<i>LCK</i>	153390	?Immunodeficiency 22, 615758 (3), Autosomal recessive
<i>LCP2</i>	601603	?Immunodeficiency 81, 619374 (3), Autosomal recessive
<i>LIG1</i>	126391	Immunodeficiency 96, 619774 (3), Autosomal recessive
<i>LIG4</i>	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
<i>LIPA</i>	613497	Wolman disease, 278000 (3), Autosomal recessive; Cholesteryl ester storage disease, 278000 (3), Autosomal recessive
<i>LPIN2</i>	605519	Majeed syndrome, 609628 (3)
<i>LRBA</i>	606453	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive
<i>LSM11</i>	617910	?Aicardi-Goutieres syndrome 8, 619486 (3), Autosomal recessive
<i>LY96</i>	605243	No OMIM phenotype
<i>LYST</i>	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
<i>MAGT1</i>	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
<i>MALT1</i>	604860	Immunodeficiency 12, 615468 (3), Autosomal recessive
<i>MAN2B1</i>	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
<i>MAN2B2</i>	618899	No OMIM phenotype
<i>MAP1LC3B2</i>	No OMIM gene	No OMIM phenotype
<i>MAP3K14</i>	604655	No OMIM phenotype
<i>MAPK8</i>	601158	No OMIM phenotype
<i>MASP1</i>	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
<i>MASP2</i>	605102	MASP2 deficiency, 613791 (3), Autosomal recessive
<i>MBL2</i>	154545	{Chronic infections, due to MBL deficiency}, 614372 (3), Autosomal dominant
<i>MCM10</i>	609357	Immunodeficiency 80 with or without cardiomyopathy, 619313 (3), Autosomal recessive

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<i>MCM4</i>	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive
<i>MEFV</i>	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
<i>MOGS</i>	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
<i>MPEG1</i>	610390	Immunodeficiency 77, 619223 (3), Autosomal dominant
<i>MPO</i>	606989	{Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; Myeloperoxidase deficiency, 254600 (3), Autosomal recessive; {Lung cancer, protection against, in smokers} (3)
<i>MRTFA</i>	606078	?Immunodeficiency 66, 618847 (3), Autosomal recessive
<i>MS4A1</i>	112210	?Immunodeficiency, common variable, 5, 613495 (3), Autosomal recessive
<i>MSH6</i>	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), Autosomal dominant, Somatic mutation
<i>MSN</i>	309845	Immunodeficiency 50, 300988 (3), X-linked recessive
<i>MTHFD1</i>	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
<i>MVK</i>	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
<i>MYD88</i>	602170	Macroglobulinemia, Waldenstrom, somatic, 153600 (3); Immunodeficiency 68, 612260 (3), Autosomal recessive
<i>MYO5B</i>	606540	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 10, 619868 (3), Autosomal recessive
<i>MYSM1</i>	612176	Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive
<i>NBAS</i>	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive
<i>NBN</i>	602667	Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
<i>NCF1</i>	608512	Chronic granulomatous disease 1, autosomal recessive, 233700 (3), Autosomal recessive
<i>NCF2</i>	608515	Chronic granulomatous disease 2, autosomal recessive, 233710 (3), Autosomal recessive
<i>NCF4</i>	601488	Chronic granulomatous disease 3, autosomal recessive, 613960 (3), Autosomal recessive
<i>NCKAP1L</i>	141180	Immunodeficiency 72 with autoinflammation, 618982 (3), Autosomal recessive

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<i>NCSTN</i>	605254	Acne inversa, familial, 1, 142690 (3), Autosomal dominant
<i>NFAT5</i>	604708	No OMIM phenotype
<i>NFE2L2</i>	600492	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 (3), Autosomal dominant
<i>NFKB1</i>	164011	Immunodeficiency, common variable, 12, 616576 (3), Autosomal dominant
<i>NFKB2</i>	164012	Immunodeficiency, common variable, 10, 615577 (3), Autosomal dominant
<i>NFKBIA</i>	164008	Ectodermal dysplasia and immunodeficiency 2, 612132 (3), Autosomal dominant
<i>NHEJ1</i>	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 (3)
<i>NHP2</i>	606470	Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive
<i>NKX2-5</i>	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
<i>NLRC4</i>	606831	?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant; Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant
<i>NLRP1</i>	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 dominant, Autosomal recessive; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant
<i>NLRP12</i>	609648	Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant
<i>NLRP3</i>	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
<i>NLRP7</i>	609661	Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive
<i>NOD2</i>	605956	Blau syndrome, 186580 (3), Autosomal dominant; {Yao syndrome}, 617321 (3), Multifactorial; {Inflammatory bowel disease 1, Crohn disease}, 266600 (3), Multifactorial
<i>NOP10</i>	606471	Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
<i>NOS2</i>	163730	{Malaria, resistance to}, 611162 (3)

