PRIMARY IMMUNODEFICIENCIES GENE PANEL DG 2.13 (381 genes)

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACD	135.2	100	98	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ACP5	196.2	100	99	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	129	99	94	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ADA	113	98	97	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	101.4	99	99	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410
ADAM17	139.4	97	93	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAR	125	100	99	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	130.2	100	100	Aspartylglucosaminuria, 208400
AICDA	139	89	82	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	68.2	98	92	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	111.8	99	96	Reticular dysgenesis, 267500
ALG13	86.7	98	94	Epileptic encephalopathy, early infantile, 36, 300884
AP1S3	114.2	90	90	{Psoriasis 15, pustular, susceptibility to}, 616106
AP3B1	95	97	90	Hermansky-Pudlak syndrome 2, 608233
AP3D1	121	98	97	?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	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
ATM	109.7	99	94	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480

ATP6AP1	112.8	99	94	Immunodeficiency 47, 300972
B2M	252.1	100	99	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
BACH2	154.8	100	99	No OMIM phenotype
BCL10	97.9	100	99	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic}, 273300, {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic},
BCL11B	79.7	96	88	?Immunodeficiency 49, 617237
BLK	115.7	100	100	Maturity-onset diabetes of the young, type 11, 613375
BLM	116.3	99	96	Bloom syndrome, 210900
BLNK	95.7	93	91	Agammaglobulinemia 4, 613502
BLOC1S6	97.2	98	91	Hermansky-pudlak syndrome 9, 614171
BTK	116.2	100	99	Agammaglobulinemia and isolated hormone deficiency, 307200 Agammaglobulinemia, X-linked 1, 300755
C1QA	120.4	100	99	C1q deficiency, 613652
C1QB	183.4	100	99	C1q deficiency, 613652
C1QC	198.1	100	98	C1q deficiency, 613652
C1R	156.9	100	100	C1r/C1s deficiency, combined, 216950
C1S	117.6	100	99	C1s deficiency, 613783
C2	129.9	100	100	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C3	145.5	100	99	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	91.5	98	95	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	90.5	98	96	C4B deficiency, 614379
C5	134.4	98	95	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C6	157.5	100	99	C6 deficiency, 612446 Combined C6/C7 deficiency

C7	132.3	99	94	C7 deficiency, 610102
C8A	120.2	100	99	C8 deficiency, type I, 613790
C8B	135.8	99	99	C8 deficiency, type II, 613789
C8G	130.1	100	100	No OMIM phenotype
C9	133.7	100	98	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
CA2	140.7	100	99	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CARD11	154.6	99	98	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11, 615206
CARD14	116.2	99	97	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	119.7	98	96	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	121.1	94	92	No OMIM phenotype Immunodeficiency, combined (Wang (2016) J Exp Med 213,2413) Immunodeficiency, primary (Schober (2017) Nat Commun 8,14209)
CASP10	117.4	99	98	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP8	144.8	95	95	Hepatocellular carcinoma, somatic, 114550 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CAVIN1	137	99	99	Lipodystrophy, congenital generalized, type 4, 613327
CCBE1	75.9	98	95	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD19	88.8	99	98	Immunodeficiency, common variable, 3, 613493
CD247	101.6	100	98	?Immunodeficiency 25, 610163
CD27	118.1	100	99	Lymphoproliferative syndrome 2, 615122
CD3D	193.8	100	100	Immunodeficiency 19, 615617
CD3E	152.1	100	99	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	156.8	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	165.4	100	99	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	126.6	95	86	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	115.1	97	93	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922

CD55	138.9	94	86	[Blood group Cromer], 613793
CD59	200.9	93	86	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	116.4	100	99	No OMIM phenotype
CD79A	128.3	99	97	Agammaglobulinemia 3, 613501
CD79B	210.7	100	100	Agammaglobulinemia 6, 612692
CD81	142.6	99	98	Immunodeficiency, common variable, 6, 613496
CD8A	110	99	99	CD8 deficiency, familial, 608957
CDCA7	109.2	100	99	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDKN2B	85.9	100	99	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 Cancer 4, 301)
CEBPE	71.1	99	95	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	81	Complement factor D deficiency, 613912
CFH	183.2	98	95	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	94	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	101	90	85	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR5	97.3	98	93	Nephropathy due to CFHR5 deficiency, 614809
CFI	145.5	96	92	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	93	Properdin deficiency,X-linked, 312060
CFTR	124	99	96	Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 Sweat chloride elevation without CF

				{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 {Hypertrypsinemia, neonatal} {Pancreatitis, idiopathic}, 167800
CHD7	150.7	99	98	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	98	Osteopetrosis, autosomal dominant 2, 166600 Osteopetrosis, autosomal recessive 4, 611490
CLEC4D	140.4	100	100	No OMIM phenotype
CLEC7A	150.7	100	99	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
CLPB	140.2	100	99	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	96	Immunodeficiency 8, 615401
CR2	160.7	100	99	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CREBBP	123.5	99	96	Rubinstein-Taybi syndrome, 180849
CSF2RA	66	89	88	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	94.8	99	97	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	94.4	99	96	?Neutrophilia, hereditary, 162830
CTC1	119	100	99	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	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	Myelokathexis, isolated WHIM syndrome, 193670

CYBA	97.3	77	71	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	110.8	99	99	Chronic granulomatous disease, X-linked, 306400 Immunodeficiency 34, mycobacteriosis, X-linked, 300645
DCLRE1C	128.8	98	94	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDX58	123.3	98	95	Singleton-Merten syndrome 2, 616298
DHFR	48.4	91	72	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	111.9	99	98	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	Immunodeficiency 40, 616433
DOCK8	129.1	100	99	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	80.9	99	95	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	76.8	99	97	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	97	Vici syndrome, 242840
ERCC2	123.7	100	99	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	113.2	99	98	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	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
FAAP24	118.5	98	94	No OMIM phenotype
FADD	142.8	100	99	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759
FAS	272.1	100	99	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic {Autoimmune lymphoproliferative syndrome}, 601859
FASLG	86.2	100	98	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980

FAT4	224.5	100	99	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FCGR1A	82.3	47	46	[IgG receptor I, phagocytic, familial deficiency of]
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	97	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700
FCGR3A	225	99	97	Immunodeficiency 20, 615707
FCGR3B	176.6	99	98	Neutropenia, alloimmune neonatal
FCN3	127.8	100	99	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	122.4	100	98	Leukocyte adhesion deficiency,type III,612840
FOXN1	112.5	100	99	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	124.6	98	91	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FPR1	216.9	100	100	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	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	97	Favism, 134700 Hemolytic anemia due to G6PD deficiency, 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GATA2	119.6	99	98	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GFI1	83.1	99	92	Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GINS1	124.3	96	83	Immunodeficiency 55, 617827
GJC2	41.9	68	58	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480

				Spastic paraplegia 44, autosomal recessive, 613206
GRHL2	134.6	100	100	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GTF2H5	113.6	100	99	Trichothiodystrophy 3, photosensitive, 616395
HAX1	136.5	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HELLS	93.7	94	86	Immunodeficiency-centromeric instability-facial anomalies syndrome 4,616911
HMOX1	128.7	95	89	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HYOU1	134	99	99	No OMIM phenotype
ICOS	160.2	100	100	Immunodeficiency, common variable, 1, 607594
IFIH1	113.5	99	97	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNAR2	138.8	98	95	?Immunodeficiency 45, 616669 {Hepatitis B virus, susceptibility to}, 610424
IFNGR1	138.5	99	97	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	93	Immunodeficiency 28, mycobacteriosis, 614889
IGHM	185.2	100	100	Agammaglobulinemia 1, 601495
IGKC	157.9	100	100	Kappa light chain deficiency, 614102
IGLL1	86.2	99	94	Agammaglobulinemia 2, 613500
IKBKB	123.5	98	94	Immunodeficiency 15, 615592
IKBKG	52.5	84	73	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 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, 1,616873
IL10	125.3	100	99	{Graft-versus-host disease, protection against}, 614395 {HIV-1, susceptibility to}, 609423

				{Rheumatoid arthritis, progression of}, 180300
IL10RA	141.9	100	99	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	168.8	98	96	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL12B	121.1	100	99	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	124.3	97	94	Immunodeficiency 30, 614891
IL17F	85.8	99	94	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	140.5	99	96	Immunodeficiency 51, 613953
IL17RC	96.1	99	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	76	Severe combined immunodeficiency due to IL2 deficiency
IL21	94.7	97	87	?Immunodeficiency, common variable, 11, 615767
IL21R	128.5	100	100	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 [IgE, elevated level of], 147050
IL2RA	116.4	100	99	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	65.2	99	97	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL36RN	99	100	100	Psoriasis 14, pustular, 614204
IL7R	129.5	99	99	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INSR	141.1	97	94	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
IRAK1	59.7	92	80	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	95.4	98	90	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676
IRF2BP2	63.2	88	70	?Immunodeficiency, common variable, 14, 617765
IRF3	116.5	99	99	{?Herpes simplex encephalitis, susceptibility to, 7}, 616532
IRF7	89.8	99	99	?Immunodeficiency 39, 616345

IRF8	114.7	99	97	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 614894
ISG15	160.1	100	100	Immunodeficiency 38, 616126
ITCH	124.7	95	94	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGB2	152.1	100	99	Leukocyte adhesion deficiency, 116920
ITK	125.2	100	99	Lymphoproliferative syndrome 1, 613011
JAGN1	147.3	100	100	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	122.8	99	98	No OMIM phenotype
JAK2	90.6	95	94	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600800
JAK3	104.2	98	95	SCID, autosomal recessive, T-negative/B-positive type, 600802
KDM6A	109	93	84	Kabuki syndrome 2, 300867
KMT2D	142.1	99	99	Kabuki syndrome 1, 147920
LAMTOR2	167	100	99	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAT	94.6	99	98	Immunodeficiency 52
LCK	161.4	98	95	?Immunodeficiency 22, 615758
LIG1	95.4	100	99	DNA ligase I deficiency
LIG4	165.6	100	99	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LPIN2	111.5	100	99	Majeed syndrome, 609628
LRBA	134.4	99	97	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	273	100	99	Agammaglobulinemia 5, 613506
LTBP3	113.5	98	94	Dental anomalies and short stature, 601216
LYST	134.6	97	93	Chediak-Higashi syndrome, 214500
MAGT1	101.8	98	95	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAL	138.6	100	99	No OMIM phenotype
MALT1	136.6	89	85	Immunodeficiency 12, 615468
MAN2B1	122.3	99	96	Mannosidosis, alpha-, types I and II, 248500
MANBA	119.9	99	97	Mannosidosis, beta, 248510

MAP3K14	111.3	99	98	No OMIM phenotype
MASP2	139.8	100	99	MASP2 deficiency, 613791
MBL2	109.8	100	99	{Chronic infections, due to MBL deficiency}, 614372
MC2R	213.1	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM4	164.3	99	98	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MEFV	108.8	94	91	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MKL1	101	96	91	Megakaryoblastic leukemia, acute
MOGS	121.6	99	99	Congenital disorder of glycosylation, type IIb, 606056
MPO	155	100	99	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}
MRE11	51.2	95	82	Ataxia-telangiectasia-like disorder, 604391
MS4A1	123.7	99	96	Immunodeficiency, common variable, 5, 613495
MSN	88.6	99	95	Immunodeficiency 50, 300988
MTHFD1	139.6	99	98	{Abruptio placentae, susceptibility to} {Spina bifida, folate-sensitive, susceptibility to}, 601634
MVK	124.3	92	90	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYD88	186.5	100	99	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
MYSM1	96	97	92	No OMIM phenotype ?Anaemia, transfusion-dependent, thrombocytopaenia, and low NK- and B- cell counts (Alsultan (2013) Blood 122, 3844)
NBAS	145.3	99	97	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBN	80.6	99	94	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCF1	23.9	25	22	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	124.3	100	99	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	Acne inversa, familial, 1, 142690

NFAT5	216.5	99	97	No OMIM phenotype
NFKB1	105.3	99	96	Immunodeficiency, common variable, 12, 616576
NFKB2	123.3	97	92	Immunodeficiency, common variable, 10, 615577
NFKBIA	116.3	98	93	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NHEJ1	80.3	100	99	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	111	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NKX2-5	83.2	100	99	Atrial septal defect 7, with or without AV conduction defects, 108900 Conotruncal heart malformations, variable, 217095 Hypoplastic left heart syndrome 2, 614435 Hypothyroidism, congenital nongoitrous, 5, 225250 Tetrology of Fallot, 187500 Ventricular septal defect 3, 614432
NLRC4	179.9	100	99	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
NLRP1	126.1	99	96	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP12	165.9	100	99	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	150.4	100	100	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900
NOD2	135.8	100	99	Blau syndrome, 186580 Yao syndrome, 617321 {Inflammatory bowel disease 1}, 266600 {Psoriatic arthritis, susceptibility to}, 607507
NOP10	160.5	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	188.4	100	100	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 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470
NSMCE3	130	99	98	Lung disease,immunodeficiency and chromosome breakage syndrome, 617241

ORAI1	237.3	93	89	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
OSTM1	80.7	90	88	Osteopetrosis, autosomal recessive 5, 259720
OTULIN	149.5	90	86	Autoinflammation,panniculitis, and dermatosis syndrome, 617099
PARN	128.4	99	98	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX5	118.8	98	95	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PBX1	111.8	99	95	Leukemia, acute pre-B-cell, 176310
PCCA	103.1	96	89	Propionicacidemia, 606054
PCCB	129.8	98	96	Propionicacidemia, 606054
PEPD	116	99	98	Prolidase deficiency, 170100
PGM3	191.4	99	99	Immunodeficiency 23, 615816
PIGA	90.5	90	81	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIK3CD	132.7	99	96	Immunodeficiency 14, 615513
PIK3R1	129.3	99	97	Immunodeficiency 36, 616005 SHORT syndrome, 269880 ?Agammaglobulinemia 7, autosomal recessive, 615214
PLCG2	118.9	100	99	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEKHM1	141.1	100	99	Osteopetrosis, autosomal recessive 6, 611497
PLG	115.4	87	87	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PMM2	141.1	99	99	Congenital disorder of glycosylation, type Ia, 212065
PNP	151.4	100	99	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POLA1	110.7	98	92	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220
POLE2	59	93	74	No OMIM phenotype
POT1	90.7	99	96	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
PRF1	122.5	91	90	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKCD	181.2	100	99	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	106.7	98	94	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	Acne inversa, familial, 2, 613736
PSMB8	118.7	100	99	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSTPIP1	88.2	99	97	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN22	134.5	98	91	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700
PTPRC	101.6	93	86	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitic C virus, susceptibility to}, 609532
RAB27A	143.9	100	99	Griscelli syndrome, type 2, 607624
RAC2	104.1	100	99	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	48	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033
RASGRP1	127.5	99	99	No OMIM phenotype
RASGRP2	97.5	99	98	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	104.1	99	94	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RECQL4	149.6	99	96	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RELB	90.6	87	75	?Immunodeficiency 53,617585
RFX5	116.9	98	96	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	91	Bare lymphocyte syndrome, type II, complementation group D, 209920
RHOH	134.9	100	100	No OMIM phenotype RHOH deficiency (Crequer (2012) J Clin Invest 122,3239)
RMRP				Anauxetic dysplasia, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	142.1	100	99	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	103.8	93	87	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	209.2	100	99	Aicardi-Goutieres syndrome 3, 610329
RNF168	215.3	100	99	RIDDLE syndrome, 611943
RNF31	154.7	99	98	No OMIM phenotype Autoinflammation, immunodeficiency, amylopectinosis and lymphangiectasia (Boisson (2015) J Exp Med 212,939)
RNU4ATAC				Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
RORC	132.3	100	100	Immunodeficiency 42, 616622
RPSA	88.8	100	99	Asplenia, isolated congenital, 271400
RSPH9	127.4	100	99	Ciliary dyskinesia, primary, 12, 612650
RTEL1	110.9	99	95	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	99	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	165.7	100	99	Ataxia-pancytopenia syndrome, 159550
SAMHD1	127.9	99	96	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SBDS	212.3	100	99	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SEMA3E	142.6	99	99	?CHARGE syndrome, 214800
SERAC1	112.5	98	94	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	97.9	97	92	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SH2B3	97.5	90	79	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450

				Thrombocythemia, somatic, 187950
SH2D1A	104.7	89	89	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	110.9	91	91	Cherubism, 118400
SKIV2L	149.1	100	99	Trichohepatoenteric syndrome 2, 614602
SLC29A3	203.6	99	99	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	124	99	97	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	230.2	99	98	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	140.2	100	99	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	81.8	99	96	Acrodermatitis enteropathica, 201100
SLC46A1	106	99	96	Folate malabsorption, hereditary, 229050
SMARCAL1	134.6	100	99	Schimke immunoosseous dysplasia, 242900
SMARCD2	92.9	87	85	Specific granule deficiency 2, 617475
SNX10	118.9	96	96	Osteopetrosis, autosomal recessive 8, 615085
SOCS4	262.8	99	99	No OMIM phenotype Autoimmunity (Arts (2015) J Intern Med epub,epub)
SP110	121.6	100	99	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SPINK5	145	99	96	Atopy, 147050 Netherton syndrome, 256500
STAT1	126.2	98	95	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	116	100	99	Immunodeficiency 44, 616636
STAT3	119.5	99	99	Autoimmune disease, multisystem, infantile-onset, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT4	144.2	98	97	{Systemic lupus erythematosus, susceptibility to, 11}, 612253
STAT5B	130.6	99	97	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STAT6	119.3	100	99	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	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1 160565

				Stormorken syndrome, 185070
STK4	138.9	100	99	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	311.4	100	100	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	102.3	88	83	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TAP1	103.3	100	99	Bare lymphocyte syndrome, type I, 604571
TAP2	95.2	99	98	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis
TAPBP	100.7	96	94	Bare lymphocyte syndrome, type I, 604571
TAZ	94	99	98	Barth syndrome, 302060
TBX1	75.3	77	67	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TCF3	67.8	98	92	Agammaglobulinemia 8,autosomal dominant,616941
TCIRG1	113.5	95	89	Osteopetrosis, autosomal recessive 1, 259700
TCN2	175.6	100	100	Transcobalamin II deficiency, 275350
TERC				Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERT	138.3	95	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	99	Immunodeficiency 46, 616740
THBD	108.2	99	97	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TICAM1	111.1	100	99	{Herpes simplex encephalitic, 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	98	{Herpes simplex encephalitis, susceptibility to, 2} 613002 {HIV1 infection, resistance to}, 609423
TLR4	132.7	100	99	Endotoxin hyporesponsiveness {Colorectal cancer, susceptibility to}, 114500 {Macular degeneration, age-related, 10}, 611488
TMC6	83.7	99	99	Epidermodysplasia verruciformis, 226400
TMC8	108.1	97	91	Epidermodysplasia verruciformis, 226400
TMEM173	100.8	98	93	STING-associated vasculopathy, infantile-onset, 615934
TNFAIP3	135.9	100	99	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF11A	146.3	93	91	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF13B	102.1	100	99	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	55.8	76	66	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	93.2	90	87	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFRSF4	51.6	97	85	?Immunodeficiency 16, 615593
TNFSF11	150.4	99	93	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	77.5	94	90	No OMIM phenotype Antibody deficiency (Wang (2013) Proc Natl Acad Sci USA 110, 5127)
TPP2	119.2	98	94	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	98	{?Herpes simplex encephalitis, susceptibility to, 3}, 614849
TRAF3IP2	116.6	99	97	?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	92	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084

TTC37	124	99	98	Trichohepatoenteric syndrome 1, 222470
TTC7A	123	99	98	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	119.2	99	98	Immunodeficiency 35, 611521
UNC13D	97	99	97	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	60	56	54	{Herpes simplex encephalitis, susceptibility to, 1}, 610551
UNG	78.4	99	94	Immunodeficiency with hyper IgM, type 5, 608106
USB1	125	99	98	Poikiloderma with neutropenia,604173
USP18	201.4	95	95	Pseudo-TORCH syndrome 2, 617397
VAV1	105.7	98	94	No OMIM phenotype
VPS13B	143.8	98	96	Cohen syndrome, 216550
VPS45	131.5	96	94	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	66.1	88	78	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WDR1	114.1	100	99	No OMIM phenotype
WIPF1	77.5	100	99	?Wiskott-Aldrich syndrome 2, 614493
WRAP53	154.4	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	107.1	91	86	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	185.6	99	99	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 : April 18th, 2018.
This list is accurate for panel version DG 2.13
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors