PID diagnostic

Gene panel

Gene panel information

Gene panel	**PID diagnostic**
Version	8
Total genes	530
Activation date	Monday 05 august 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ACD	100 %	609377	?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant
ACP5	100 %	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
ACTB	100 %	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant
ADA	99.97 %	608958	Adenosine deaminase deficiency, partial, 102700 (3), Autosomal recessive, Somatic mosaicism; Severe combined immunodeficiency due to ADA deficiency, 102700 (3), Autosomal recessive, Somatic mosaicism
ADA2	100 %	607575	Sneddon syndrome, 182410 (3), Autosomal recessive; Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 (3), Autosomal recessive
ADAM17	99.94 %	603639	?Inflammatory skin and bowel disease, neonatal, 1, 614328 (3), Autosomal recessive
ADAR	99.84 %	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
AGR2	99.98 %	606358	Respiratory infections, recurrent, and failure to thrive with or without diarrhea, 620233 (3), Autosomal recessive
AICDA	99.94 %	605257	Immunodeficiency with hyper-IgM, type 2, 605258 (3), Autosomal recessive
AIRE	99.95 %	607358	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal recessive, Autosomal dominant
AK2	99.39 %	103020	Reticular dysgenesis, 267500 (3), Autosomal recessive
ALPI	100 %	171740	*No OMIM phenotypes*
ALPK1	99.92 %	607347	ROSAH syndrome, 614979 (3), Autosomal dominant
ANGPT1	99.97 %	601667	?Angioedema, hereditary, 5, 619361 (3), Autosomal dominant
ANKZF1	100 %	617541	*No OMIM phenotypes*
AP1S3	100 %	615781	{Psoriasis 15, pustular, susceptibility to}, 616106 (3), Autosomal dominant
AP3B1	99.89 %	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP3D1	100 %	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
APOL1	99.99 %	603743	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 (3), Autosomal dominant
ARHGEF1	99.97 %	601855	?Immunodeficiency 62, 618459 (3), Autosomal recessive
ARPC1B	99.92 %	604223	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718 (3), Autosomal recessive
ARPC5	99.85 %	604227	*No OMIM phenotypes*

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ATAD3A	99.62 %	612316	Harel-Yoon syndrome, 617183 (3), Autosomal recessive, Autosomal dominant; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
ATG16L1	99.95 %	610767	{Inflammatory bowel disease (Crohn disease) 10}, 611081 (3)
ATG4A	99.86 %	300663	*No OMIM phenotypes*
ATM	99.83 %	607585	Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3)
ATP6AP1	100 %	300197	Immunodeficiency 47, 300972 (3), X-linked recessive
ATRIP	99.97 %	606605	*No OMIM phenotypes*
B2M	100 %	109700	?Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Immunodeficiency 43, 241600 (3), Autosomal recessive
BACH2	99.99 %	605394	Immunodeficiency 60 and autoimmunity, 618394 (3), Autosomal dominant
BCL10	99.74 %	603517	{Lymphoma, follicular, somatic}, 605027 (3); ?Immunodeficiency 37, 616098 (3), Autosomal recessive; {Sezary syndrome, somatic} (3); {Male germ cell tumor, somatic}, 273300 (3); Lymphoma, MALT, somatic, 137245 (3); {Mesothelioma, somatic}, 156240 (3)
BCL11B	100 %	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
BLK	99.98 %	191305	Maturity-onset diabetes of the young, type 11, 613375 (3), Autosomal dominant
BLM	99.8 %	604610	Bloom syndrome, 210900 (3), Autosomal recessive
BLNK	99.9 %	604515	?Agammaglobulinemia 4, 613502 (3), Autosomal recessive
BLOC1S6	99.98 %	604310	?Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive
BTK	99.88 %	300300	Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive; Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 (3), X-linked recessive
C1QA	99.99 %	120550	C1q deficiency 1, 613652 (3), Autosomal recessive
C1QB	99.58 %	120570	C1q deficiency 2, 620321 (3)
C1QC	99.97 %	120575	C1q deficiency 3, 620322 (3)
C1R	99.99 %	613785	Ehlers-Danlos syndrome, periodontal type, 1, 130080 (3), Autosomal dominant
C1S	99.98 %	120580	C1s deficiency, 613783 (3); Ehlers-Danlos syndrome, periodontal type, 2, 617174 (3), Autosomal dominant
C2	99.99 %	613927	C2 deficiency, 217000 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant
C2orf69	99.97 %	619219	Combined oxidative phosphorylation deficiency 53, 619423 (3), Autosomal recessive
C3	100 %	120700	C3 deficiency, 613779 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3), Autosomal dominant; {Macular degeneration, age-related, 9}, 611378 (3)
C4A	18.13 %	120810	[Blood group, Rodgers], 614374 (3); C4a deficiency, 614380 (3), Autosomal recessive
C4B	21.28 %	120820	C4B deficiency, 614379 (3)
C5	99.92 %	120900	C5 deficiency, 609536 (3), Autosomal recessive; [Eculizumab, poor response to], 615749 (3), Autosomal dominant
C6	99.97 %	217050	C6 deficiency, 612446 (3), Autosomal recessive
C7	99.94 %	217070	C7 deficiency, 610102 (3)

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C8A	99.95 %	120950	C8 deficiency, type I, 613790 (3), Autosomal recessive
C8B	99.37 %	120960	C8 deficiency, type II, 613789 (3), Autosomal recessive
C8G	99.99 %	120930	*No OMIM phenotypes*
C9	99.89 %	120940	C9 deficiency, 613825 (3); {Macular degeneration, age-related, 15, susceptibility to}, 615591 (3), Autosomal dominant
CARD11	99.97 %	607210	B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive
CARD14	99.99 %	607211	Psoriasis 2, 602723 (3), Autosomal dominant; Pityriasis rubra pilaris, 173200 (3), Autosomal dominant
CARD8	99.99 %	609051	?Inflammatory bowel disease (Crohn disease) 30, 619079 (3), Autosomal dominant
CARD9	100 %	607212	Immunodeficiency 103, susceptibility to fungal infection, 212050 (3), Autosomal recessive
CARMIL2	99.99 %	610859	Immunodeficiency 58, 618131 (3), Autosomal recessive
CASP10	99.85 %	601762	Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
CASP8	99.92 %	601763	{Breast cancer, protection against}, 114480 (3), Somatic mutation, Autosomal dominant; ?Caspase 8 lymphadenopathy syndrome, 607271 (3), Autosomal recessive; Hepatocellular carcinoma, somatic, 114550 (3); {Lung cancer, protection against}, 211980 (3), Somatic mutation, Autosomal dominant
CBL	99.95 %	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant
CBLB	99.55 %	604491	Autoimmune disease, multisystem, infantile-onset, 3, 620430 (3), Autosomal recessive
CCBE1	99.52 %	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
CCDC28B	99.99 %	610162	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive
CD19	99.98 %	107265	Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive
CD247	99.79 %	186780	?Immunodeficiency 25, 610163 (3), Autosomal recessive
CD27	99.95 %	186711	Lymphoproliferative syndrome 2, 615122 (3), Autosomal recessive
CD28	99.99 %	186760	*No OMIM phenotypes*
CD3D	100 %	186790	Immunodeficiency 19, severe combined, 615617 (3), Autosomal recessive
CD3E	100 %	186830	Immunodeficiency 18, 615615 (3), Autosomal recessive; Immunodeficiency 18, SCID variant, 615615 (3), Autosomal recessive
CD3G	100 %	186740	Immunodeficiency 17, CD3 gamma deficient, 615607 (3), Autosomal recessive
CD4	100 %	186940	Immunodeficiency 79, 619238 (3), Autosomal recessive; OKT4 epitope deficiency, 613949 (3)
CD40	100 %	109535	Immunodeficiency with hyper-IgM, type 3, 606843 (3), Autosomal recessive
CD40LG	99.88 %	300386	Immunodeficiency, X-linked, with hyper-IgM, 308230 (3), X-linked recessive
CD46	99.86 %	120920	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 (3), Autosomal recessive, Autosomal dominant
CD55	74.12 %	125240	[Blood group Cromer], 613793 (3), Autosomal recessive; Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 (3), Autosomal recessive

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CD59	100 %	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive
CD70	99.99 %	602840	Lymphoproliferative syndrome 3, 618261 (3), Autosomal recessive
CD79A	99.97 %	112205	Agammaglobulinemia 3, 613501 (3), Autosomal recessive
CD79B	99.93 %	147245	Agammaglobulinemia 6, 612692 (3), Autosomal recessive
CD81	99.97 %	186845	Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive
CD8A	99.97 %	186910	CD8 deficiency, familial, 608957 (3), Autosomal recessive
CDC42	98.05 %	116952	Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant
CDCA7	99.88 %	609937	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910 (3), Autosomal recessive
CEBPE	100 %	600749	?Immunodeficiency 108 with autoinflammation, 260570 (3), Autosomal recessive; Specific granule deficiency, 245480 (3), Autosomal recessive, Autosomal dominant
CFB	99.97 %	138470	?Complement factor B deficiency, 615561 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant
CFD	99.99 %	134350	Complement factor D deficiency, 613912 (3), Autosomal recessive
CFH	99.12 %	134370	{Macular degeneration, age-related, 4}, 610698 (3), Autosomal dominant; Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal recessive, Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal recessive, Autosomal dominant
CFHR1	84.44 %	134371	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant
CFHR2	90.26 %	600889	*No OMIM phenotypes*
CFHR3	91.62 %	605336	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant
CFHR4	99.86 %	605337	*No OMIM phenotypes*
CFHR5	99.68 %	608593	Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant
CFI	99.87 %	217030	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant; Complement factor I deficiency, 610984 (3), Autosomal recessive
CFP	99.96 %	300383	Properdin deficiency, X-linked, 312060 (3), X-linked recessive
CFTR	99.45 %	602421	Cystic fibrosis, 219700 (3), Autosomal recessive; Sweat chloride elevation without CF (3); Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; {Hypertrypsinemia, neonatal} (3)
CHD7	99.99 %	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CHUK	99.83 %	600664	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 (3), Autosomal recessive; ?Cocoon syndrome, 613630 (3), Autosomal recessive
CIB1	99.92 %	602293	Epidermodysplasia verruciformis 3, 618267 (3), Autosomal recessive
CIITA	99.99 %	600005	{Rheumatoid arthritis, susceptibility to}, 180300 (3); Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive

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CLCN7	99.99 %	602727	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant
CLEC7A	99.98 %	606264	Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive; {Aspergillosis, susceptibility to}, 614079 (3)
CLPB	99.97 %	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
COL7A1	99.99 %	120120	Nail disorder, nonsyndromic congenital, 8, 607523 (3), Autosomal dominant; Epidermolysis bullosa dystrophica, Bart type, 132000 (3), Autosomal dominant; Epidermolysis bullosa dystrophica inversa, 226600 (3), Autosomal recessive; Epidermolysis bullosa dystrophica, autosomal recessive, 226600 (3), Autosomal recessive; Epidermolysis bullosa, pretibial, 131850 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa dystrophica, autosomal dominant, 131750 (3), Autosomal dominant; Transient bullous of the newborn, 131705 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa dystrophica, localisata variant, 226600 (3), Autosomal recessive
COPA	99.61 %	601924	{Autoimmune interstitial lung, joint, and kidney disease}, 616414 (3), Autosomal dominant
COPG1	99.98 %	615525	*No OMIM phenotypes*
CORO1A	91.71 %	605000	Immunodeficiency 8, 615401 (3), Autosomal recessive
CR2	99.97 %	120650	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3); ?Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive
CRACR2A	100 %	614178	*No OMIM phenotypes*
CSF2RA	93.86 %	306250	Surfactant metabolism dysfunction, pulmonary, 4, 300770 (3)
CSF2RB	100 %	138981	Surfactant metabolism dysfunction, pulmonary, 5, 614370 (3), Autosomal recessive
CSF3R	99.97 %	138971	Neutropenia, severe congenital, 7, autosomal recessive, 617014 (3), Autosomal recessive; ?Neutrophilia, hereditary, 162830 (3), Autosomal dominant
CTC1	100 %	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTLA4	99.99 %	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
CTNNBL1	100 %	611537	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846 (3), Autosomal recessive
CTPS1	98.63 %	123860	Immunodeficiency 24, 615897 (3), Autosomal recessive
CTSC	99.97 %	602365	Periodontitis 1, juvenile, 170650 (3), Autosomal recessive; Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive
CXCR2	100 %	146928	?WHIM syndrome 2, 619407 (3), Autosomal recessive
CXCR4	99.98 %	162643	WHIM syndrome 1, 193670 (3), Autosomal dominant; Myelokathexis, isolated, 193670 (3), Autosomal dominant
CYBA	99.96 %	608508	Chronic granulomatous disease 4, autosomal recessive, 233690 (3), Autosomal recessive

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CYBB	99.87 %	300481	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive; Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive
CYBC1	100 %	618334	Chronic granulomatous disease 5, autosomal recessive, 618935 (3), Autosomal recessive
DBF4	98.6 %	604281	*No OMIM phenotypes*
DBR1	99.92 %	607024	{Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441 (3), Autosomal recessive
DCLRE1B	99.91 %	609683	Dyskeratosis congenita, autosomal recessive 8, 620133 (3), Autosomal recessive
DCLRE1C	99.79 %	605988	Severe combined immunodeficiency, Athabascan type, 602450 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive
DDX58	99.84 %	609631	Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant
DEF6	100 %	610094	Immunodeficiency 87 and autoimmunity, 619573 (3), Autosomal recessive
DHFR	98.89 %	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
DIAPH1	99.95 %	602121	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive
DKC1	99.59 %	300126	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 (3), X-linked dominant; Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DNAJC17	100 %	616844	*No OMIM phenotypes*
DNAJC21	99.67 %	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
DNASE1	100 %	125505	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
DNASE1L3	99.9 %	602244	Systemic lupus erythematosus 16, 614420 (3), Autosomal recessive
DNASE2	100 %	126350	Autoinflammatory-pancytopenia syndrome, 619858 (3), Autosomal recessive
DNMT3B	99.98 %	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive; Facioscapulohumeral muscular dystrophy 4, digenic, 619478 (3), Digenic dominant
DOCK11	99.45 %	300681	Autoinflammatory disease, multisystem, with immune dysregulation, X-linked, 301109 (3), X-linked recessive
DOCK2	100 %	603122	Immunodeficiency 40, 616433 (3), Autosomal recessive
DOCK8	99.86 %	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive
DPP9	100 %	608258	Hatipoglu immunodeficiency syndrome, 620331 (3), Autosomal recessive
DTNBP1	99.89 %	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
DUOX2	98.85 %	606759	Thyroid dyshormonogenesis 6, 607200 (3), Autosomal recessive
DUT	99.9 %	601266	Bone marrow failure and diabetes mellitus syndrome, 620044 (3), Autosomal recessive
EFL1	99.83 %	617538	Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive
ELANE	100 %	130130	Neutropenia, cyclic, 162800 (3), Autosomal dominant; Neutropenia, severe congenital 1, autosomal dominant, 202700 (3), Autosomal dominant
ELF4	99.97 %	300775	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074 (3), X-linked recessive
EPG5	99.95 %	615068	Vici syndrome, 242840 (3), Autosomal recessive
ERBIN	99.69 %	606944	*No OMIM phenotypes*
ERCC6L2	99.94 %	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive

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ETV6	99.99 %	600618	Thrombocytopenia 5, 616216 (3), Autosomal dominant; Leukemia, acute myeloid, somatic, 601626 (3)
EXTL3	99.99 %	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
F12	99.99 %	610619	Angioedema, hereditary, 3, 610618 (3), Autosomal dominant; Factor XII deficiency, 234000 (3), Autosomal recessive
FAAP24	99.95 %	610884	*No OMIM phenotypes*
FADD	99.97 %	602457	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759 (3), Autosomal recessive
FAM111B	99.98 %	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
FAS	99.99 %	134637	Squamous cell carcinoma, burn scar-related, somatic (3); Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant
FASLG	99.84 %	134638	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant
FAT4	99.98 %	612411	Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive
FCGR3A	99.93 %	146740	Immunodeficiency 20, 615707 (3), Autosomal recessive
FCHO1	99.99 %	613437	Immunodeficiency 76, 619164 (3), Autosomal recessive
FCN3	99.48 %	604973	Immunodeficiency due to ficolin 3 deficiency, 613860 (3), Autosomal recessive
FERMT1	99.9 %	607900	Kindler syndrome, 173650 (3), Autosomal recessive
FERMT3	99.99 %	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive
FGL2	99.96 %	605351	*No OMIM phenotypes*
FMNL2	99.9 %	616285	*No OMIM phenotypes*
FNIP1	99.87 %	610594	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705 (3), Autosomal recessive
FOXI3	100 %	612351	Craniofacial microsomia 2, 620444 (3), Autosomal recessive, Autosomal dominant
FOXN1	99.97 %	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
FOXP3	99.93 %	300292	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive
FPR1	100 %	136537	*No OMIM phenotypes*
G6PC3	99.98 %	611045	Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive
G6PD	99.97 %	305900	Hemolytic anemia, G6PD deficient (favism), 300908 (3), X-linked; {Resistance to malaria due to G6PD deficiency}, 611162 (3)
GATA1	99.97 %	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

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GATA2	99.99 %	137295	{Leukemia, acute myeloid, susceptibility to}, 601626 (3), Somatic mutation, Autosomal dominant; Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3)
GFI1	99.88 %	600871	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 (3), Autosomal dominant; Neutropenia, severe congenital 2, autosomal dominant, 613107 (3), Autosomal dominant
GIMAP5	100 %	608086	Portal hypertension, noncirrhotic, 2, 619463 (3), Autosomal recessive
GIMAP6	100 %	616960	*No OMIM phenotypes*
GINS1	99.99 %	610608	Immunodeficiency 55, 617827 (3), Autosomal recessive
GINS4	100 %	610611	*No OMIM phenotypes*
GTF3A	99.99 %	600860	*No OMIM phenotypes*
GUCY2C	99.9 %	601330	Diarrhea 6, 614616 (3), Autosomal dominant; Meconium ileus, 614665 (3), Autosomal recessive
HAVCR2	99.93 %	606652	T-cell lymphoma, subcutaneous panniculitis-like, 618398 (3), Autosomal recessive
HAX1	100 %	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
HCK	100 %	142370	Autoinflammation with pulmonary and cutaneous vasculitis, 620296 (3), Autosomal dominant
HELLS	99.78 %	603946	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911 (3), Autosomal recessive
HMOX1	99.95 %	141250	Heme oxygenase-1 deficiency, 614034 (3), Autosomal recessive; {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (3)
HPS1	100 %	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
HPS4	99.98 %	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
HPS6	100 %	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
HSPA1L	100 %	140559	*No OMIM phenotypes*
HTRA2	99.99 %	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
HYOU1	99.97 %	601746	?Immunodeficiency 59 and hypoglycemia, 233600 (3), Autosomal recessive
ICOS	99.95 %	604558	Immunodeficiency, common variable, 1, 607594 (3), Autosomal recessive
ICOSLG	5.95 %	605717	*No OMIM phenotypes*
IFIH1	99.84 %	606951	Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IFNAR1	99.75 %	107450	Immunodeficiency 106, susceptibility to viral infections, 619935 (3), Autosomal recessive
IFNAR2	89.62 %	602376	{Hepatitis B virus, susceptibility to}, 610424 (3); Immunodeficiency 45, 616669 (3), Autosomal recessive
IFNG	99.5 %	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)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
IFNGR1	99.87 %	107470	{H. pylori infection, susceptibility to}, 600263 (3); Immunodeficiency 27A, mycobacteriosis, AR, 209950 (3), Autosomal recessive; Immunodeficiency 27B, mycobacteriosis, AD, 615978 (3), Autosomal dominant; {Tuberculosis infection, protection against}, 607948 (3); {Tuberculosis, susceptibility to}, 607948 (3); {Hepatitis B virus infection, susceptibility to}, 610424 (3)
IFNGR2	99.95 %	147569	Immunodeficiency 28, mycobacteriosis, 614889 (3), Autosomal recessive
IGHM	100 %	147020	Agammaglobulinemia 1, 601495 (3), Autosomal recessive
IGKC	99.99 %	147200	Kappa light chain deficiency, 614102 (3), Autosomal recessive
IGLL1	100 %	146770	Agammaglobulinemia 2, 613500 (3), Autosomal recessive
IKBKB	99.93 %	603258	Immunodeficiency 15B, 615592 (3), Autosomal recessive; Immunodeficiency 15A, 618204 (3), Autosomal dominant
IKBKG	57.34 %	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
IKZF1	99.92 %	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
IKZF2	99.99 %	606234	*No OMIM phenotypes*
IKZF3	99.96 %	606221	?Immunodeficiency 84, 619437 (3), Autosomal dominant
IL10	100 %	124092	{Rheumatoid arthritis, progression of}, 180300 (3); {Graft-versus-host disease, protection against}, 614395 (3); {HIV-1, susceptibility to}, 609423 (3)
IL10RA	99.99 %	146933	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148 (3), Autosomal recessive
IL10RB	99.99 %	123889	{Hepatitis B virus, susceptibility to}, 610424 (3); Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 (3), Autosomal recessive
IL12B	99.98 %	161561	Immunodeficiency 29, mycobacteriosis, 614890 (3), Autosomal recessive
IL12RB1	94.11 %	601604	Immunodeficiency 30, 614891 (3), Autosomal recessive
IL12RB2	97.66 %	601642	*No OMIM phenotypes*
IL17F	99.99 %	606496	?Candidiasis, familial, 6, autosomal dominant, 613956 (3)
IL17RA	100 %	605461	Immunodeficiency 51, 613953 (3), Autosomal recessive
IL17RC	100 %	610925	Candidiasis, familial, 9, 616445 (3), Autosomal recessive
IL18BP	99.99 %	604113	{?Hepatitis, fulminant viral, susceptibility to}, 618549 (3), Autosomal recessive
IL1RL1	99.76 %	601203	*No OMIM phenotypes*
IL1RN	99.64 %	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
IL21	99.95 %	605384	?Immunodeficiency, common variable, 11, 615767 (3), Autosomal recessive
IL21R	99.7 %	605383	[IgE, elevated level of], 147050 (3), Autosomal dominant; Immunodeficiency 56, 615207 (3), Autosomal recessive
IL23R	97.64 %	607562	{Inflammatory bowel disease 17, protection against}, 612261 (3); {Psoriasis, protection against}, 605606 (3)
IL2RA	99.99 %	147730	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive; {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3)
IL2RB	100 %	146710	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495 (3), Autosomal recessive
IL2RG	99.86 %	308380	Combined immunodeficiency, X-linked, moderate, 312863 (3), X-linked recessive; Severe combined immunodeficiency, X-linked, 300400 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
IL36RN	100 %	605507	Psoriasis 14, pustular, 614204 (3), Autosomal recessive
IL37	100 %	605510	?Inflammatory bowel disease (infantile ulcerative colitis) 31, 619398 (3), Autosomal recessive
IL6R	92.46 %	147880	[Interleukin 6, serum level of, QTL], 614752 (3); Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944 (3), Autosomal recessive; [Interleukin-6 receptor, soluble, serum level of, QTL], 614689 (3)
IL6ST	99.88 %	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
IL7R	99.99 %	146661	Immunodeficiency 104, severe combined, 608971 (3), Autosomal recessive
INO80	99.96 %	610169	*No OMIM phenotypes*
IPO8	97.96 %	605600	VISS syndrome, 619472 (3), Autosomal recessive
IRAK1	99.98 %	300283	*No OMIM phenotypes*
IRAK4	98.85 %	606883	Immunodeficiency 67, 607676 (3), Autosomal recessive
IRF2BP2	100 %	615332	?Immunodeficiency, common variable, 14, 617765 (3), Autosomal dominant
IRF3	99.96 %	603734	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532 (3), Autosomal dominant
IRF4	99.99 %	601900	[Skin/hair/eye pigmentation, variation in, 8], 611724 (3)
IRF7	100 %	605047	?Immunodeficiency 39, 616345 (3), Autosomal recessive
IRF8	99.99 %	601565	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 (3), Autosomal dominant; Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 (3), Autosomal recessive
IRF9	100 %	147574	Immunodeficiency 65, susceptibility to viral infections, 618648 (3), Autosomal recessive
ISG15	100 %	147571	Immunodeficiency 38, 616126 (3), Autosomal recessive
ITCH	95.57 %	606409	Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3), Autosomal recessive
ITGB2	100 %	600065	Leukocyte adhesion deficiency, 116920 (3), Autosomal recessive
ITK	99.91 %	186973	Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive
ITPKB	99.99 %	147522	*No OMIM phenotypes*
IVNS1ABP	99.24 %	609209	Immunodeficiency 70, 618969 (3), Autosomal dominant
JAGN1	100 %	616012	Neutropenia, severe congenital, 6, autosomal recessive, 616022 (3), Autosomal recessive
JAK1	99.32 %	147795	Autoinflammation, immune dysregulation, and eosinophilia, 618999 (3), Autosomal dominant
JAK3	99.99 %	600173	SCID, autosomal recessive, T-negative/B-positive type, 600802 (3), Autosomal recessive
KARS1	99.98 %	601421	Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive; Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196 (3), Autosomal recessive
KCNA5	100 %	176267	Atrial fibrillation, familial, 7, 612240 (3), Autosomal dominant
KDM6A	99.74 %	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KLF2	99.98 %	602016	*No OMIM phenotypes*

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KMT2A	99.97 %	159555	Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
KMT2D	99.98 %	602113	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186 (3), Autosomal dominant; Kabuki syndrome 1, 147920 (3), Autosomal dominant
KRAS	99.13 %	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)
LACC1	99.99 %	613409	Juvenile arthritis, 618795 (3), Autosomal recessive
LAMTOR2	99.92 %	610389	Immunodeficiency due to defect in MAPBP-interacting protein, 610798 (3), Autosomal recessive
LAT	99.85 %	602354	Immunodeficiency 52, 617514 (3), Autosomal recessive
LCK	99.56 %	153390	?Immunodeficiency 22, 615758 (3), Autosomal recessive
LCP1	99.98 %	153430	*No OMIM phenotypes*
LCP2	99.58 %	601603	?Immunodeficiency 81, 619374 (3), Autosomal recessive
LIG1	99.93 %	126391	Immunodeficiency 96, 619774 (3), Autosomal recessive
LIG4	100 %	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
LIPA	99.96 %	613497	Wolman disease, 620151 (3), Autosomal recessive; Cholesteryl ester storage disease, 278000 (3), Autosomal recessive
LPIN2	100 %	605519	Majeed syndrome, 609628 (3)
LRBA	99.76 %	606453	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive
LSM11	100 %	617910	?Aicardi-Goutieres syndrome 8, 619486 (3), Autosomal recessive
LY96	99.97 %	605243	*No OMIM phenotypes*
LYN	99.95 %	165120	Autoinflammatory disease, systemic, with vasculitis, 620376 (3), Autosomal dominant
LYST	99.87 %	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
MAGT1	99.54 %	300715	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3), X-linked recessive; Congenital disorder of glycosylation, type Icc, 301031 (3), X-linked recessive
MALT1	99.71 %	604860	Immunodeficiency 12, 615468 (3), Autosomal recessive
MAN1B1	99.94 %	604346	Rafiq syndrome, 614202 (3), Autosomal recessive
MAN2B1	99.99 %	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MAN2B2	99.98 %	618899	*No OMIM phenotypes*
MAP1LC3B2	100 %		*No OMIM phenotypes*
MAP3K14	99.98 %	604655	Immunodeficiency 112, 620449 (3), Autosomal recessive
MAPK8	99.64 %	601158	*No OMIM phenotypes*
MASP2	99.95 %	605102	MASP2 deficiency, 613791 (3), Autosomal recessive
MBL2	99.93 %	154545	{Chronic infections, due to MBL deficiency}, 614372 (3), Autosomal dominant
MCM10	99.99 %	609357	Immunodeficiency 80 with or without cardiomyopathy, 619313 (3), Autosomal recessive
MCM4	99.96 %	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MCTS1	99.4 %	300587	*No OMIM phenotypes*
MECOM	99.97 %	165215	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738 (3), Autosomal dominant
MEFV	100 %	608107	Neutrophilic dermatosis, acute febrile, 608068 (3), Autosomal dominant; Familial Mediterranean fever, AR, 249100 (3), Autosomal recessive; Familial Mediterranean fever, AD, 134610 (3), Autosomal dominant
MOGS	100 %	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
MPEG1	100 %	610390	Immunodeficiency 77, 619223 (3), Autosomal dominant
MPO	99.97 %	606989	{Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; Myeloperoxidase deficiency, 254600 (3), Autosomal recessive; {Lung cancer, protection against, in smokers} (3)
MRTFA	92.99 %	606078	?Immunodeficiency 66, 618847 (3), Autosomal recessive
MS4A1	99.6 %	112210	?Immunodeficiency, common variable, 5, 613495 (3), Autosomal recessive
MSH6	99.97 %	600678	Lynch syndrome 5, 614350 (3), Autosomal dominant; Mismatch repair cancer syndrome 3, 619097 (3), Autosomal recessive; {Endometrial cancer, familial}, 608089 (3), Somatic mutation, Autosomal dominant
MSN	99.98 %	309845	Immunodeficiency 50, 300988 (3), X-linked recessive
MTHFD1	100 %	172460	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 (3), Autosomal recessive
MVK	99.97 %	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
MYD88	99.99 %	602170	Macroglobulinemia, Waldenstrom, somatic, 153600 (3); Immunodeficiency 68, 612260 (3), Autosomal recessive
MYO5B	100 %	606540	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 10, 619868 (3), Autosomal recessive
MYSM1	94.16 %	612176	Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive
NBAS	99.86 %	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive
NBN	99.93 %	602667	Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NCF1	57.22 %	608512	Chronic granulomatous disease 1, autosomal recessive, 233700 (3), Autosomal recessive
NCF2	99.85 %	608515	Chronic granulomatous disease 2, autosomal recessive, 233710 (3), Autosomal recessive
NCF4	100 %	601488	Chronic granulomatous disease 3, autosomal recessive, 613960 (3), Autosomal recessive
NCKAP1L	99.77 %	141180	Immunodeficiency 72 with autoinflammation, 618982 (3), Autosomal recessive
NCSTN	99.82 %	605254	Acne inversa, familial, 1, 142690 (3), Autosomal dominant
NFAT5	99.93 %	604708	*No OMIM phenotypes*
NFATC1	100 %	600489	*No OMIM phenotypes*
NFE2L2	99.97 %	600492	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 (3), Autosomal dominant
NFKB1	99.8 %	164011	Immunodeficiency, common variable, 12, 616576 (3), Autosomal dominant
NFKB2	99.98 %	164012	Immunodeficiency, common variable, 10, 615577 (3), Autosomal dominant
NFKBIA	99.99 %	164008	Ectodermal dysplasia and immunodeficiency 2, 612132 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NHEJ1	99.91 %	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 (3)
NHP2	99.96 %	606470	Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive
NKX2-5	99.75 %	600584	Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Ventricular septal defect 3, 614432 (3), Autosomal dominant; Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant
NLRC4	99.95 %	606831	?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant; Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant
NLRP1	95.26 %	606636	{Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 (3); ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 (3), Autosomal recessive; Autoinflammation with arthritis and dyskeratosis, 617388 (3), Autosomal recessive, Autosomal dominant; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant
NLRP12	99.99 %	609648	Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant
NLRP3	100 %	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NLRP7	99.99 %	609661	Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive
NOD2	99.98 %	605956	Blau syndrome, 186580 (3), Autosomal dominant; {Yao syndrome}, 617321 (3), Multifactorial; {Inflammatory bowel disease 1, Crohn disease}, 266600 (3), Multifactorial
NOP10	99.99 %	606471	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400 (3), Autosomal dominant; ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425 (3), Autosomal recessive; ?Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
NOS2	96.11 %	163730	{Malaria, resistance to}, 611162 (3)
NPC1	99.99 %	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NRAS	99.66 %	164790	Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3)
NSMCE3	100 %	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive
NUDCD3	99.98 %	610296	*No OMIM phenotypes*
OAS1	99.96 %	164350	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinemia, 618042 (3), Autosomal dominant
ORAI1	99.63 %	610277	Immunodeficiency 9, 612782 (3), Autosomal recessive; Myopathy, tubular aggregate, 2, 615883 (3), Autosomal dominant
OSTM1	99.56 %	607649	Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive
OTULIN	99.95 %	615712	Autoinflammation, panniculitis, and dermatosis syndrome, 617099 (3), Autosomal recessive; {Immunodeficiency 107, susceptibility to invasive staphylococcus aureus infection}, 619986 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
P2RY8	100 %	300525	*No OMIM phenotypes*
PARN	99.75 %	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371 (3), Autosomal dominant
PAX1	100 %	167411	Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive
PAX5	99.82 %	167414	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3)
PCCA	99.9 %	232000	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	99.97 %	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PDCD1	99.99 %	600244	{Multiple sclerosis, disease progression, modifier of}, 126200 (3), Multifactorial; {Systemic lupus erythematosus, susceptibility to, 2}, 605218 (3)
PEPD	99.98 %	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
PGM3	99.94 %	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PI4KA	99.76 %	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
PIK3CD	99.99 %	602839	Immunodeficiency 14A, autosomal dominant, 615513 (3), Autosomal dominant; Immunodeficiency 14B, autosomal recessive, 619281 (3), Autosomal recessive; ?Roifman-Chitayat syndrome, digenic, 613328 (3), Digenic recessive
PIK3CG	99.72 %	601232	Immunodeficiency 97 with autoinflammation, 619802 (3), Autosomal recessive
PIK3R1	99.86 %	171833	Immunodeficiency 36, 616005 (3), Autosomal dominant; ?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; SHORT syndrome, 269880 (3), Autosomal dominant
PLCG2	99.99 %	600220	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 (3), Autosomal dominant; Familial cold autoinflammatory syndrome 3, 614468 (3), Autosomal dominant
PLEKHM1	99.77 %	611466	?Osteopetrosis, autosomal recessive 6, 611497 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 3, 618107 (3), Autosomal dominant
PLG	99.89 %	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Angioedema, hereditary, 4, 619360 (3), Autosomal dominant; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
PMS2	70.47 %	600259	Lynch syndrome 4, 614337 (3); Mismatch repair cancer syndrome 4, 619101 (3), Autosomal recessive
PMVK	99.6 %	607622	Porokeratosis 1, multiple types, 175800 (3), Autosomal dominant
PNP	100 %	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive
POLA1	99.57 %	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive
POLD1	99.96 %	174761	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant
POLD2	99.95 %	600815	*No OMIM phenotypes*
POLD3	99.97 %	611415	*No OMIM phenotypes*
POLE	99.99 %	174762	{Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive; IMAGE-I syndrome, 618336 (3), Autosomal recessive
POLE2	99.87 %	602670	*No OMIM phenotypes*

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
POLR3A	99.97 %	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
POLR3C	99.89 %	617454	*No OMIM phenotypes*
POLR3E	99.15 %	617815	*No OMIM phenotypes*
POLR3F	99.97 %	617455	?Immunodeficiency 101 (varicella zoster virus-specific), 619872 (3), Autosomal dominant
POMP	99.95 %	613386	Proteasome-associated autoinflammatory syndrome 2, 618048 (3), Autosomal dominant; Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive
POU2AF1	99.43 %	601206	*No OMIM phenotypes*
PRF1	100 %	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
PRIM1	99.07 %	176635	Primordial dwarfism-immunodeficiency-lipodystrophy syndrome, 620005 (3), Autosomal recessive
PRKCD	99.96 %	176977	Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive
PRKDC	99.93 %	600899	Immunodeficiency 26, with or without neurologic abnormalities, 615966 (3), Autosomal recessive
PSEN1	100 %	104311	Pick disease, 172700 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; ?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant
PSENEN	100 %	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant
PSMA3	99.96 %	176843	*No OMIM phenotypes*
PSMB10	99.98 %	176847	Proteasome-associated autoinflammatory syndrome 5, 619175 (3), Autosomal recessive
PSMB4	99.83 %	602177	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591 (3), Autosomal recessive
PSMB8	99.96 %	177046	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040 (3), Autosomal recessive
PSMB9	99.68 %	177045	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591 (3), Autosomal recessive
PSMG2	99.98 %	609702	?Proteasome-associated autoinflammatory syndrome 4, 619183 (3), Autosomal recessive
PSTPIP1	99.91 %	606347	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 (3), Autosomal dominant
PTCRA	99.99 %	606817	*No OMIM phenotypes*
PTEN	99.89 %	601728	{Glioma susceptibility 2}, 613028 (3), Autosomal dominant; {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
PTPN2	99.98 %	176887	*No OMIM phenotypes*
PTPRC	93.9 %	151460	Immunodeficiency 105, severe combined, 619924 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RAB27A	99.94 %	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
RAC2	99.99 %	602049	Immunodeficiency 73A with defective neutrophil chemotaxix 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
RAG1	100 %	179615	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (3)
RAG2	100 %	179616	Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive
RANBP2	99.37 %	601181	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033 (3), Autosomal dominant
RASGRP1	100 %	603962	Immunodeficiency 64, 618534 (3), Autosomal recessive
RBCK1	100 %	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
RC3H1	99.22 %	609424	?Immune dysregulation and systemic hyperinflammation syndrome, 618998 (3)
RECQL4	100 %	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive
REL	96.99 %	164910	Immunodeficiency 92, 619652 (3), Autosomal recessive
RELA	99.99 %	164014	Autoinflammatory disease, familial, Behcet-like-3, 618287 (3), Autosomal dominant
RELB	99.97 %	604758	?Immunodeficiency 53, 617585 (3), Autosomal recessive
RFX5	99.88 %	601863	Bare lymphocyte syndrome, type II, complementation group C, 209920 (3), Autosomal recessive; Bare lymphocyte syndrome, type II, complementation group E, 209920 (3), Autosomal recessive
RFXANK	100 %	603200	Bare lymphocyte syndrome, type II, complementation group B, 209920 (3), Autosomal recessive
RFXAP	99.98 %	601861	Bare lymphocyte syndrome, type II, complementation group D, 209920 (3), Autosomal recessive
RHBDF2	99.95 %	614404	Tylosis with esophageal cancer, 148500 (3), Autosomal dominant
RHOG	100 %	179505	*No OMIM phenotypes*
RHOH	99.99 %	602037	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307 (3), Autosomal recessive
RIPK1	99.93 %	603453	Immunodeficiency 57 with autoinflammation, 618108 (3), Autosomal recessive; Autoinflammation with episodic fever and lymphadenopathy, 618852 (3), Autosomal dominant
RIPK3	100 %	605817	*No OMIM phenotypes*
RMRP	100 %	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive
RNASEH2A	99.95 %	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
RNASEH2B	99.94 %	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
RNASEH2C	99.99 %	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
RNF168	99.97 %	612688	RIDDLE syndrome, 611943 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RNF31	100 %	612487	*No OMIM phenotypes*
RNU4ATAC	99.95 %	601428	Roifman syndrome, 616651 (3), Autosomal recessive; Lowry-Wood syndrome, 226960 (3), Autosomal recessive; Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive
RNU7-1	33.9 %	617876	Aicardi-Goutieres syndrome 9, 619487 (3), Autosomal recessive
RORC	99.42 %	602943	Immunodeficiency 42, 616622 (3), Autosomal recessive
RPSA	0 %	150370	Asplenia, isolated congenital, 271400 (3), Autosomal dominant
RTEL1	100 %	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 syndrome, telomere-related, 3, 616373 (3), Autosomal dominant
SAMD9	99.93 %	610456	Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive; Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 (3), Autosomal dominant; MIRAGE syndrome, 617053 (3), Autosomal dominant
SAMD9L	99.95 %	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
SAMHD1	99.98 %	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
SASH3	99.99 %	300441	Immunodeficiency 102, 301082 (3), X-linked recessive
SBDS	99.93 %	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
SCGN	99.98 %	609202	*No OMIM phenotypes*
SEC61A1	99.99 %	609213	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056 (3), Autosomal dominant
SEMA3E	99.13 %	608166	*No OMIM phenotypes*
SERPING1	100 %	606860	Angioedema, hereditary, 1 and 2, 106100 (3), Autosomal recessive, Autosomal dominant; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant
SGPL1	99.95 %	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive
SH2D1A	98.98 %	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
SH3BP2	100 %	602104	Cherubism, 118400 (3), Autosomal dominant
SH3KBP1	99.95 %	300374	?Immunodeficiency 61, 300310 (3), X-linked recessive
SKIV2L	99.98 %	600478	Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive
SLC29A3	99.98 %	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
SLC35C1	100 %	605881	Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive
SLC37A4	99.9 %	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
SLC39A4	100 %	607059	Acrodermatitis enteropathica, 201100 (3), Autosomal recessive
SLC39A7	100 %	601416	Agammaglobulinemia 9, autosomal recessive, 619693 (3), Autosomal recessive
SLC46A1	100 %	611672	Folate malabsorption, hereditary, 229050 (3), Autosomal recessive
SLC7A7	99.99 %	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
SLC9A3	100 %	182307	Diarrhea 8, secretory sodium, congenital, 616868 (3), Autosomal recessive
SLCO2A1	99.99 %	601460	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 (3), Autosomal dominant; Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SMARCAL1	99.97 %	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
SMARCD2	99.99 %	601736	Specific granule deficiency 2, 617475 (3), Autosomal recessive
SNORA31	99.79 %	619378	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 10}, 619396 (3), Autosomal dominant
SNX10	99.96 %	614780	Osteopetrosis, autosomal recessive 8, 615085 (3), Autosomal recessive
SOCS1	99.98 %	603597	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375 (3), Autosomal dominant
SOCS4	99.92 %	616337	*No OMIM phenotypes*
SP110	99.99 %	604457	{Mycobacterium tuberculosis, susceptibility to}, 607948 (3); Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive
SPI1	99.81 %	165170	Agammaglobulinemia 10, autosomal dominant, 619707 (3), Autosomal dominant
SPINK5	99.91 %	605010	Netherton syndrome, 256500 (3), Autosomal recessive
SPPL2A	99.88 %	608238	Immunodeficiency 86, mycobacteriosis, 619549 (3), Autosomal recessive
SRP54	99.83 %	604857	Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant
SRP68	99.99 %	604858	*No OMIM phenotypes*
SRP72	99.91 %	602122	Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant
STAT1	99.83 %	600555	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 (3), Autosomal dominant; Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 (3), Autosomal dominant; Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 (3), Autosomal recessive
STAT2	99.89 %	600556	Pseudo-TORCH syndrome 3, 618886 (3), Autosomal recessive; Immunodeficiency 44, 616636 (3), Autosomal recessive
STAT3	99.97 %	102582	Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant
STAT4	99.77 %	600558	Disabling pansclerotic morphea of childhood, 620443 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to, 11}, 612253 (3)
STAT5B	99.5 %	604260	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 (3), Autosomal recessive; Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 (3), Autosomal dominant; Leukemia, acute promyelocytic, somatic, 102578 (3)
STAT6	99.87 %	601512	*No OMIM phenotypes*
STIM1	99.99 %	605921	Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive
STING1	99.87 %	612374	STING-associated vasculopathy, infantile-onset, 615934 (3), Autosomal dominant
STK4	99.91 %	604965	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 (3), Autosomal recessive
STN1	99.88 %	613128	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive
STX11	100 %	605014	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive
STXBP2	100 %	601717	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101 (3), Autosomal recessive
STXBP3	85.74 %	608339	*No OMIM phenotypes*
SYK	99.96 %	600085	Immunodeficiency 82 with systemic inflammation, 619381 (3), Autosomal dominant
TAFAZZIN	99.98 %	300394	Barth syndrome, 302060 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TAP1	99.97 %	170260	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
TAP2	99.94 %	170261	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 (3), Autosomal recessive
TAPBP	99.98 %	601962	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
TBK1	99.07 %	604834	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 (3), Autosomal dominant
TBX1	99.95 %	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
TBX2	99.97 %	600747	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223 (3), Autosomal dominant
TBX21	99.99 %	604895	Asthma and nasal polyps, 208550 (3), Autosomal recessive; ?Immunodeficiency 88, 619630 (3), Autosomal recessive; {Asthma, aspirin-induced, susceptibility to}, 208550 (3), Autosomal recessive
TCF3	100 %	147141	Agammaglobulinemia 8B, autosomal recessive, 619824 (3), Autosomal recessive; Agammaglobulinemia 8A, autosomal dominant, 616941 (3), Autosomal dominant
TCIRG1	99.99 %	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive
TCN2	100 %	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
TERC	98.59 %	602322	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant
TERT	100 %	187270	Dyskeratosis congenita, autosomal dominant 2, 613989 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 4, 613989 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 9}, 615134 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Somatic mutation, Autosomal dominant
TET2	99.99 %	612839	Myelodysplastic syndrome, somatic, 614286 (3); Immunodeficiency 75, 619126 (3), Autosomal recessive
TFRC	99.87 %	190010	Immunodeficiency 46, 616740 (3), Autosomal recessive
TGFB1	100 %	190180	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 (3), Autosomal recessive; Camurati-Engelmann disease, 131300 (3), Autosomal dominant; {Cystic fibrosis lung disease, modifier of}, 219700 (3), Autosomal recessive
TGFBR1	99.94 %	190181	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant
TGFBR2	99.98 %	190182	Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3)
THBD	100 %	188040	Thrombophilia 12 due to thrombomodulin defect, 614486 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant
TICAM1	99.99 %	607601	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850 (3), Autosomal recessive, Autosomal dominant
TINF2	100 %	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TIRAP	100 %	606252	{Malaria, protection against}, 611162 (3); {Tuberculosis, protection against}, 607948 (3); {Bacteremia, protection against}, 614382 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TLR3	99.99 %	603029	{HIV1 infection, resistance to}, 609423 (3); {Immunodeficiency 83, susceptibility to viral infections}, 613002 (3), Autosomal recessive, Autosomal dominant
TLR4	99.99 %	603030	*No OMIM phenotypes*
TLR7	99.98 %	300365	Immunodeficiency 74, COVID19-related, X-linked, 301051 (3), X-linked recessive; Systemic lupus erythematosus 17, 301080 (3), X-linked dominant
TLR8	99.98 %	300366	Immunodeficiency 98 with autoinflammation, X-linked, 301078 (3), X-linked, Somatic mosaicism
TMC6	100 %	605828	Epidermodysplasia verruciformis, 226400 (3), Autosomal recessive
TMC8	99.92 %	605829	Epidermodysplasia verruciformis 2, 618231 (3), Autosomal recessive
TNFAIP3	99.94 %	191163	Autoinflammatory syndrome, familial, Behcet-like 1, 616744 (3), Autosomal dominant
TNFRSF11A	100 %	603499	Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive; {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant; Osteolysis, familial expansile, 174810 (3), Autosomal dominant
TNFRSF13B	99.43 %	604907	Immunodeficiency, common variable, 2, 240500 (3), Autosomal recessive, Autosomal dominant; Immunoglobulin A deficiency 2, 609529 (3)
TNFRSF13C	99.99 %	606269	Immunodeficiency, common variable, 4, 613494 (3), Autosomal recessive
TNFRSF1A	100 %	191190	{Multiple sclerosis, susceptibility to, 5}, 614810 (3); Periodic fever, familial, 142680 (3), Autosomal dominant
TNFRSF4	100 %	600315	?Immunodeficiency 16, 615593 (3), Autosomal recessive
TNFRSF9	99.99 %	602250	Immunodeficiency 109 with lymphoproliferation, 620282 (3), Autosomal recessive
TNFSF11	99.89 %	602642	Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive
TNFSF12	100 %	602695	*No OMIM phenotypes*
TNFSF13	100 %	604472	*No OMIM phenotypes*
TOP2B	99.72 %	126431	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296 (3), Autosomal dominant
TPP2	99.89 %	190470	Immunodeficiency 78 with autoimmunity and developmental delay, 619220 (3), Autosomal recessive
TRAC	100 %	186880	Immunodeficiency 7, TCR-alpha/beta deficient, 615387 (3), Autosomal recessive
TRAF3	99.97 %	601896	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849 (3), Autosomal dominant
TRAF3IP2	100 %	607043	?Candidiasis, familial, 8, 615527 (3), Autosomal recessive; {Psoriasis susceptibility 13}, 614070 (3)
TREX1	100 %	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
TRIM22	100 %	606559	*No OMIM phenotypes*
TRNT1	99.97 %	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
TRPM4	99.99 %	606936	Progressive familial heart block, type IB, 604559 (3), Autosomal dominant; Erythrokeratodermia variabilis et progressiva 6, 618531 (3), Autosomal dominant
TTC37	99.82 %	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
TTC7A	99.77 %	609332	Gastrointestinal defects and immunodeficiency syndrome, 243150 (3), Autosomal recessive
TYK2	99.99 %	176941	Immunodeficiency 35, 611521 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
UBA1	99.93 %	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive; VEXAS syndrome, somatic, 301054 (3)
UHRF1	99.83 %	607990	*No OMIM phenotypes*
UNC13D	100 %	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3), Autosomal recessive
UNC93B1	99.75 %	608204	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551 (3), Autosomal recessive
UNG	100 %	191525	Immunodeficiency with hyper IgM, type 5, 608106 (3), Autosomal recessive
USB1	89.62 %	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive
USP18	93.05 %	607057	Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive
VPS13B	99.9 %	607817	Cohen syndrome, 216550 (3), Autosomal recessive
VPS45	93.94 %	610035	Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive
WAS	99.9 %	300392	Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive; Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive; Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive
WDR1	99.99 %	604734	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550 (3), Autosomal recessive
WIPF1	99.87 %	602357	Wiskott-Aldrich syndrome 2, 614493 (3), Autosomal recessive
WRAP53	100 %	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
XIAP	99.36 %	300079	Lymphoproliferative syndrome, X-linked, 2, 300635 (3), X-linked recessive
ZAP70	99.95 %	176947	Immunodeficiency 48, 269840 (3), Autosomal recessive; Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive
ZBTB24	99.99 %	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
ZNF341	100 %	618269	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282 (3), Autosomal recessive
ZNFX1	99.99 %	618931	Immunodeficiency 91 and hyperinflammation, 619644 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2023-07-31**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

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.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.