

PID diagnostic panel		
versie	18-04-2019 (414 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 dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal recessive, Autosomal dominant
<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), Autosomal recessive, Somatic mosaicism; Severe combined immunodeficiency due to ADA deficiency, 102700 (3), Autosomal recessive, Somatic mosaicism
<i>ADA2</i>	607575	Polyarteritis nodosa, childhood-onset, 615688 (3), Autosomal recessive; ?Sneddon syndrome, 182410 (3), Autosomal recessive
<i>ADAM17</i>	603639	?Inflammatory skin and bowel disease, neonatal, 1, 614328 (3), Autosomal recessive
<i>ADAR</i>	146920	Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive; Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant
<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 recessive, Autosomal dominant
<i>AK2</i>	103020	Reticular dysgenesis, 267500 (3), Autosomal recessive
<i>AKT1</i>	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
<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>AP4E1</i>	607244	Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive; Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant

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<i>APOL1</i>	603743	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 (3); {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 (3)
<i>ARPC1B</i>	604223	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718 (3), Autosomal recessive
<i>ATM</i>	607585	Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant; Lymphoma, B-cell non-Hodgkin, somatic (3); Lymphoma, mantle cell, somatic (3); T-cell prolymphocytic leukemia, 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>ATP6V0A2</i>	611716	Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive; Wrinkly skin syndrome, 278250 (3), Autosomal recessive
<i>B2M</i>	109700	?Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Immunodeficiency 43, 241600 (3), Autosomal recessive
<i>BACH2</i>	605394	No OMIM phenotype
<i>BCL10</i>	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)
<i>BCL11B</i>	606558	?Immunodeficiency 49, 617237 (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 and isolated hormone deficiency, 307200 (3), X-linked recessive; Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive
<i>BTLA</i>	607925	No OMIM phenotype
<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)
<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)

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<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); [Eculizumab, poor response to], 615749 (3), Autosomal dominant
<i>C6</i>	217050	C6 deficiency, 612446 (3); Combined C6/C7 deficiency (3)
<i>C6orf106</i>	612217	No OMIM phenotype
<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)
<i>CA2</i>	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
<i>CARD11</i>	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
<i>CARD14</i>	607211	Pityriasis rubra pilaris, 173200 (3), Autosomal dominant; Psoriasis 2, 602723 (3), Autosomal dominant
<i>CARD9</i>	607212	Candidiasis, familial, 2, autosomal recessive, 212050 (3), Autosomal recessive
<i>CARMIL2</i>	610859	No OMIM phenotype
<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	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 (3), Autosomal recessive; {Breast cancer, protection against}, 114480 (3), Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); {Lung cancer, protection against}, 211980 (3), Autosomal recessive
<i>CBL</i>	165360	?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation; Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant
<i>CCBE1</i>	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), 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>CD3D</i>	186790	Immunodeficiency 19, 615617 (3), Autosomal recessive

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<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	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 recessive, Autosomal dominant
<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	No OMIM phenotype
<i>CD79A</i>	112205	Agammaglobulinemia 3, 613501 (3), Autosomal recessive
<i>CD79B</i>	147245	Agammaglobulinemia 6, 612692 (3), Autosomal recessive
<i>CD80</i>	112203	No OMIM phenotype
<i>CD81</i>	186845	Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive
<i>CD86</i>	601020	No OMIM phenotype
<i>CD8A</i>	186910	CD8 deficiency, familial, 608957 (3), Autosomal recessive
<i>CDCA7</i>	609937	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910 (3), Autosomal recessive
<i>CDKN2A</i>	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
<i>CEBPE</i>	600749	Specific granule deficiency, 245480 (3), Autosomal recessive
<i>CFB</i>	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)
<i>CFD</i>	134350	Complement factor D deficiency, 613912 (3), Autosomal recessive
<i>CFH</i>	134370	Basal lamina 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)
<i>CFHR1</i>	134371	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant

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<i>CFHR2</i>	600889	No OMIM phenotype
<i>CFHR3</i>	605336	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant
<i>CFHR4</i>	605337	No OMIM phenotype
<i>CFHR5</i>	608593	Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant
<i>CFI</i>	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
<i>CFP</i>	300383	Properdin deficiency, X-linked, 312060 (3), X-linked recessive
<i>CFTR</i>	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)
<i>CHD7</i>	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
<i>CIITA</i>	600005	Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive; {Rheumatoid arthritis, susceptibility to}, 180300 (3)
<i>CLCN7</i>	602727	Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3)
<i>CLEC7A</i>	606264	{Aspergillosis, susceptibility to}, 614079 (3); Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive
<i>CLPB</i>	616254	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 (3), Autosomal recessive
<i>CNBP</i>	116955	Myotonic dystrophy 2, 602668 (3), Autosomal dominant
<i>COLEC11</i>	612502	3MC syndrome 2, 265050 (3), Autosomal recessive
<i>COPA</i>	601924	{Autoimmune interstitial lung, joint, and kidney disease}, 616414 (3), Autosomal dominant
<i>CORO1A</i>	605000	Immunodeficiency 8, 615401 (3), Autosomal recessive
<i>CPT2</i>	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
<i>CR2</i>	120650	Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive; {Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3)
<i>CSF2RA</i>	425000	No OMIM phenotype

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<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
<i>CTC1</i>	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
<i>CTLA4</i>	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
<i>CTPS1</i>	123860	Immunodeficiency 24, 615897 (3), Autosomal recessive
<i>CTSC</i>	602365	Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive; Periodontitis 1, juvenile, 170650 (3), Autosomal recessive
<i>CXCL12</i>	600835	{AIDS, resistance to}, 609423 (3)
<i>CXCR4</i>	162643	Myelokathexis, isolated (3); WHIM syndrome, 193670 (3), Autosomal dominant
<i>CYBA</i>	608508	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690 (3), Autosomal recessive
<i>CYBB</i>	300481	Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive; Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive
<i>CYBC1 (C17orf6)</i>	No OMIM gene	No OMIM phenotype
<i>DCLRE1B</i>	609683	No OMIM phenotype
<i>DCLRE1C</i>	605988	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, Athabaskan type, 602450 (3), Autosomal recessive
<i>DDX58</i>	609631	Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant
<i>DGCR6</i>	601279	No OMIM phenotype
<i>DGKE</i>	601440	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (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>DNASE2</i>	126350	No OMIM phenotype
<i>DNMT3B</i>	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive
<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
<i>DOK3</i>	611435	No OMIM phenotype
<i>DTNBP1</i>	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive

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<i>ELANE</i>	130130	Neutropenia, cyclic, 162800 (3), Autosomal dominant; Neutropenia, severe congenital 1, autosomal dominant, 202700 (3), Autosomal dominant
<i>EPG5</i>	615068	Vici syndrome, 242840 (3), Autosomal recessive
<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>EZR</i>	123900	No OMIM phenotype
<i>FAAP24</i>	610884	No OMIM phenotype
<i>FADD</i>	602457	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759 (3), Autosomal recessive
<i>FAS</i>	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)
<i>FASLG</i>	134638	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Autosomal recessive
<i>FASN</i>	600212	No OMIM phenotype
<i>FAT4</i>	612411	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive; Van Maldergem syndrome 2, 615546 (3), Autosomal recessive
<i>FCGR1A</i>	146760	[IgG receptor I, phagocytic, familial deficiency of] (3)
<i>FCGR2A</i>	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
<i>FCGR2B</i>	604590	{Malaria, resistance to}, 611162 (3); {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
<i>FCGR3A</i>	146740	Immunodeficiency 20, 615707 (3), Autosomal recessive
<i>FCGR3B</i>	610665	Neutropenia, alloimmune neonatal (3)
<i>FCGRT</i>	601437	No OMIM phenotype
<i>FCN3</i>	604973	Immunodeficiency due to ficolin 3 deficiency, 613860 (3), Autosomal recessive
<i>FERMT3</i>	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive
<i>FOXN1</i>	600838	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal recessive
<i>FOXP3</i>	300292	{Diabetes mellitus, type I, susceptibility to}, 222100 (3), Autosomal recessive; 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

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<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	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
<i>GATA2</i>	137295	Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Leukemia, acute myeloid, susceptibility to}, 601626 (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>GINS1</i>	610608	Immunodeficiency 55, 617827 (3), Autosomal recessive
<i>HAX1</i>	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
<i>HELLS</i>	603946	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911 (3), Autosomal recessive
<i>HYOU1</i>	601746	No OMIM phenotype
<i>ICOS</i>	604558	Immunodeficiency, common variable, 1, 607594 (3), Autosomal recessive
<i>ICOSLG</i>	605717	No OMIM phenotype
<i>IFIH1</i>	606951	Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
<i>IFNAR2</i>	602376	{Hepatitis B virus, susceptibility to}, 610424 (3); ?Immunodeficiency 45, 616669 (3), Autosomal recessive
<i>IFNGR1</i>	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)
<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 15, 615592 (3), Autosomal recessive

<i>IKBKG</i>	300248	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 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)
<i>IKZF1</i>	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
<i>IL10</i>	124092	{Graft-versus-host disease, protection against}, 614395 (3); {HIV-1, susceptibility to}, 609423 (3); {Rheumatoid arthritis, progression of}, 180300 (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>IL1RL1</i>	601203	No OMIM phenotype
<i>IL1RN</i>	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)
<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>IL2RA</i>	147730	{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3); Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (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
<i>IL36RN</i>	605507	Psoriasis 14, pustular, 614204 (3), Autosomal recessive
<i>IL6ST</i>	600694	No OMIM phenotype
<i>IL7R</i>	146661	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971 (3), Autosomal recessive
<i>INO80</i>	610169	No OMIM phenotype

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<i>INPP5D</i>	601582	No OMIM phenotype
<i>IRAK1</i>	300283	No OMIM phenotype
<i>IRAK2</i>	603304	No OMIM phenotype
<i>IRAK4</i>	606883	IRAK4 deficiency, 607676 (3); Invasive pneumococcal disease, recurrent isolated, 1, 610799 (3)
<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>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>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>JAGN1</i>	616012	Neutropenia, severe congenital, 6, autosomal recessive, 616022 (3), Autosomal recessive
<i>JAK1</i>	147795	No OMIM phenotype
<i>JAK2</i>	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
<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>KMT2A</i>	159555	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 (2), Autosomal dominant; Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
<i>KMT2D</i>	602113	Kabuki syndrome 1, 147920 (3), Autosomal dominant
<i>KRAS</i>	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; Lung cancer, somatic, 211980 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; Pancreatic carcinoma, somatic, 260350 (3); RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3)
<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

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<i>LCK</i>	153390	?Immunodeficiency 22, 615758 (3), Autosomal recessive
<i>LIG1</i>	126391	No OMIM phenotype
<i>LIG4</i>	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
<i>LIPA</i>	613497	Cholesteryl ester storage disease, 278000 (3), Autosomal recessive; Wolman 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>LRRC8A</i>	608360	?Agammaglobulinemia 5, 613506 (3), Autosomal dominant
<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)
<i>MALT1</i>	604860	Immunodeficiency 12, 615468 (3), Autosomal recessive
<i>MAP3K14</i>	604655	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>MCM4</i>	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive
<i>MEFV</i>	608107	Familial Mediterranean fever, AD, 134610 (3), Autosomal dominant; Familial Mediterranean fever, AR, 249100 (3), Autosomal recessive
<i>MLPH</i>	606526	GrisCELLI syndrome, type 3, 609227 (3), Autosomal recessive
<i>MOGS</i>	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
<i>MPO</i>	606989	{Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; {Lung cancer, protection against, in smokers} (3); Myeloperoxidase deficiency, 254600 (3), Autosomal recessive
<i>MRE11</i>	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
<i>MRTFA (MKL1)</i>	606078	Megakaryoblastic leukemia, acute (3)
<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; Endometrial cancer, familial, 608089 (3); Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive
<i>MSN</i>	309845	Immunodeficiency 50, 300988 (3), X-linked recessive
<i>MST1</i>	142408	No OMIM phenotype
<i>MTHFD1</i>	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
<i>MVK</i>	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Mevalonic aciduria, 610377 (3), Autosomal recessive; Prokeratosis 3, multiple types, 175900 (3), Autosomal dominant

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<i>MYD88</i>	602170	Macroglobulinemia, Waldenstrom, somatic, 153600 (3); Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 (3)
<i>MYO5A</i>	160777	GrisCELLI syndrome, type 1, 214450 (3), Autosomal recessive
<i>MYSM1</i>	612176	No OMIM phenotype
<i>NBAS</i>	608025	Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive; Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive
<i>NBN</i>	602667	Aplastic anemia, 609135 (3); Leukemia, acute lymphoblastic, 613065 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
<i>NCF1</i>	608512	Chronic granulomatous disease due to deficiency of NCF-1, 233700 (3), Autosomal recessive
<i>NCF2</i>	608515	Chronic granulomatous disease due to deficiency of NCF-2, 233710 (3), Autosomal recessive
<i>NCF4</i>	601488	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960 (3), Autosomal recessive
<i>NCSTN</i>	605254	Acne inversa, familial, 1, 142690 (3), Autosomal dominant
<i>NEIL2</i>	608933	No OMIM phenotype
<i>NFAT5</i>	604708	No OMIM phenotype
<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, anhidrotic, with T-cell immunodeficiency, 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	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
<i>NLRC4</i>	606831	Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant; ?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant
<i>NLRP1</i>	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)
<i>NLRP12</i>	609648	Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant

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<i>NLRP3</i>	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
<i>NLRP7</i>	609661	Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive
<i>NOD2</i>	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
<i>NOP10</i>	606471	Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
<i>NRAS</i>	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)
<i>NSMCE3</i>	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive
<i>OFD1</i>	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
<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
<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>PEPD</i>	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
<i>PGM3</i>	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
<i>PIGA</i>	311770	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3)
<i>PIK3CD</i>	602839	Immunodeficiency 14, 615513 (3), Autosomal dominant
<i>PIK3R1</i>	171833	?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; Immunodeficiency 36, 616005 (3), Autosomal dominant; SHORT syndrome, 269880 (3), Autosomal dominant

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<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>PMS2</i>	600259	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3); Mismatch repair cancer syndrome, 276300 (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
<i>POLE</i>	174762	{Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive
<i>POLE2</i>	602670	No OMIM phenotype
<i>POT1</i>	606478	{Glioma susceptibility 9}, 616568 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848 (3), Autosomal dominant
<i>PRF1</i>	170280	Aplastic anemia, 609135 (3); Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Lymphoma, non-Hodgkin, 605027 (3)
<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>PSENE1</i>	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant
<i>PSMB4</i>	602177	No OMIM phenotype
<i>PSMB8</i>	177046	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040 (3), Autosomal recessive
<i>PSMB9</i>	177045	No OMIM phenotype
<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	Bannayan-Riley-Ruvalcaba syndrome, 153480 (3); Cowden syndrome 1, 158350 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); {Glioma susceptibility 2}, 613028 (3); Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; Malignant melanoma, somatic, 155600 (3); {Meningioma}, 607174 (3), Autosomal dominant; PTEN hamartoma tumor syndrome (3); {Prostate cancer, somatic}, 176807 (3); Squamous cell carcinoma, head and neck, somatic, 275355 (3); VATER association with macrocephaly and ventriculomegaly, 276950 (3), Autosomal recessive
<i>PTPN11</i>	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

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<i>PTPN6</i>	176883	No OMIM phenotype
<i>PTPRC</i>	151460	{Hepatitis C virus, susceptibility to}, 609532 (3); Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 (3), Autosomal recessive
<i>RAB27A</i>	603868	GrisCELLI syndrome, type 2, 607624 (3), Autosomal recessive
<i>RAC2</i>	602049	Neutrophil immunodeficiency syndrome, 608203 (3)
<i>RAG1</i>	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
<i>RAG2</i>	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
<i>RANBP2</i>	601181	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033 (3), Autosomal dominant
<i>RASGRP1</i>	603962	No OMIM phenotype
<i>RBCK1</i>	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
<i>RECQL4</i>	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive; Rothmund-Thomson syndrome, 268400 (3), Autosomal recessive
<i>RELA</i>	164014	No OMIM phenotype
<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>RHOH</i>	602037	No OMIM phenotype
<i>RIPK1</i>	603453	No OMIM phenotype
<i>RMRP</i>	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (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>RORC</i>	602943	Immunodeficiency 42, 616622 (3), Autosomal recessive

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<i>RPSA</i>	150370	Asplenia, isolated congenital, 271400 (3), Autosomal dominant
<i>RRAS2</i>	600098	Ovarian carcinoma (3)
<i>RTEL1</i>	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
<i>RTL1</i>	611896	No OMIM phenotype
<i>SAMD9</i>	610456	MIRAGE syndrome, 617053 (3), Autosomal dominant; Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive
<i>SAMD9L</i>	611170	Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant
<i>SAMHD1</i>	606754	Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive; ?Chilblain lupus 2, 614415 (3), Autosomal dominant
<i>SBDS</i>	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
<i>SEMA3E</i>	608166	?CHARGE syndrome, 214800 (3), Autosomal dominant
<i>SERPING1</i>	606860	Angioedema, hereditary, types I and II, 106100 (3), Autosomal recessive, Autosomal dominant; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant
<i>SH2D1A</i>	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
<i>SH3BP2</i>	602104	Cherubism, 118400 (3), Autosomal dominant
<i>SKIV2L</i>	600478	Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive
<i>SLC11A1</i>	600266	{Buruli ulcer, susceptibility to}, 610446 (3); {Mycobacterium tuberculosis, susceptibility to infection by}, 607948 (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; Glycogen storage disease Ic, 232240 (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>SMARCAL1</i>	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
<i>SMARCD2</i>	601736	Specific granule deficiency 2, 617475 (3), Autosomal recessive
<i>SP110</i>	604457	Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive; {Mycobacterium tuberculosis, susceptibility to}, 607948 (3)
<i>SPINK5</i>	605010	Netherton syndrome, 256500 (3), Autosomal recessive
<i>SRP72</i>	602122	Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant

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<i>STAT1</i>	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
<i>STAT2</i>	600556	Immunodeficiency 44, 616636 (3), Autosomal recessive
<i>STAT3</i>	102582	Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant; Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant
<i>STAT4</i>	600558	{Systemic lupus erythematosus, susceptibility to, 11}, 612253 (3)
<i>STAT5B</i>	604260	Growth hormone insensitivity with immunodeficiency, 245590 (3); Leukemia, acute promyelocytic, somatic, 102578 (3)
<i>STIM1</i>	605921	Immunodeficiency 10, 612783 (3), Autosomal recessive; Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (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, 613101 (3)
<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; Wegener-like granulomatosis (3)
<i>TAPBP</i>	601962	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
<i>TAZ</i>	300394	Barth syndrome, 302060 (3), X-linked 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	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
<i>TCF3</i>	147141	Agammaglobulinemia 8, autosomal dominant, 616941 (3), Autosomal dominant
<i>TCF7L1</i>	604652	No OMIM phenotype
<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	{Aplastic anemia}, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant; {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743 (3), Autosomal dominant

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<i>TERT</i>	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; {Melanoma, cutaneous malignant, 9}, 615134 (3); {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 (3), Autosomal dominant
<i>TFRC</i>	190010	Immunodeficiency 46, 616740 (3), Autosomal recessive
<i>TGFBR1</i>	190181	Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant; {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant
<i>THBD</i>	188040	{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant; Thrombophilia due to thrombomodulin defect, 614486 (3)
<i>TICAM1</i>	607601	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850 (3), Autosomal recessive, Autosomal dominant
<i>TINF2</i>	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
<i>TIRAP</i>	606252	{Bacteremia, protection against}, 614382 (3); {Malaria, protection against}, 611162 (3); {Pneumococcal disease, invasive, protection against}, 610799 (3); {Tuberculosis, protection against}, 607948 (3)
<i>TLR3</i>	603029	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002 (3), Autosomal recessive, Autosomal dominant; {HIV1 infection, resistance to}, 609423 (3)
<i>TMC6</i>	605828	Epidermodysplasia verruciformis, 226400 (3), Autosomal recessive
<i>TMC8</i>	605829	Epidermodysplasia verruciformis, 226400 (3), Autosomal recessive
<i>TMEM173</i>	612374	STING-associated vasculopathy, infantile-onset, 615934 (3), Autosomal dominant
<i>TNFAIP3</i>	191163	Autoinflammatory syndrome, familial, Behcet-like, 616744 (3), Autosomal dominant
<i>TNFRSF13B</i>	604907	Immunodeficiency, common variable, 2, 240500 (3), Autosomal recessive, Autosomal dominant; 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>TNFSF12</i>	602695	No OMIM phenotype
<i>TPP1</i>	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
<i>TPP2</i>	190470	No OMIM phenotype

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<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	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
<i>TRIM21</i>	109092	No OMIM phenotype
<i>TRNT1</i>	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
<i>TTC37</i>	614589	Trichohepatoenteric syndrome 1, 222470 (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>UNC119</i>	604011	?Cone-rod dystrophy (3); ?Immunodeficiency 13, 615518 (3), Autosomal dominant
<i>UNC13D</i>	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3)
<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>VAV2</i>	600428	No OMIM phenotype
<i>VIPAS39</i>	613401	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
<i>VPS13B</i>	607817	Cohen syndrome, 216550 (3), Autosomal recessive
<i>VPS33B</i>	608552	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
<i>VPS45</i>	610035	Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive
<i>WAS</i>	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
<i>WDR1</i>	604734	No OMIM phenotype

<i>WIPF1</i>	602357	?Wiskott-Aldrich syndrome 2, 614493 (3)
<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>XRCC4</i>	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
<i>ZAP70</i>	176947	Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive; Immunodeficiency 48, 269840 (3), Autosomal recessive
<i>ZBTB24</i>	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
<i>ZNF341</i>	No OMIM gene	No OMIM phenotype

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern. OMIM release used for OMIM disease identifiers and descriptions: July 04, 2018

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.