<i>NPC1</i>	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
<i>NRAS</i>	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)
<i>NSMCE3</i>	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive
<i>OAS1</i>	164350	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinemia, 618042 (3), Autosomal dominant
<i>ORAI1</i>	610277	Immunodeficiency 9, 612782 (3), Autosomal recessive; Myopathy, tubular aggregate, 2, 615883 (3), Autosomal dominant
<i>OSTM1</i>	607649	Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive
<i>OTULIN</i>	615712	Autoinflammation, panniculitis, and dermatosis syndrome, 617099 (3), Autosomal recessive; {Immunodeficiency 107, susceptibility to invasive staphylococcus aureus infection}, 619986 (3), Autosomal dominant
<i>PARN</i>	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 (3), Autosomal dominant
<i>PAX1</i>	167411	Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive
<i>PCCA</i>	232000	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PCCB</i>	232050	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PDCD1</i>	600244	{Multiple sclerosis, disease progression, modifier of}, 126200 (3), Multifactorial; {Systemic lupus erythematosus, susceptibility to, 2}, 605218 (3)
<i>PEPD</i>	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
<i>PGM3</i>	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
<i>PI4KA</i>	600286	Spastic paraplegia 84, autosomal recessive, 619621 (3), Autosomal recessive; Gastrointestinal defects and immunodeficiency syndrome 2, 619708 (3), Autosomal recessive; Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive
<i>PIK3CD</i>	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
<i>PIK3CG</i>	601232	Immunodeficiency 97 with autoinflammation, 619802 (3), Autosomal recessive

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<i>PIK3R1</i>	171833	Immunodeficiency 36, 616005 (3), Autosomal dominant; ?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; SHORT syndrome, 269880 (3), Autosomal dominant
<i>PLCG2</i>	600220	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 (3), Autosomal dominant; Familial cold autoinflammatory syndrome 3, 614468 (3), Autosomal dominant
<i>PLEKHM1</i>	611466	?Osteopetrosis, autosomal recessive 6, 611497 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 3, 618107 (3), Autosomal dominant
<i>PLG</i>	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Angioedema, hereditary, 4, 619360 (3), Autosomal dominant; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
<i>PMS2</i>	600259	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3); Mismatch repair cancer syndrome 4, 619101 (3), Autosomal recessive
<i>PNP</i>	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive
<i>POLA1</i>	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive
<i>POLD1</i>	174761	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant
<i>POLD2</i>	600815	No OMIM phenotype
<i>POLE</i>	174762	{Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive; IMAGE-I syndrome, 618336 (3), Autosomal recessive
<i>POLE2</i>	602670	No OMIM phenotype
<i>POLR3A</i>	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
<i>POLR3C</i>	617454	No OMIM phenotype
<i>POLR3F</i>	617455	?Immunodeficiency 101 (varicella zoster virus-specific), 619872 (3), Autosomal dominant
<i>POMP</i>	613386	Proteasome-associated autoinflammatory syndrome 2, 618048 (3), Autosomal dominant; Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive
<i>POU2AF1</i>	601206	No OMIM phenotype
<i>PRF1</i>	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
<i>PRIM1</i>	176635	Primordial dwarfism-immunodeficiency-lipodystrophy syndrome, 620005 (3), Autosomal recessive

<i>PRKCD</i>	176977	Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive
<i>PRKDC</i>	600899	Immunodeficiency 26, with or without neurologic abnormalities, 615966 (3), Autosomal recessive
<i>PSEN1</i>	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
<i>PSENE1</i>	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant
<i>PSMA3</i>	176843	No OMIM phenotype
<i>PSMB10</i>	176847	Proteasome-associated autoinflammatory syndrome 5, 619175 (3), Autosomal recessive
<i>PSMB4</i>	602177	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591 (3), Autosomal recessive
<i>PSMB8</i>	177046	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040 (3), Autosomal recessive
<i>PSMB9</i>	177045	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591 (3), Autosomal recessive
<i>PSMG2</i>	609702	?Proteasome-associated autoinflammatory syndrome 4, 619183 (3), Autosomal recessive
<i>PSTPIP1</i>	606347	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 (3), Autosomal dominant
<i>PSTPIP2</i>	616046	No OMIM phenotype
<i>PTEN</i>	601728	{Glioma susceptibility 2}, 613028 (3); {Meningioma}, 607174 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos disease, 158350 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3); Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant
<i>PTPN11</i>	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)
<i>PTPN2</i>	176887	No OMIM phenotype
<i>PTPN6</i>	176883	No OMIM phenotype
<i>PTPRC</i>	151460	Immunodeficiency 105, severe combined, 619924 (3), Autosomal recessive
<i>RAB27A</i>	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive

<i>RAC2</i>	602049	Immunodeficiency 73A with defective neutrophil chemotaxis and leukocytosis, 608203 (3), Autosomal dominant; ?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 (3), Autosomal recessive; Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986 (3), Autosomal dominant
<i>RAG1</i>	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)
<i>RAG2</i>	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
<i>RANBP2</i>	601181	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033 (3), Autosomal dominant
<i>RASGRP1</i>	603962	Immunodeficiency 64, 618534 (3), Autosomal recessive
<i>RBCK1</i>	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
<i>RC3H1</i>	609424	?Immune dysregulation and systemic hyperinflammation syndrome, 618998 (3)
<i>RECQL4</i>	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive
<i>REL</i>	164910	Immunodeficiency 92, 619652 (3), Autosomal recessive
<i>RELA</i>	164014	?Mucocutaneous ulceration, chronic, 618287 (3), Autosomal dominant
<i>RELB</i>	604758	?Immunodeficiency 53, 617585 (3), Autosomal recessive
<i>RFX5</i>	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
<i>RFXANK</i>	603200	MHC class II deficiency, complementation group B, 209920 (3), Autosomal recessive
<i>RFXAP</i>	601861	Bare lymphocyte syndrome, type II, complementation group D, 209920 (3), Autosomal recessive
<i>RHOG</i>	179505	No OMIM phenotype
<i>RHOH</i>	602037	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307 (3), Autosomal recessive
<i>RIGI (DDX58)</i>	609631	Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant
<i>RIPK1</i>	603453	Immunodeficiency 57 with autoinflammation, 618108 (3), Autosomal recessive; Autoinflammation with episodic fever and lymphadenopathy, 618852 (3), Autosomal dominant

<i>RMRP</i>	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive
<i>RNASEH2A</i>	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
<i>RNASEH2B</i>	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
<i>RNASEH2C</i>	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
<i>RNF168</i>	612688	RIDDLE syndrome, 611943 (3), Autosomal recessive
<i>RNF31</i>	612487	No OMIM phenotype
<i>RNU7-1</i>	617876	Aicardi-Goutieres syndrome 9, 619487 (3), Autosomal recessive
<i>RORC</i>	602943	Immunodeficiency 42, 616622 (3), Autosomal recessive
<i>RPSA</i>	150370	Asplenia, isolated congenital, 271400 (3), Autosomal dominant
<i>RTEL1</i>	608833	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal dominant, Autosomal recessive
<i>SAMD9</i>	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
<i>SAMD9L</i>	611170	Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant; Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 (3), Autosomal dominant; Spinocerebellar ataxia 49, 619806 (3), Autosomal dominant
<i>SAMHD1</i>	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
<i>SASH3</i>	300441	Immunodeficiency 102, 301082 (3), X-linked recessive
<i>SBDS</i>	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
<i>SDHA</i>	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
<i>SEC61A1</i>	609213	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056 (3), Autosomal dominant
<i>SEMA3E</i>	608166	?CHARGE syndrome, 214800 (3), Autosomal dominant
<i>SERPING1</i>	606860	Angioedema, hereditary, 1 and 2, 106100 (3), Autosomal dominant, Autosomal recessive; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant
<i>SGPL1</i>	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive
<i>SH2D1A</i>	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
<i>SH3BP2</i>	602104	Cherubism, 118400 (3), Autosomal dominant

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<i>SH3KBP1</i>	300374	?Immunodeficiency 61, 300310 (3), X-linked recessive
<i>SKIC2</i> (<i>SKIV2L</i>)	600478	Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive
<i>SKIC3</i> (<i>TTC37</i>)	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
<i>SLC11A1</i>	600266	{ <i>Mycobacterium tuberculosis</i> , susceptibility to infection by}, 607948 (3); {Buruli ulcer, susceptibility to}, 610446 (3)
<i>SLC29A3</i>	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
<i>SLC35C1</i>	605881	Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive
<i>SLC37A4</i>	602671	Glycogen storage disease Ib, 232220 (3), Autosomal recessive; Congenital disorder of glycosylation, type IIw, 619525 (3), Autosomal dominant; Glycogen storage disease Ic, 232240 (3), Autosomal recessive
<i>SLC39A7</i>	601416	Agammaglobulinemia 9, autosomal recessive, 619693 (3), Autosomal recessive
<i>SLC46A1</i>	611672	Folate malabsorption, hereditary, 229050 (3), Autosomal recessive
<i>SLC7A7</i>	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
<i>SLC9A3</i>	182307	Diarrhea 8, secretory sodium, congenital, 616868 (3), Autosomal recessive
<i>SLCO2A1</i>	601460	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 (3), Autosomal dominant; Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441 (3), Autosomal recessive
<i>SMARCAL1</i>	606622	Schimke immunosseous dysplasia, 242900 (3), Autosomal recessive
<i>SMARCD2</i>	601736	Specific granule deficiency 2, 617475 (3), Autosomal recessive
<i>SNORA31</i>	619378	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 10}, 619396 (3), Autosomal dominant
<i>SNX10</i>	614780	Osteopetrosis, autosomal recessive 8, 615085 (3), Autosomal recessive
<i>SOCS1</i>	603597	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375 (3), Autosomal dominant
<i>SOCS4</i>	616337	No OMIM phenotype
<i>SP110</i>	604457	{ <i>Mycobacterium tuberculosis</i> , susceptibility to}, 607948 (3); Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive
<i>SPI1</i>	165170	Agammaglobulinemia 10, autosomal dominant, 619707 (3), Autosomal dominant
<i>SPINK5</i>	605010	Netherton syndrome, 256500 (3), Autosomal recessive
<i>SPPL2A</i>	608238	Immunodeficiency 86, mycobacteriosis, 619549 (3), Autosomal recessive
<i>SRP54</i>	604857	Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant
<i>SRP72</i>	602122	Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant

<i>STAT1</i>	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
<i>STAT2</i>	600556	Pseudo-TORCH syndrome 3, 618886 (3), Autosomal recessive; Immunodeficiency 44, 616636 (3), Autosomal recessive
<i>STAT3</i>	102582	Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant
<i>STAT4</i>	600558	{Systemic lupus erythematosus, susceptibility to, 11}, 612253 (3)
<i>STAT5B</i>	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)
<i>STAT6</i>	601512	No OMIM phenotype
<i>STIM1</i>	605921	Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive
<i>STING1</i>	612374	STING-associated vasculopathy, infantile-onset, 615934 (3), Autosomal dominant
<i>STK4</i>	604965	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 (3)
<i>STN1</i>	613128	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive
<i>STX11</i>	605014	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive
<i>STXBP2</i>	601717	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101 (3)
<i>STXBP3</i>	608339	No OMIM phenotype
<i>SYK</i>	600085	Immunodeficiency 82 with systemic inflammation, 619381 (3), Autosomal dominant
<i>TAFAZZIN</i>	300394	Barth syndrome, 302060 (3), X-linked recessive
<i>TAP1</i>	170260	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
<i>TAP2</i>	170261	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 (3), Autosomal recessive
<i>TAPBP</i>	601962	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
<i>TBK1</i>	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

<i>TBX1</i>	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
<i>TBX21</i>	604895	Asthma and nasal polyps, 208550 (3), Autosomal recessive; ?Immunodeficiency 88, 619630 (3), Autosomal recessive; {Asthma, aspirin-induced, susceptibility to}, 208550 (3), Autosomal recessive
<i>TCF3</i>	147141	Agammaglobulinemia 8B, autosomal recessive, 619824 (3), Autosomal recessive; Agammaglobulinemia 8A, autosomal dominant, 616941 (3), Autosomal dominant
<i>TCIRG1</i>	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive
<i>TCN2</i>	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
<i>TERC</i>	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
<i>TERT</i>	187270	Dyskeratosis congenita, autosomal dominant 2, 613989 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 4, 613989 (3), Autosomal dominant, Autosomal recessive; {Melanoma, cutaneous malignant, 9}, 615134 (3); Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Autosomal dominant, Somatic mutation
<i>TET2</i>	612839	Myelodysplastic syndrome, somatic, 614286 (3); Immunodeficiency 75, 619126 (3), Autosomal recessive
<i>TFRC</i>	190010	Immunodeficiency 46, 616740 (3), Autosomal recessive
<i>TGFB1</i>	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
<i>TGFBR1</i>	190181	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant
<i>TGFBR2</i>	190182	Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3)
<i>THBD</i>	188040	Thrombophilia 12 due to thrombomodulin defect, 614486 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant
<i>TICAM1</i>	607601	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850 (3), Autosomal dominant, Autosomal recessive

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<i>TINF2</i>	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
<i>TIRAP</i>	606252	{Malaria, protection against}, 611162 (3); {Tuberculosis, protection against}, 607948 (3); {Bacteremia, protection against}, 614382 (3)
<i>TLR3</i>	603029	{HIV1 infection, resistance to}, 609423 (3); {Immunodeficiency 83, susceptibility to viral infections}, 613002 (3), Autosomal dominant, Autosomal recessive
<i>TLR4</i>	603030	Macular degeneration, age-related, 10, 611488 (2)
<i>TLR7</i>	300365	Immunodeficiency 74, COVID19-related, X-linked, 301051 (3), X-linked recessive; Systemic lupus erythematosus 17, 301080 (3), X-linked dominant
<i>TLR8</i>	300366	Immunodeficiency 98 with autoinflammation, X-linked, 301078 (3), Somatic mosaicism, X-linked
<i>TMC6</i>	605828	Epidermodysplasia verruciformis, 226400 (3), Autosomal recessive
<i>TMC8</i>	605829	Epidermodysplasia verruciformis 2, 618231 (3), Autosomal recessive
<i>TNFAIP3</i>	191163	Autoinflammatory syndrome, familial, Behcet-like 1, 616744 (3), Autosomal dominant
<i>TNFRSF11A</i>	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
<i>TNFRSF13B</i>	604907	Immunodeficiency, common variable, 2, 240500 (3), Autosomal dominant, Autosomal recessive; Immunoglobulin A deficiency 2, 609529 (3)
<i>TNFRSF13C</i>	606269	Immunodeficiency, common variable, 4, 613494 (3), Autosomal recessive
<i>TNFRSF1A</i>	191190	{Multiple sclerosis, susceptibility to, 5}, 614810 (3); Periodic fever, familial, 142680 (3), Autosomal dominant
<i>TNFRSF4</i>	600315	?Immunodeficiency 16, 615593 (3), Autosomal recessive
<i>TNFRSF9</i>	602250	No OMIM phenotype
<i>TNFSF11</i>	602642	Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive
<i>TNFSF12</i>	602695	No OMIM phenotype
<i>TNFSF13</i>	604472	No OMIM phenotype
<i>TOP2B</i>	126431	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296 (3), Autosomal dominant
<i>TPP2</i>	190470	Immunodeficiency 78 with autoimmunity and developmental delay, 619220 (3), Autosomal recessive
<i>TRAC</i>	186880	Immunodeficiency 7, TCR-alpha/beta deficient, 615387 (3), Autosomal recessive
<i>TRAF3</i>	601896	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849 (3)
<i>TRAF3IP2</i>	607043	?Candidiasis, familial, 8, 615527 (3), Autosomal recessive; {Psoriasis susceptibility 13}, 614070 (3)

<i>TREX1</i>	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal dominant, Autosomal recessive; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
<i>TRIM22</i>	606559	No OMIM phenotype
<i>TRNT1</i>	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
<i>TTC7A</i>	609332	Gastrointestinal defects and immunodeficiency syndrome, 243150 (3), Autosomal recessive
<i>TYK2</i>	176941	Immunodeficiency 35, 611521 (3), Autosomal recessive
<i>UBA1</i>	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive; VEXAS syndrome, somatic, 301054 (3)
<i>UNC119</i>	604011	?Immunodeficiency 13, 615518 (3), Autosomal dominant; ?Cone-rod dystrophy (3)
<i>UNC13D</i>	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3), Autosomal recessive
<i>UNC93B1</i>	608204	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551 (3)
<i>UNG</i>	191525	Immunodeficiency with hyper IgM, type 5, 608106 (3), Autosomal recessive
<i>USB1</i>	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive
<i>USP18</i>	607057	Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive
<i>VAV1</i>	164875	No OMIM phenotype
<i>VPS13B</i>	607817	Cohen syndrome, 216550 (3), Autosomal recessive
<i>VPS45</i>	610035	Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive
<i>WAS</i>	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
<i>WDR1</i>	604734	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550 (3), Autosomal recessive
<i>WIPF1</i>	602357	Wiskott-Aldrich syndrome 2, 614493 (3), Autosomal recessive
<i>WRAP53</i>	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
<i>XBP1</i>	194355	{Major affective disorder-7, susceptibility to}, 612371 (3)
<i>XIAP</i>	300079	Lymphoproliferative syndrome, X-linked, 2, 300635 (3), X-linked recessive

<i>ZAP70</i>	176947	Immunodeficiency 48, 269840 (3), Autosomal recessive; Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive
<i>ZBTB24</i>	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
<i>ZNF341</i>	618269	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282 (3), Autosomal recessive
<i>ZNFX1</i>	618931	Immunodeficiency 91 and hyperinflammation, 619644 (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: August 24, 2022

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.