PRIMARY IMMUNODEFICIENCIES GENE PANEL DG 2.16 (394 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ACD	159,6	100.0%	100.0%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACP5	172,6	100.0%	99.6%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	80,5	100.0%	99.7%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADA	104,6	100.0%	99.6%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	83,5	99.9%	97.6%	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	119	99.8%	98.6%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAR	109,2	99.9%	99.3%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	142,7	100.0%	100.0%	Aspartylglucosaminuria, 208400
AICDA	128,4	100.0%	99.5%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	102,3	100.0%	99.9%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	96,1	98.7%	94.4%	Reticular dysgenesis, 267500
ALG13	77,3	98.5%	92.1%	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884
AP1S3	110,7	90.5%	90.4%	{Psoriasis 15, pustular, susceptibility to}, 616106
AP3B1	112,1	99.5%	96.5%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	125,2	98.4%	97.9%	?Hermansky-Pudlak syndrome 10, 617050
APOL1	146,1	100.0%	100.0%	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
ARHGEF1	105,4	100.0%	99.1%	No OMIM phenotype
ARPC1B	139,6	100.0%	100.0%	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
ATM	110,9	99.6%	97.2%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, 0 Lymphoma, mantle cell, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0 {Breast cancer, susceptibility to}, 114480

ATP6AP1	105,6	99.8%	96.9%	Immunodeficiency 47, 300972
B2M	198,9	100.0%	99.8%	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
BACH2	159	100.0%	99.8%	Immunodeficiency 60, 618394
BCL10	126,9	100.0%	100.0%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic}, 273300 {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic}, 0
BCL11B	127,4	99.9%	98.0%	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BLK	127,9	100.0%	100.0%	Maturity-onset diabetes of the young, type 11, 613375
BLM	111	99.6%	98.0%	Bloom syndrome, 210900
BLNK	91,4	97.0%	93.1%	?Agammaglobulinemia 4, 613502
BLOC1S6	101,1	99.2%	95.1%	?Hermansky-pudlak syndrome 9, 614171
BTK	98	99.9%	99.0%	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
C17orf62	NC	NC	NC	No OMIM phenotype
C1QA	196,1	100.0%	100.0%	C1q deficiency, 613652
C1QB	161,5	100.0%	100.0%	C1q deficiency, 613652
C1QC	187	100.0%	99.6%	C1q deficiency, 613652
C1R	151	100.0%	100.0%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	96,8	99.8%	97.8%	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	126,3	100.0%	100.0%	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C3	141,6	100.0%	99.4%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	86,4	98.5%	96.1%	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	83	98.7%	96.6%	C4B deficiency, 614379
C5	120,9	99.5%	97.7%	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C6	139,1	100.0%	99.6%	C6 deficiency, 612446 Combined C6/C7 deficiency, 0

C7	113,1	99.7%	97.3%	C7 deficiency, 610102
C8A	105,6	100.0%	99.4%	C8 deficiency, type I, 613790
C8B	105,6	99.9%	98.7%	C8 deficiency, type II, 613789
C8G	164,2	100.0%	100.0%	No OMIM phenotype
C9	121	99.9%	99.3%	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
CA2	137,4	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CARD11	138,2	100.0%	99.6%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206 Immunodeficiency 11B with atopic dermatitis, 617638
CARD14	124,5	100.0%	99.2%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	133,2	100.0%	99.5%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	137,3	98.4%	96.6%	Immunodeficiency 58, 618131
CASP10	106,2	99.8%	98.2%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP8	128,1	95.6%	95.2%	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CAVIN1	174,1	100.0%	100.0%	Lipodystrophy, congenital generalized, type 4, 613327
CCBE1	75,3	99.8%	99.1%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD19	108,8	100.0%	99.9%	Immunodeficiency, common variable, 3, 613493
CD247	95,5	100.0%	99.5%	?Immunodeficiency 25, 610163
CD27	105,3	100.0%	100.0%	Lymphoproliferative syndrome 2, 615122
CD3D	138,9	100.0%	99.9%	Immunodeficiency 19, 615617
CD3E	125,9	100.0%	98.9%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	136,6	100.0%	100.0%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	147,8	100.0%	99.9%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	109,6	97.2%	87.3%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	125,5	99.7%	98.7%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD55	134,6	95.5%	90.4%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 [Blood group Cromer], 613793
CD59	149,6	93.5%	85.8%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300

CD70	109	100.0%	99.2%	Lymphoproliferative syndrome 3, 618261
CD79A	133,5	100.0%	99.3%	Agammaglobulinemia 3, 613501
CD79B	194,3	100.0%	100.0%	Agammaglobulinemia 6, 612692
CD81	158,3	100.0%	100.0%	Immunodeficiency, common variable, 6, 613496
CD8A	150,8	100.0%	99.9%	CD8 deficiency, familial, 608957
CDCA7	112	100.0%	99.5%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDKN2B	121,9	100.0%	100.0%	No OMIM phenotype Renal cell carcinoma (Jafri (2015) Cancer Discov 5, 723) Multiple endocrine neoplasia 1 (Agarwal (2009) J Clin Endocrinol Metab 94, 1826) ?Melanoma (Foley (2015) EBioMedicine 2,74) ?Parathyroid adenoma (Costa-Guda (2013) Horm
CEBPE	99,1	100.0%	99.9%	Specific granule deficiency, 245480
CFB	119,4	100.0%	99.9%	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFD	113,8	96.9%	89.7%	Complement factor D deficiency, 613912
CFH	155,4	99.4%	97.9%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	158,4	93.6%	90.8%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR2	134,4	99.6%	96.8%	No OMIM phenotype
CFHR3	98,4	93.8%	91.6%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR4	121,1	100.0%	99.8%	No OMIM phenotype
CFHR5	96,6	99.8%	97.5%	Nephropathy due to CFHR5 deficiency, 614809
CFI	139	99.5%	97.0%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFP	98,4	99.7%	97.4%	Properdin deficiency, X-linked, 312060
CFTR	113,5	99.4%	97.4%	Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF, 0 {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal}, 0 {Pancreatitis, hereditary}, 167800

CHD7	137	99.9%	99.4%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CIITA	148,8	100.0%	99.9%	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CLCN7	146,7	99.8%	98.7%	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
CLEC4D	136,5	100.0%	99.9%	No OMIM phenotype
CLEC7A	148,5	100.0%	99.9%	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
CLPB	125,6	99.8%	97.9%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COLEC11	180,6	100.0%	100.0%	3MC syndrome 2, 265050
COPA	107,6	100.0%	99.2%	{Autoimmune interstitial lung, joint, and kidney disease}, 616414
CORO1A	150,8	99.9%	98.5%	Immunodeficiency 8, 615401
CR2	131,7	100.0%	99.9%	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CREBBP	110,7	99.4%	97.0%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CSF2RA	53,7	90.0%	88.0%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	123,2	99.8%	98.4%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	105,4	99.7%	98.6%	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	105,5	100.0%	99.3%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	141	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type V, 616100 {Celiac disease, susceptibility to, 3}, 609755 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Hashimoto thyroiditis}, 140300 {Systemic lupus erythematosus, susceptibility to}, 152700
CTPS1	108,8	100.0%	99.9%	Immunodeficiency 24, 615897
CTSC	116,2	100.0%	100.0%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CXCR4	122,8	100.0%	100.0%	Myelokathexis, isolated, 0 WHIM syndrome, 193670
CYBA	110,5	96.7%	86.9%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	99,1	99.9%	99.0%	Chronic granulomatous disease, X-linked, 306400 Immunodeficiency 34, mycobacteriosis, X-linked, 300645
DCLRE1C	138,9	99.9%	97.2%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450

DDX58	112,3	99.8%	99.1%	Singleton-Merten syndrome 2, 616298
DHFR	50	94.1%	83.1%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	91,2	99.8%	97.7%	Dyskeratosis congenita, X-linked, 305000
DNASE1	168,5	100.0%	100.0%	{Systemic lupus erythematosus, susceptibility to}, 152700
DNASE2	93,2	99.3%	96.6%	No OMIM phenotype
DNMT3B	116,4	100.0%	99.9%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	119,5	100.0%	99.5%	Immunodeficiency 40, 616433
DOCK8	112,1	100.0%	99.6%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	141,5	100.0%	99.3%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	91,7	100.0%	98.9%	No OMIM phenotype ?Immunodeficiency, primary, modifier of (Stray-Pedersen (2017) J Allergy Clin Immunol 139,232) ?Hypogammaglobulinaemia (Stewart (2005) Curr Opin Allergy Clin Immunol 5,510)
EPG5	110,3	99.3%	97.9%	Vici syndrome, 242840
ERCC2	128	100.0%	99.8%	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	92	99.9%	98.4%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
EXTL3	184,1	100.0%	100.0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
F12	151,3	99.9%	99.5%	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
FAAP24	112,6	99.8%	97.1%	No OMIM phenotype
FADD	181,7	100.0%	100.0%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAS	226	99.9%	99.6%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0 {Autoimmune lymphoproliferative syndrome}, 601859
FASLG	82	100.0%	99.0%	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FAT4	190,3	100.0%	99.9%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FCGR1A	53,8	47.6%	44.9%	[IgG receptor I, phagocytic, familial deficiency of], 0
FCGR2A	164,6	100.0%	100.0%	{Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162 {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700
FCGR2B	119,7	99.9%	96.9%	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700

FCGR3A	163,2	99.6%	96.7%	Immunodeficiency 20, 615707
FCGR3B	137,4	98.8%	97.1%	Neutropenia, alloimmune neonatal, 0
FCN3	124,7	100.0%	100.0%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	144,9	100.0%	99.9%	Leukocyte adhesion deficiency, type III, 612840
FOXN1	133	100.0%	99.5%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	115,6	99.1%	94.8%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FPR1	148,2	100.0%	99.9%	No OMIM phenotype {Periodontitis, aggressive, association with} (Gunji (2007) Biochem Biophys Res Commun 364,7) {Earlier onset of Alzheimer disease, association with} (Velez (2016) Am J Med Genet B Neuropsychiatr Genet 171,1116)
G6PC	146,8	100.0%	99.9%	Glycogen storage disease Ia, 232200
G6PC3	114,6	100.0%	100.0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	114,4	99.5%	97.4%	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GATA2	115	100.0%	99.0%	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GFI1	105,7	100.0%	100.0%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 ?Neutropenia, severe congenital 2, autosomal dominant, 613107
GINS1	122,7	98.1%	90.6%	Immunodeficiency 55, 617827
GJC2	45,3	92.6%	75.4%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GRHL2	116,8	100.0%	100.0%	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GTF2H5	81,8	99.9%	95.9%	Trichothiodystrophy 3, photosensitive, 616395
HAX1	137,4	100.0%	100.0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HELLS	102,8	98.8%	92.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HMOX1	137,4	96.5%	90.7%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HYOU1	130,8	99.9%	99.5%	?Immunodeficiency 59 and hypoglycemia, 233600
ICOS	156,4	100.0%	99.9%	Immunodeficiency, common variable, 1, 607594
IFIH1	110,8	99.8%	98.2%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNAR2	129,5	99.5%	96.9%	?Immunodeficiency 45, 616669

				{Hepatitis B virus, susceptibility to}, 610424
IFNGR1	145	99.9%	99.3%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948
IFNGR2	125,2	97.0%	93.5%	Immunodeficiency 28, mycobacteriosis, 614889
IGHM	170	100.0%	100.0%	Agammaglobulinemia 1, 601495
IGLL1	92,2	100.0%	99.6%	Agammaglobulinemia 2, 613500
IKBKB	110	99.3%	96.4%	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	60,1	88.1%	78.8%	Ectodermal dysplasia and immunodeficiency 1, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IKZF1	181,7	100.0%	100.0%	Immunodeficiency, common variable, 13, 616873
IL10	95,6	100.0%	97.0%	{Graft-versus-host disease, protection against}, 614395 {HIV-1, susceptibility to}, 609423 {Rheumatoid arthritis, progression of}, 180300
IL10RA	144,4	100.0%	100.0%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	130,1	100.0%	99.3%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL12B	94,8	99.9%	97.0%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	112,9	98.0%	95.6%	Immunodeficiency 30, 614891
IL17F	73,1	99.2%	94.0%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	149,1	100.0%	100.0%	Immunodeficiency 51, 613953
IL17RC	123,6	100.0%	100.0%	Candidiasis, familial, 9, 616445
IL1RN	139,3	100.0%	99.7%	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL2	70,6	97.6%	90.5%	Severe combined immunodeficiency due to IL2 deficiency
IL21	74,9	99.8%	95.0%	?Immunodeficiency, common variable, 11, 615767
IL21R	145	100.0%	100.0%	Immunodeficiency 56, 615207 [IgE, elevated level of], 147050

IL2RA	100,6	99.9%	98.7%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	59,3	99.3%	94.0%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL36RN	92,8	100.0%	100.0%	Psoriasis 14, pustular, 614204
IL7R	114,1	100.0%	99.9%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INO80	96	99.8%	98.4%	No OMIM phenotype
INSR	116,4	99.0%	95.1%	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
IRAK1	81,6	99.9%	97.2%	No OMIM phenotype {Atherothrombotic cerebral infarction, association with} (Yamada (2008) Stroke 39,2211) Sepsis, susceptibility,association with} (Fang (2011) Chin Med J (Engl) 124, 2248)
IRAK4	103,9	99.7%	96.5%	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676
IRF2BP2	86,8	100.0%	99.3%	?Immunodeficiency, common variable, 14, 617765
IRF3	137	100.0%	99.6%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532
IRF4	196,4	100.0%	99.9%	[Skin/hair/eye pigmentation, variation in, 8], 611724
IRF7	152,6	100.0%	99.8%	?Immunodeficiency 39, 616345
IRF8	111,8	99.9%	98.6%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
ISG15	184,2	100.0%	100.0%	Immunodeficiency 38, 616126
ITCH	116,3	95.4%	94.6%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGB2	156,8	100.0%	100.0%	Leukocyte adhesion deficiency, 116920
ITK	103,1	99.9%	99.1%	Lymphoproliferative syndrome 1, 613011
JAGN1	118,5	100.0%	100.0%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	105,4	100.0%	99.4%	No OMIM phenotype
JAK2	103,5	97.6%	95.0%	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600880
JAK3	122,1	98.8%	97.2%	SCID, autosomal recessive, T-negative/B-positive type, 600802
KDM6A	97,7	95.3%	87.8%	Kabuki syndrome 2, 300867

KMT2D	136,2	100.0%	99.7%	Kabuki syndrome 1, 147920
LACC1	144,5	100.0%	99.2%	No OMIM phenotype
LAMTOR2	172,2	100.0%	100.0%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAT	114,4	100.0%	99.3%	Immunodeficiency 52, 617514
LCK	148,6	99.3%	97.3%	?Immunodeficiency 22, 615758
LIG1	105,5	100.0%	99.2%	DNA ligase I deficiency
LIG4	173,4	100.0%	99.8%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LPIN2	97,8	100.0%	99.6%	Majeed syndrome, 609628
LRBA	129,8	100.0%	99.6%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	223,3	100.0%	100.0%	?Agammaglobulinemia 5, 613506
LTBP3	147,5	100.0%	99.6%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LYST	136,3	99.4%	97.8%	Chediak-Higashi syndrome, 214500
MAGT1	96,8	98.2%	96.3%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAL2	162	100.0%	100.0%	No OMIM phenotype
MALT1	129,4	93.0%	89.4%	Immunodeficiency 12, 615468
MAN2B1	128,6	99.9%	98.6%	Mannosidosis, alpha-, types I and II, 248500
MANBA	118,3	99.5%	97.5%	Mannosidosis, beta, 248510
MAP3K14	120,3	99.3%	99.2%	No OMIM phenotype
MASP2	121,9	100.0%	99.5%	MASP2 deficiency, 613791
MBL2	94,4	100.0%	99.7%	{Chronic infections, due to MBL deficiency}, 614372
MC2R	148,3	100.0%	99.2%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM4	133,1	100.0%	99.5%	Immunodeficiency 54, 609981
MEFV	126,8	98.6%	96.5%	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MKL1	NC	NC	NC	Megakaryoblastic leukemia, acute, 0
MOGS	141	100.0%	100.0%	Congenital disorder of glycosylation, type IIb, 606056
MRE11	49,7	97.3%	86.0%	Ataxia-telangiectasia-like disorder 1, 604391
MS4A1	127,6	99.9%	98.8%	Immunodeficiency, common variable, 5, 613495
MSN	66,9	97.5%	91.7%	Immunodeficiency 50, 300988
MTHFD1	115,4	99.8%	97.4%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MVK	121,4	91.0%	90.5%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900

MYD88	194,7	100.0%	99.8%	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
MYSM1	111	99.8%	98.4%	Bone marrow failure syndrome 4, 618116
NBAS	138,5	99.9%	99.1%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBN	93,8	99.8%	98.4%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCF1	23,1	27.8%	22.5%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	109,2	99.8%	98.2%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	148,6	100.0%	100.0%	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	92,7	100.0%	99.6%	Acne inversa, familial, 1, 142690
NFAT5	174,3	99.8%	98.7%	No OMIM phenotype
NFKB1	93	99.9%	98.7%	Immunodeficiency, common variable, 12, 616576
NFKB2	135	99.1%	96.5%	Immunodeficiency, common variable, 10, 615577
NFKBIA	134,6	95.3%	89.4%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHEJ1	58,5	99.7%	92.8%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	121,9	100.0%	99.2%	Dyskeratosis congenita, autosomal recessive 2, 613987
NLRC4	159,4	100.0%	99.9%	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050
NLRP1	117,7	99.5%	97.6%	Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP12	161,7	100.0%	100.0%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	134,6	100.0%	99.9%	CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Muckle-Wells syndrome, 191900
NLRP7	124,5	99.9%	98.8%	Hydatidiform mole, recurrent, 1, 231090
NOD2	125,3	100.0%	99.9%	Blau syndrome, 186580 {Inflammatory bowel disease 1, Crohn disease}, 266600 {Psoriatic arthritis, susceptibility to}, 607507 {Yao syndrome}, 617321
NOP10	120,5	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	145,5	100.0%	100.0%	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470

				Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
NSMCE3	194	100.0%	100.0%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
ORAI1	198,9	99.8%	98.2%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
OSTM1	109,3	98.2%	92.5%	Osteopetrosis, autosomal recessive 5, 259720
OTULIN	132,6	98.7%	95.2%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
PARN	127,3	99.9%	99.5%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX5	105,6	99.2%	96.0%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PBX1	111,7	99.9%	98.2%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	99,2	99.3%	95.5%	Propionicacidemia, 606054
PCCB	111,8	99.3%	96.9%	Propionicacidemia, 606054
PEPD	117,4	100.0%	99.6%	Prolidase deficiency, 170100
PGM3	149,3	99.9%	99.6%	Immunodeficiency 23, 615816
PIGA	70,9	92.9%	84.0%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIK3CD	158,2	99.5%	97.8%	Immunodeficiency 14, 615513
PIK3R1	124,3	99.9%	98.9%	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PLCG2	105,8	100.0%	99.3%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEKHM1	127,8	100.0%	99.9%	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLG	93,4	87.8%	86.8%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PMM2	127,7	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PNP	108,6	100.0%	99.5%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POLA1	104	99.3%	95.4%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220
POLE2	68,8	97.2%	88.9%	No OMIM phenotype

POMP	124,6	99.9%	97.6%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 Proteasome-associated autoinflammatory syndrome 2, 618048
POT1	97,7	99.9%	98.5%	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
PRF1	138,1	91.2%	90.6%	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKCD	164,7	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	97,9	99.3%	96.5%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	111,6	100.0%	99.9%	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PSENEN	90,1	100.0%	100.0%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMA3	73	99.2%	95.4%	No OMIM phenotype
PSMB4	119,2	100.0%	99.9%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	113,5	100.0%	98.8%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	81,5	99.9%	97.7%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMG2	121,1	100.0%	99.3%	No OMIM phenotype
PSTPIP1	103,8	99.9%	98.5%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN22	127,8	99.6%	95.9%	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700
PTPRC	100,6	98.7%	93.9%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitis C virus, susceptibility to}, 609532
RAB27A	126,1	100.0%	99.8%	Griscelli syndrome, type 2, 607624
RAC2	100,6	100.0%	99.4%	Neutrophil immunodeficiency syndrome, 608203
RAG1	150,9	100.0%	100.0%	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAG2	186,2	100.0%	100.0%	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RANBP2	102,3	50.2%	49.3%	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033

RASGRP1	110,1	100.0%	99.5%	No OMIM phenotype
RASGRP2	102,5	100.0%	99.7%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	107,9	100.0%	99.2%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RECQL4	159,9	100.0%	99.8%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RELB	108	99.2%	92.7%	?Immunodeficiency 53, 617585
RFX5	109	99.8%	97.5%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	123,1	100.0%	99.3%	MHC class II deficiency, complementation group B, 209920
RFXAP	116,5	100.0%	99.3%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RHOH	123,4	100.0%	100.0%	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307
RIPK1	103,6	99.9%	98.7%	Immunodeficiency 57, 618108
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	129,8	100.0%	99.7%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	100,8	98.9%	95.2%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	281,7	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
RNF168	182	100.0%	99.6%	RIDDLE syndrome, 611943
RNF31	148,3	100.0%	99.6%	No OMIM phenotype Autoinflammation, immunodeficiency, amylopectinosis and lymphangiectasia (Boisson (2015) J Exp Med 212,939)
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
RORC	118,6	100.0%	100.0%	Immunodeficiency 42, 616622
RPSA	64	100.0%	99.4%	Asplenia, isolated congenital, 271400
RSPH9	131,2	99.7%	97.1%	Ciliary dyskinesia, primary, 12, 612650
RTEL1	131,1	99.7%	97.7%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
SAMD9	163,9	100.0%	99.9%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	171,8	100.0%	100.0%	Ataxia-pancytopenia syndrome, 159550
SAMHD1	133,4	99.8%	98.5%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SBDS	166,2	100.0%	100.0%	Shwachman-Diamond syndrome, 260400

				{Aplastic anemia, susceptibility to}, 609135
SEMA3E	130,9	100.0%	99.6%	?CHARGE syndrome, 214800
SERAC1	111	99.7%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	96,7	99.5%	96.7%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SH2B3	108,4	99.9%	97.6%	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SH2D1A	108,9	97.8%	92.4%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	139,3	91.9%	91.4%	Cherubism, 118400
SKIV2L	138,4	100.0%	99.7%	Trichohepatoenteric syndrome 2, 614602
SLC29A3	173,3	100.0%	99.5%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	127,1	100.0%	99.4%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	187,8	100.0%	99.8%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	114,3	100.0%	99.6%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	114,2	100.0%	99.0%	Acrodermatitis enteropathica, 201100
SLC46A1	111,1	99.9%	98.4%	Folate malabsorption, hereditary, 229050
SMARCAL1	113,2	100.0%	99.6%	Schimke immunoosseous dysplasia, 242900
SMARCD2	92,3	87.3%	85.8%	Specific granule deficiency 2, 617475
SNX10	131,4	96.2%	95.7%	Osteopetrosis, autosomal recessive 8, 615085
SOCS4	223,3	100.0%	99.1%	No OMIM phenotype Autoimmunity (Arts (2015) J Intern Med epub,epub)
SP110	109,3	100.0%	99.8%	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SPINK5	128	99.9%	99.5%	Netherton syndrome, 256500
SPPL2A	57,9	85.7%	70.8%	No OMIM phenotype
STAT1	117,8	99.6%	97.7%	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	110	100.0%	99.8%	Immunodeficiency 44, 616636
STAT3	103,2	100.0%	99.0%	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT4	144,2	99.9%	99.4%	{Systemic lupus erythematosus, susceptibility to, 11}, 612253
STAT5B	114,1	99.8%	97.8%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578

STAT6	112,8	100.0%	99.8%	No OMIM phenotype {Schistosomiasis infection, association with} (He (2008) Genes Immun 9, 195) {Atopic asthma, association with} (Gao (2004) J Med Genet 41,535)
STIM1	120,7	99.8%	96.8%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STK4	122,5	100.0%	99.7%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	298,3	100.0%	100.0%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	100,2	83.7%	80.4%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TAP1	117,7	99.9%	97.3%	Bare lymphocyte syndrome, type I, 604571
TAP2	93	99.6%	98.4%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	116,6	96.6%	96.5%	Bare lymphocyte syndrome, type I, 604571
TAZ	114,5	99.3%	95.8%	Barth syndrome, 302060
TBX1	101,2	93.0%	86.9%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TCF3	91,1	99.1%	95.6%	Agammaglobulinemia 8, autosomal dominant, 616941
TCIRG1	131,4	99.2%	96.6%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	148,5	100.0%	100.0%	Transcobalamin II deficiency, 275350
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERT	144,1	99.7%	97.6%	{Dyskeratosis congenita, autosomal dominant 2}, 613989 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742
TFRC	132	99.9%	99.0%	Immunodeficiency 46, 616740
THBD	181,1	100.0%	100.0%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TICAM1	120	100.0%	99.9%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850
TINF2	177,1	100.0%	100.0%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TIRAP	138,1	100.0%	99.8%	{Bacteremia, protection against}, 614382 {Malaria, protection against}, 611162 {Pneumococcal disease, invasive, protection against}, 610799

				{Tuberculosis, protection against}, 607948
TLR3	167,2	100.0%	99.4%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002 {HIV1 infection, resistance to}, 609423
TLR4	129,7	100.0%	99.7%	Endotoxin hyporesponsiveness {Colorectal cancer, susceptibility to}, 114500 {Macular degeneration, age-related, 10}, 611488
TMC6	91,1	100.0%	99.7%	Epidermodysplasia verruciformis, 226400
TMC8	133	100.0%	99.7%	Epidermodysplasia verruciformis 2, 618231
TMEM173	95,3	99.1%	94.0%	STING-associated vasculopathy, infantile-onset, 615934
TNFAIP3	149,4	100.0%	100.0%	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF11A	131	96.1%	95.2%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF13B	100,8	100.0%	99.7%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	98,4	96.2%	82.4%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	106,8	92.5%	89.7%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFRSF4	79,8	99.5%	96.8%	?Immunodeficiency 16, 615593
TNFSF11	129,8	100.0%	100.0%	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	90,7	99.9%	98.8%	No OMIM phenotype Antibody deficiency (Wang (2013) Proc Natl Acad Sci USA 110, 5127)
TPP2	115,5	99.5%	97.3%	No OMIM phenotype Evans syndrome, immunodeficiency and premature immunosenescence (Stepensky (2015) Blood 125, 753)
TRAC	127,5	100.0%	100.0%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	109	100.0%	98.9%	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849
TRAF3IP2	111,1	100.0%	98.3%	?Candidiasis, familial, 8, 615527 {Psoriasis susceptibility 13}, 614070
TREX1	233,4	100.0%	100.0%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRNT1	101,5	99.2%	96.5%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TTC37	135,1	99.9%	99.2%	Trichohepatoenteric syndrome 1, 222470
TTC7A	115	99.8%	98.0%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	129,9	100.0%	99.5%	Immunodeficiency 35, 611521

UNC13D	108,2	99.8%	98.7%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	62,9	61.4%	60.1%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551
UNG	116,8	99.5%	95.6%	Immunodeficiency with hyper IgM, type 5, 608106
USB1	118,2	99.8%	97.2%	Poikiloderma with neutropenia, 604173
USP18	144,4	95.9%	95.9%	Pseudo-TORCH syndrome 2, 617397
VAV1	103,8	98.5%	96.2%	No OMIM phenotype
VPS13B	134,5	99.3%	98.0%	Cohen syndrome, 216550
VPS45	126,5	97.3%	94.4%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	70,4	94.2%	83.6%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WDR1	98,9	99.9%	98.7%	No OMIM phenotype
WIPF1	89,1	100.0%	99.1%	?Wiskott-Aldrich syndrome 2, 614493
WRAP53	162,8	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	89,4	93.3%	87.9%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	186,1	100.0%	99.9%	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840
ZBTB24	155,5	100.0%	100.0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors