PRIMARY IMMUNODEFICIENCIES GENE PANEL DG 2.15 (386 genes)

Releasedate: 31-01-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACD	135.2	100	98.2	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACP5	196.2	100	99.9	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	129	99.1	94.2	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADA	113	98.9	97.3	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	101.4	99.9	99.1	?Sneddon syndrome, 182410 Polyarteritis nodosa, childhood-onset, 615688
ADAM17	139.4	97.6	93.8	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAR	125	100	99.8	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	130.2	100	100	Aspartylglucosaminuria, 208400
AICDA	139	89.8	82.6	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	68.2	98.9	92	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	111.8	99.8	96.6	Reticular dysgenesis, 267500
ALG13	86.7	98.7	94.1	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884
AP1S3	114.2	90.3	90.1	{Psoriasis 15, pustular, susceptibility to}, 616106
AP3B1	95	97.8	90.2	Hermansky-Pudlak syndrome 2, 608233
AP3D1	121	98.1	97.8	?Hermansky-Pudlak syndrome 10, 617050
APOL1	192.2	100	100	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
ARPC1B	126.4	100	99.9	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
ATM	109.7	99	94	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	112.8	99.1	94.7	Immunodeficiency 47, 300972
B2M	252.1	100	99.9	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
BACH2	154.8	100	99.9	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
BCL10	97.9	100	99.8	?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	79.7	96.6	88.6	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BLK	115.7	100	100	Maturity-onset diabetes of the young, type 11, 613375
BLM	116.3	99.4	96.5	Bloom syndrome, 210900
BLNK	95.7	93.7	91.3	?Agammaglobulinemia 4, 613502
BLOC1S6	97.2	98.7	91.3	?Hermansky-pudlak syndrome 9, 614171
BTK	116.2	100	99.6	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
C1QA	120.4	100	99	C1q deficiency, 613652
C1QB	183.4	100	99.9	C1q deficiency, 613652
C1QC	198.1	100	98.9	C1q deficiency, 613652
C1R	156.9	100	100	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	117.6	100	99.7	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	129.9	100	100	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C3	145.5	100	99.7	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	91.5	98.1	95.9	C4a deficiency, 614380

				[Blood group, Rodgers], 614374
C4B	90.5	98.5	96.5	C4B deficiency, 614379
C5	134.4	98.4	95.3	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C6	157.5	100	99.9	C6 deficiency, 612446 Combined C6/C7 deficiency, 0
C7	132.3	99.1	94.8	C7 deficiency, 610102
C8A	120.2	100	99.8	C8 deficiency, type I, 613790
C8B	135.8	99.9	99.5	C8 deficiency, type II, 613789
C8G	130.1	100	100	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
C9	133.7	100	98.5	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
CA2	140.7	100	99.3	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CARD11	154.6	99.9	98.6	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206 Immunodeficiency 11B with atopic dermatitis, 617638
CARD14	116.2	99.7	97.7	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	119.7	98.3	96.4	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	121.1	94.9	92.7	Immunodeficiency 58, 618131
CASP10	117.4	99.5	98	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP8	144.8	95.6	95.5	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CAVIN1	137	99.9	99.3	Lipodystrophy, congenital generalized, type 4, 613327
CCBE1	75.9	98.9	95.5	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD19	88.8	99.9	98.4	Immunodeficiency, common variable, 3, 613493
CD247	101.6	100	98.9	?Immunodeficiency 25, 610163
CD27	118.1	100	99.6	Lymphoproliferative syndrome 2, 615122
CD3D	193.8	100	100	Immunodeficiency 19, 615617

CD3E	152.1	100	99.9	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	156.8	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	165.4	100	99.9	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	126.6	95.9	86.8	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	115.1	97.8	93.2	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD55	138.9	94.4	86.2	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 [Blood group Cromer], 613793
CD59	200.9	93.6	86.5	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	116.4	100	99.3	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
CD79A	128.3	99.8	97	Agammaglobulinemia 3, 613501
CD79B	210.7	100	100	Agammaglobulinemia 6, 612692
CD81	142.6	99.9	98.1	Immunodeficiency, common variable, 6, 613496
CD8A	110	99.9	99	CD8 deficiency, familial, 608957
CDCA7	109.2	100	99.3	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDKN2B	85.9	100	99.8	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	71.1	99.3	95.8	Specific granule deficiency, 245480
CFB	147.1	100	100	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFD	80.6	89.7	81.6	Complement factor D deficiency, 613912
CFH	183.2	98.7	95.3	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	236.5	95.8	94.2	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR2	171.7	96.8	89	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)

CFHR3	101	90.6	85.6	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR4	139.7	99.6	97.3	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
CFHR5	97.3	98.7	93.4	Nephropathy due to CFHR5 deficiency, 614809
CFI	145.5	96.6	92.8	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFP	95.9	98.4	93.4	Properdin deficiency, X-linked, 312060
CFTR	124	99.1	96.3	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	150.7	99.9	98.9	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CIITA	125	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CLCN7	129.7	99.5	98.2	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
CLEC4D	140.4	100	100	No OMIM phenotype plays a role in immunity (Zhu et al.,Immunity 2013).
CLEC7A	150.7	100	99.9	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
CLPB	140.2	100	99.5	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COLEC11	203	100	100	3MC syndrome 2, 265050
COPA	133.2	100	100	{Autoimmune interstitial lung, joint, and kidney disease}, 616414
CORO1A	154.4	99.8	96.9	Immunodeficiency 8, 615401
CR2	160.7	100	99.8	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CREBBP	123.5	99.4	96.7	Rubinstein-Taybi syndrome 1, 180849
CSF2RA	66	89.9	88.2	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	94.8	99.6	97.8	Surfactant metabolism dysfunction, pulmonary, 5, 614370

CSF3R	94.4	99.3	96.5	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	119	100	99.8	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	193.9	100	100	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	143.1	100	99.6	Immunodeficiency 24, 615897
CTSC	127.5	100	100	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CXCR4	202.7	100	99.9	Myelokathexis, isolated, 0 WHIM syndrome, 193670
CYBA	97.3	77.9	71	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	110.8	99.9	99.2	Chronic granulomatous disease, X-linked, 306400 Immunodeficiency 34, mycobacteriosis, X-linked, 300645
DCLRE1C	128.8	98.2	94.5	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDX58	123.3	98.6	95	Singleton-Merten syndrome 2, 616298
DHFR	48.4	91.1	72	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	111.9	99.6	98.1	Dyskeratosis congenita, X-linked, 305000
DNASE1	198.8	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700
DNMT3B	124.8	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	143.8	100	99.9	Immunodeficiency 40, 616433
DOCK8	129.1	100	99.8	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	80.9	99.7	95.9	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	76.8	99.8	97.5	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	126	99.3	97.7	Vici syndrome, 242840
ERCC2	123.7	100	99.7	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730

ERCC3	113.2	99.9	98.9	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
EXTL3	206.4	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
F12	111.4	100	99.5	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
FAAP24	118.5	98.5	94.8	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
FADD	142.8	100	99.6	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAS	272.1	100	99.3	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0 {Autoimmune lymphoproliferative syndrome}, 601859
FASLG	86.2	100	98.5	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FAT4	224.5	100	99.9	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FCGR1A	82.3	47.5	46.5	[IgG receptor I, phagocytic, familial deficiency of], 0
FCGR2A	244.9	100	100	{Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162 {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700
FCGR2B	176.9	99.9	97.2	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700
FCGR3A	225	99.6	97.7	Immunodeficiency 20, 615707
FCGR3B	176.6	99.1	98.2	Neutropenia, alloimmune neonatal, 0
FCN3	127.8	100	99.4	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	122.4	100	98.9	Leukocyte adhesion deficiency, type III, 612840
FOXN1	112.5	100	99.5	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	124.6	98.7	91.6	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FPR1	216.9	100	100	No OMIM phenotype Congenital defects of Phagocyte number,function or both Nanamori M et al.,2004
G6PC	180.7	100	100	Glycogen storage disease Ia, 232200
G6PC3	123.7	100	100	Dursun syndrome, 612541

				Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	118.3	99.5	97.5	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GATA2	119.6	99.9	98.5	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GFI1	83.1	99	92.9	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 ?Neutropenia, severe congenital 2, autosomal dominant, 613107
GINS1	124.3	96.6	83.2	Immunodeficiency 55, 617827
GJC2	41.9	68.9	58.6	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GRHL2	134.6	100	100	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GTF2H5	113.6	100	99.1	Trichothiodystrophy 3, photosensitive, 616395
HAX1	136.5	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HELLS	93.7	94.1	86.8	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HMOX1	128.7	95.8	89.5	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HYOU1	134	99.9	99.7	?Immunodeficiency 59 and hypoglycemia, 233600
ICOS	160.2	100	100	Immunodeficiency, common variable, 1, 607594
IFIH1	113.5	99.6	97.1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNAR2	138.8	98.5	95.4	?Immunodeficiency 45, 616669 {Hepatitis B virus, susceptibility to}, 610424
IFNGR1	138.5	99.2	97.3	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	142.3	93.2	93.1	Immunodeficiency 28, mycobacteriosis, 614889

IGHM	185.2	100	100	Agammaglobulinemia 1, 601495
IGLL1	86.2	99.3	94.9	Agammaglobulinemia 2, 613500
IKBKB	123.5	98.5	94.2	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	52.5	84.6	73.2	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	183.4	100	100	Immunodeficiency, common variable, 13, 616873
IL10	125.3	100	99.9	{Graft-versus-host disease, protection against}, 614395 {HIV-1, susceptibility to}, 609423 {Rheumatoid arthritis, progression of}, 180300
IL10RA	141.9	100	99.9	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	168.8	98.7	96.1	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL12B	121.1	100	99.9	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	124.3	97	94.7	Immunodeficiency 30, 614891
IL17F	85.8	99.1	94.4	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	140.5	99.8	96.9	Immunodeficiency 51, 613953
IL17RC	96.1	99.8	99	Candidiasis, familial, 9, 616445
IL1RN	162.8	100	100	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL2	65.3	93.4	76.9	No OMIM phenotype Severe combined immunodeficiency due to IL2 deficiency,Combined T-cell and B-cell immunodeficiencies
IL21	94.7	97.8	87.5	?Immunodeficiency, common variable, 11, 615767
IL21R	128.5	100	100	Immunodeficiency 56, 615207 [IgE, elevated level of], 147050
IL2RA	116.4	100	99.5	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	65.2	99.8	97.3	Combined immunodeficiency, X-linked, moderate, 312863

				Severe combined immunodeficiency, X-linked, 300400
IL36RN	99	100	100	Psoriasis 14, pustular, 614204
IL7R	129.5	99.9	99.4	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INO80	105.1	99.2	97.1	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
INSR	141.1	97.1	94.5	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
IRAK1	59.7	92.8	80.4	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
IRAK4	95.4	98.3	90.1	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676
IRF2BP2	63.2	88.2	70.8	?Immunodeficiency, common variable, 14, 617765
IRF3	116.5	99.9	99.1	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532
IRF7	89.8	99.9	99.1	?Immunodeficiency 39, 616345
IRF8	114.7	99.6	97.4	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
ISG15	160.1	100	100	Immunodeficiency 38, 616126
ITCH	124.7	95.4	94.8	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGB2	152.1	100	99.8	Leukocyte adhesion deficiency, 116920
ITK	125.2	100	99.6	Lymphoproliferative syndrome 1, 613011
JAGN1	147.3	100	100	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	122.8	99.8	98.7	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
JAK2	90.6	95.9	94.1	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600880
JAK3	104.2	98.2	95.2	SCID, autosomal recessive, T-negative/B-positive type, 600802
KDM6A	109	93.2	84.3	Kabuki syndrome 2, 300867
KMT2D	142.1	99.9	99	Kabuki syndrome 1, 147920

LAMTOR2	167	100	99.9	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAT	94.6	99.8	98.4	Immunodeficiency 52, 617514
LCK	161.4	98.2	95.5	?Immunodeficiency 22, 615758
LIG1	95.4	100	99	No OMIM phenotype DNA ligase I deficiency Combined T-cell and B-cell immunodeficiencies 1 studie Barnes DE et al.,1992
LIG4	165.6	100	99.6	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LPIN2	111.5	100	99.6	Majeed syndrome, 609628
LRBA	134.4	99.3	97.8	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	273	100	99.9	?Agammaglobulinemia 5, 613506
LTBP3	113.5	98.7	94.7	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LYST	134.6	97.8	93.9	Chediak-Higashi syndrome, 214500
MAGT1	101.8	98.4	95.8	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAL	138.6	100	99.9	No OMIM phenotype
MALT1	136.6	89.1	85.4	Immunodeficiency 12, 615468
MAN2B1	122.3	99.1	96.2	Mannosidosis, alpha-, types I and II, 248500
MANBA	119.9	99.7	97.2	Mannosidosis, beta, 248510
MAP3K14	111.3	99.3	98.4	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
MASP2	139.8	100	99.3	MASP2 deficiency, 613791
MBL2	109.8	100	99.6	{Chronic infections, due to MBL deficiency}, 614372
MC2R	213.1	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM4	164.3	99.9	98.8	Immunodeficiency 54, 609981
MEFV	108.8	94.9	91	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MKL1	101	96.2	91.1	Megakaryoblastic leukemia, acute, 0
MOGS	121.6	99.8	99.1	Congenital disorder of glycosylation, type IIb, 606056
MRE11	51.2	95.3	82.3	Ataxia-telangiectasia-like disorder 1, 604391
MS4A1	123.7	99.4	96.2	Immunodeficiency, common variable, 5, 613495

MSN	88.6	99	95	Immunodeficiency 50, 300988
MTHFD1	139.6	99.8	98.4	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MVK	124.3	92.1	90.4	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYD88	186.5	100	99.9	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
MYSM1	96	97.6	92.5	Bone marrow failure syndrome 4, 618116
NBAS	145.3	99.5	97.6	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBN	80.6	99.1	94.6	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCF1	23.9	25.8	22.1	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	124.3	100	99.4	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	158.7	100	100	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	111.9	100	99.8	Acne inversa, familial, 1, 142690
NFAT5	216.5	99.2	97.8	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
NFKB1	105.3	99.3	96.8	Immunodeficiency, common variable, 12, 616576
NFKB2	123.3	97.5	92.6	Immunodeficiency, common variable, 10, 615577
NFKBIA	116.3	98.5	93.8	Ectodermal dysplasia and immunodeficiency, 612132
NHEJ1	80.3	100	99.1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	111	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NLRC4	179.9	100	99.7	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050
NLRP1	126.1	99	96.5	Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP12	165.9	100	99.9	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	150.4	100	100	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	135.9	99.8	98.7	Hydatidiform mole, recurrent, 1, 231090
NOD2	135.8	100	99.7	Blau syndrome, 186580 {Inflammatory bowel disease 1, Crohn disease}, 266600 {Psoriatic arthritis, susceptibility to}, 607507 {Yao syndrome}, 617321
NOP10	160.5	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	188.4	100	100	?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	130	99.9	98.5	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
ORAI1	237.3	93.8	89.8	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
OSTM1	80.7	90.8	88.4	Osteopetrosis, autosomal recessive 5, 259720
OTULIN	149.5	90.5	86.3	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
PARN	128.4	99.9	98	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX5	118.8	98.3	95.4	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PBX1	111.8	99.3	95.2	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	103.1	96.4	89.2	Propionicacidemia, 606054
PCCB	129.8	98.7	96.4	Propionicacidemia, 606054
PEPD	116	99.6	98.5	Prolidase deficiency, 170100
PGM3	191.4	99.9	99.7	Immunodeficiency 23, 615816
PIGA	90.5	90.4	81.3	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818

PIK3CD	132.7	99.2	96.8	Immunodeficiency 14, 615513
PIK3R1	129.3	99.7	97.3	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PLCG2	118.9	100	99.8	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEKHM1	141.1	100	99.9	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLG	115.4	87.8	87	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PMM2	141.1	99.9	99.4	Congenital disorder of glycosylation, type Ia, 212065
PNP	151.4	100	99.5	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POLA1	110.7	98.2	92.8	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220
POLE2	59	93.4	74.6	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
POMP	114.4	95.2	87.5	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 Proteasome-associated autoinflammatory syndrome 2, 618048
POT1	90.7	99.6	96	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
PRF1	122.5	91.2	90.8	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKCD	181.2	100	99.9	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	106.7	98.4	94.8	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	149.5	100	100	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	67.6	100	98.4	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMA3	66.5	100	97.8	No OMIM phenotype autoinflammatory disorder (INFEVERS website http://fmf.igh.cnrs.fr/ISSAID/infevers/) and based on literature it underlies a human primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)

PSMB4	122.5	100	99.2	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	118.7	100	99.8	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	85.6	99.5	95.7	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSTPIP1	88.2	99.7	97.7	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN22	134.5	98	91.9	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700
PTPRC	101.6	93.9	86.3	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitis C virus, susceptibility to}, 609532
RAB27A	143.9	100	99.9	Griscelli syndrome, type 2, 607624
RAC2	104.1	100	99.4	Neutrophil immunodeficiency syndrome, 608203
RAG1	206.9	100	100	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	221	100	100	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RANBP2	110.9	49.7	48.9	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033
RASGRP1	127.5	99.9	99.5	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
RASGRP2	97.5	99.9	98.6	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	104.1	99.2	94.9	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RECQL4	149.6	99.2	96.5	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RELB	90.6	87.9	75.1	?Immunodeficiency 53, 617585
RFX5	116.9	98.7	96.3	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	105.9	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	84.8	94.4	91.6	Bare lymphocyte syndrome, type II, complementation group D, 209920
RHOH	134.9	100	100	No OMIM phenotype Combined T-cell and B-cell immunodeficiencies

				1 studie Crequer A et al.,2012
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	142.1	100	99.9	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	103.8	93.2	87.5	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	209.2	100	99.9	Aicardi-Goutieres syndrome 3, 610329
RNF168	215.3	100	99.1	RIDDLE syndrome, 611943
RNF31	154.7	99.5	98.2	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	132.3	100	100	Immunodeficiency 42, 616622
RPSA	88.8	100	99.7	Asplenia, isolated congenital, 271400
RSPH9	127.4	100	99.6	Ciliary dyskinesia, primary, 12, 612650
RTEL1	110.9	99.2	95.1	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
SAMD9	159.1	99.9	99.3	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	165.7	100	99.9	Ataxia-pancytopenia syndrome, 159550
SAMHD1	127.9	99.6	96.6	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SBDS	212.3	100	99.9	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SEMA3E	142.6	99.9	99	?CHARGE syndrome, 214800
SERAC1	112.5	98.8	94.6	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	97.9	97.3	92.6	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SH2B3	97.5	90.7	79	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950

SH2D1A	104.7	89.9	89.4	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	110.9	91.4	91.4	Cherubism, 118400
SKIV2L	149.1	100	99.8	Trichohepatoenteric syndrome 2, 614602
SLC29A3	203.6	99.9	99.5	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	124	99.9	97.7	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	230.2	99.9	98.4	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	140.2	100	99.9	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	81.8	99.2	96	Acrodermatitis enteropathica, 201100
SLC46A1	106	99.4	96.4	Folate malabsorption, hereditary, 229050
SMARCAL1	134.6	100	99.9	Schimke immunoosseous dysplasia, 242900
SMARCD2	92.9	87	85.7	Specific granule deficiency 2, 617475
SNX10	118.9	96.2	96.1	Osteopetrosis, autosomal recessive 8, 615085
SOCS4	262.8	99.9	99.3	No OMIM phenotype Autoimmunity (Arts (2015) J Intern Med epub,epub)
SP110	121.6	100	99.5	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SPINK5	145	99.4	96.5	Netherton syndrome, 256500
STAT1	126.2	98	95.8	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	116	100	99.9	Immunodeficiency 44, 616636
STAT3	119.5	99.9	99	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT4	144.2	98.7	97.1	{Systemic lupus erythematosus, susceptibility to, 11}, 612253
STAT5B	130.6	99.7	97.2	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STAT6	119.3	100	99.7	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	145.3	100	99.2	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STK4	138.9	100	99.3	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868

STX11	311.4	100	100	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	102.3	88.9	83.8	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TAP1	103.3	100	99.1	Bare lymphocyte syndrome, type I, 604571
TAP2	95.2	99.6	98.6	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	100.7	96.1	94.3	Bare lymphocyte syndrome, type I, 604571
TAZ	94	99.9	98.8	Barth syndrome, 302060
TBX1	75.3	77.1	67.4	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TCF3	67.8	98.9	92.3	Agammaglobulinemia 8, autosomal dominant, 616941
TCIRG1	113.5	95.4	89.4	Osteopetrosis, autosomal recessive 1, 259700
TCN2	175.6	100	100	Transcobalamin II deficiency, 275350
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERT	138.3	95.3	92	{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	157.2	99.9	99.1	Immunodeficiency 46, 616740
THBD	108.2	99.8	97.8	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TICAM1	111.1	100	99.1	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850
TINF2	184	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TIRAP	136.4	100	100	{Bacteremia, protection against}, 614382 {Malaria, protection against}, 611162 {Pneumococcal disease, invasive, protection against}, 610799 {Tuberculosis, protection against}, 607948
TLR3	185.4	99.8	98.6	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002 {HIV1 infection, resistance to}, 609423
TLR4	132.7	100	99.9	Endotoxin hyporesponsiveness

				{Colorectal cancer, susceptibility to}, 114500 {Macular degeneration, age-related, 10}, 611488
TMC6	83.7	99.9	99	Epidermodysplasia verruciformis, 226400
TMC8	108.1	97.6	91.6	Epidermodysplasia verruciformis 2, 618231
TMEM173	100.8	98.7	93.4	STING-associated vasculopathy, infantile-onset, 615934
TNFAIP3	135.9	100	99.9	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF11A	146.3	93.3	91.4	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF13B	102.1	100	99.7	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	55.8	76.5	66.8	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	93.2	90.8	87.9	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFRSF4	51.6	97.3	85.4	?Immunodeficiency 16, 615593
TNFSF11	150.4	99.3	93.2	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	77.5	94.7	90.6	No OMIM phenotype Antibody deficiency (Wang (2013) Proc Natl Acad Sci USA 110, 5127)
TPP2	119.2	98.9	94.3	No OMIM phenotype Evans syndrome, immunodeficiency and premature immunosenescence (Stepensky (2015) Blood 125, 753)
TRAC	170.9	100	100	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	130.6	99.9	98.4	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849
TRAF3IP2	116.6	99.9	97.7	?Candidiasis, familial, 8, 615527 {Psoriasis susceptibility 13}, 614070
TREX1	242.4	100	100	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRNT1	104.6	97.8	92.3	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TTC37	124	99.6	98.1	Trichohepatoenteric syndrome 1, 222470
TTC7A	123	99.9	98.3	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	119.2	99.9	98.8	Immunodeficiency 35, 611521

UNC13D	97	99.6	97.7	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	60	56.8	54.5	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551
UNG	78.4	99.5	94.2	Immunodeficiency with hyper IgM, type 5, 608106
USB1	125	99.9	98.2	Poikiloderma with neutropenia, 604173
USP18	201.4	95.9	95.9	Pseudo-TORCH syndrome 2, 617397
VAV1	105.7	98.3	94.7	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
VPS13B	143.8	98.6	96.8	Cohen syndrome, 216550
VPS45	131.5	96.2	94.9	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	66.1	88.2	78.7	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WDR1	114.1	100	99.2	No OMIM phenotype primary (auto)immune disorder (Picard et al.,J Clin Immunol (2018) 38:96)
WIPF1	77.5	100	99.2	?Wiskott-Aldrich syndrome 2, 614493
WRAP53	154.4	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	107.1	91.9	86.6	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	185.6	99.9	99.5	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840
ZBTB24	178.1	100	100	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 : December 31st, 2018.
This list is accurate for panel version DG 2.15
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors