PRIMARY IMMUNODEFICIENCIES GENE PANEL DG 2.9

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACD	139.5	100%	99%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ACP5	231.8	100%	99%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	118.8	98%	94%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ADA	128.2	99%	98%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADAM17	152.2	99%	95%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAR	136.8	100%	99%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	175.8	100%	100%	Aspartylglucosaminuria, 208400
AICDA	149.7	98%	90%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	83.4	99%	97%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	110.8	99%	94%	Reticular dysgenesis, 267500
ALG13	104.2	99%	96%	Epileptic encephalopathy, early infantile, 36, 300884
AP3B1	128.1	99%	95%	Hermansky-Pudlak syndrome 2, 608233
APOL1	194.7	100%	100%	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
ATM	132.3	99%	96%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480
BLM	139.5	99%	97%	Bloom syndrome, 210900
BLNK	120.2	95%	90%	Agammaglobulinemia 4, 613502
BLOC1S6	111.3	99%	96%	Hermansky-pudlak syndrome 9, 614171
BTK	139.7	100%	99%	Agammaglobulinemia and isolated hormone deficiency, 307200 Agammaglobulinemia, X-linked 1, 300755

C1QA	147.6	100%	98%	C1q deficiency, 613652
C1QB	225.6	100%	100%	C1q deficiency, 613652
C1QC	215.1	100%	99%	C1q deficiency, 613652
C1R	171.2	100%	100%	C1r/C1s deficiency, combined, 216950
C1S	138	100%	99%	C1s deficiency, 613783
C2	20.1	82%	41%	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C3	167.5	100%	99%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	22.1	76%	46%	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	20.5	73%	43%	C4B deficiency, 614379
C5	149.2	98%	96%	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C6	185.4	100%	99%	C6 deficiency, 612446 Combined C6/C7 deficiency
C7	159	99%	96%	C7 deficiency, 610102
C8A	134.3	100%	99%	C8 deficiency, type I, 613790
C8B	154.8	100%	99%	C8 deficiency, type II, 613789
C9	169.7	100%	99%	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
CARD11	176.4	99%	97%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11, 615206
CARD14	130.2	99%	98%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	129.1	99%	96%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	139.2	95%	93%	No OMIM phenotype Immunodeficiency, combined (Wang (2016) J Exp Med 213,2413) Immunodeficiency, primary (Schober (2017) Nat Commun 8,14209)
CASP10	127.9	99%	97%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP8	172.1	100%	99%	Hepatocellular carcinoma, somatic, 114550

				?Autoimmune lymphoproliferative syndrome, type IIB, 607271 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CD19	105.5	100%	99%	Immunodeficiency, common variable, 3, 613493
CD247	104.3	100%	99%	?Immunodeficiency 25, 610163
CD27	138.1	100%	99%	Lymphoproliferative syndrome 2, 615122
CD3D	211.9	100%	100%	Immunodeficiency 19, 615617
CD3E	171.6	100%	99%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	195.3	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	196.1	100%	99%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	150.6	98%	90%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	157.5	99%	95%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD55	167.3	92%	85%	[Blood group Cromer], 613793
CD59	211.7	94%	88%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	155.7	99%	98%	Agammaglobulinemia 3, 613501
CD79B	268.6	100%	100%	Agammaglobulinemia 6, 612692
CD81	168.9	100%	99%	Immunodeficiency, common variable, 6, 613496
CD8A	117.9	99%	99%	CD8 deficiency, familial, 608957
CDCA7	129.5	99%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDKN2B	75.9	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	84.5	100%	99%	Specific granule deficiency, 245480
CECR1	106	99%	98%	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410
CFB	23	84%	51%	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFD	76.4	95%	87%	Complement factor D deficiency, 613912
CFH	216.6	99%	97%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814

				{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	202.7	92%	90%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	101.2	88%	83%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR5	127.2	98%	95%	Nephropathy due to CFHR5 deficiency, 614809
CFI	198	98%	97%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFP	109.2	99%	95%	Properdin deficiency,X-linked, 312060
CFTR	158.3	99%	97%	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	168.4	100%	99%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CIITA	140.9	100%	99%	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CLEC4D	159.2	100%	100%	No OMIM phenotype
CLEC7A	177.5	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
COLEC11	221.8	100%	99%	3MC syndrome 2, 265050
COPA	144.7	100%	99%	{Autoimmune interstitial lung, joint, and kidney disease}, 616414
CORO1A	187	99%	96%	Immunodeficiency 8, 615401
CR2	192	100%	99%	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CREBBP	141.3	98%	96%	Rubinstein-Taybi syndrome, 180849
CSF2RA	68.1	90%	88%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF3R	101.3	99%	96%	?Neutrophilia, hereditary, 162830
CTC1	122.4	99%	99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	212.7	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
CTSC	142.5	100%	100%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CXCR4	182.7	100%	99%	Myelokathexis, isolated WHIM syndrome, 193670
CYBA	101.8	88%	80%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	132.1	99%	99%	Chronic granulomatous disease, X-linked, 306400 Immunodeficiency 34, mycobacteriosis, X-linked, 300645
DCLRE1C	158.5	98%	95%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDX58	147.7	99%	98%	Singleton-Merten syndrome 2, 616298
DHFR	61.8	95%	81%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	122.5	100%	98%	Dyskeratosis congenita, X-linked, 305000
DNASE1	230.9	100%	100%	{Systemic lupus erythematosus, susceptibility to}, 152700
DNMT3B	143.6	99%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	165.9	100%	99%	Immunodeficiency 40, 616433
DOCK8	148.8	100%	99%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	115.1	99%	98%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	85.7	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	138.8	99%	98%	Vici syndrome, 242840
ERCC2	143	100%	99%	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	120.8	100%	99%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
F12	131.4	100%	99%	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
FADD	140.3	99%	99%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759

FAM105B	177	95%	89%	Autoinflammation,panniculitis, and dermatosis syndrome, 617099
FAS	294.7	100%	99%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic {Autoimmune lymphoproliferative syndrome}, 601859
FASLG	98.6	99%	98%	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FCGR1A	77.7	47%	45%	[IgG receptor I, phagocytic, familial deficiency of]
FCGR2A	237.8	100%	100%	{Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162 {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700
FCGR2B	157.9	99%	96%	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700
FCGR3A	242.2	100%	99%	Immunodeficiency 20, 615707
FCGR3B	180.2	99%	98%	Neutropenia, alloimmune neonatal
FCN3	145.2	100%	99%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	139.2	99%	98%	Leukocyte adhesion deficiency,type III,612840
FOXN1	127.1	100%	99%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	130.9	98%	92%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FPR1	220	100%	99%	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	201.1	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	143.5	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	138.1	99%	96%	Favism, 134700 Hemolytic anemia due to G6PD deficiency, 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GATA2	145.4	100%	99%	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GFI1	99.4	99%	96%	Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107

GJC2	50	88%	71%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GRHL2	152.8	100%	100%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GTF2H5	152.5	100%	99%	Trichothiodystrophy 3, photosensitive, 616395
HAX1	157.7	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HELLS	110.7	96%	90%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4,616911
ICOS	186.3	100%	100%	Immunodeficiency, common variable, 1, 607594
IFIH1	143.7	99%	98%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNGR1	171.1	99%	98%	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	164	94%	93%	Immunodeficiency 28, mycobacteriosis, 614889
IGHM	204.2	100%	100%	Agammaglobulinemia 1, 601495
IGLL1	82.2	99%	95%	Agammaglobulinemia 2, 613500
IKBKB	129.9	98%	95%	Immunodeficiency 15, 615592
IKBKG	57.4	83%	72%	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	202.5	100%	99%	Immunodeficiency,common variable, 1,616873
IL10RA	164.2	100%	99%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	214.6	99%	97%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL12B	122.6	100%	99%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	138.8	98%	96%	Immunodeficiency 30, 614891
IL17F	92.7	99%	95%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	145.2	99%	97%	Immunodeficiency 51, 613953

IL1RN	174.7	100%	100%	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL2	86.2	97%	89%	Severe combined immunodeficiency due to IL2 deficiency
IL21R	149.6	100%	100%	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 [IgE, elevated level of], 147050
IL2RA	140	100%	99%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	75.4	99%	97%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL36RN	94.7	100%	99%	Psoriasis 14, pustular, 614204
IL7R	162.6	100%	99%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INSR	151.6	97%	94%	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
IRAK4	110.5	99%	95%	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676
IRF7	113.3	100%	99%	?Immunodeficiency 39, 616345
IRF8	124.4	99%	98%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 614894
ISG15	183.3	100%	100%	Immunodeficiency 38, 616126
ITCH	143	95%	95%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGB2	181.5	100%	99%	Leukocyte adhesion deficiency, 116920
ITK	150.8	100%	99%	Lymphoproliferative syndrome 1, 613011
JAGN1	166.5	100%	100%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK2	113.7	96%	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	113.3	98%	95%	SCID, autosomal recessive, T-negative/B-positive type, 600802
KMT2D	158.6	100%	99%	Kabuki syndrome 1, 147920
KRAS	89.6	99%	99%	Bladder cancer, somatic, 109800

				Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
LAMTOR2	204.7	100%	99%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LCK	183.6	98%	95%	?Immunodeficiency 22, 615758
LIG1	106.1	99%	98%	DNA ligase I deficiency
LIG4	207.5	100%	99%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LPIN2	124.6	100%	99%	Majeed syndrome, 609628
LRBA	164.3	99%	98%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	300.4	100%	99%	Agammaglobulinemia 5, 613506
LTBP3	128.7	98%	97%	Dental anomalies and short stature, 601216
LYST	171	98%	96%	Chediak-Higashi syndrome, 214500
MAGT1	118.7	98%	97%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAL	159.5	100%	100%	No OMIM phenotype
MALT1	153.1	90%	87%	Immunodeficiency 12, 615468
MAN2B1	134.6	98%	95%	Mannosidosis, alpha-, types I and II, 248500
MANBA	153.6	99%	98%	Mannosidosis, beta, 248510
MASP2	179.7	100%	99%	MASP2 deficiency, 613791
MBL2	136.9	100%	99%	{Chronic infections, due to MBL deficiency}, 614372
MC2R	201.9	100%	99%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM4	175.6	100%	99%	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MEFV	135.7	96%	92%	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MKL1	108.9	99%	95%	Megakaryoblastic leukemia, acute
MPO	160.6	100%	99%	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}

MRE11A	64.3	97%	89%	Ataxia-telangiectasia-like disorder, 604391
MS4A1	145.2	99%	97%	Immunodeficiency, common variable, 5, 613495
MTHFD1	159.4	99%	98%	{Abruptio placentae, susceptibility to} {Spina bifida, folate-sensitive, susceptibility to}, 601634
MVK	167.2	100%	99%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYD88	195.9	100%	99%	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
NBN	106	99%	98%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCF1	23.9	26%	22%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	145.9	99%	97%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	188.3	100%	100%	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	129.2	100%	99%	Acne inversa, familial, 1, 142690
NFKB1	124.4	99%	96%	Immunodeficiency, common variable, 12, 616576
NFKB2	146.9	99%	96%	Immunodeficiency, common variable, 10, 615577
NFKBIA	126	98%	93%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NHEJ1	85.5	100%	99%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	101.6	100%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987
NKX2-5	96.3	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	202.8	100%	99%	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
NLRP1	144.7	99%	96%	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP12	184.3	99%	99%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	162.9	100%	99%	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100

				Muckle-Wells syndrome, 191900
NOD2	149.9	100%	99%	Blau syndrome, 186580 Yao syndrome, 617321 {Inflammatory bowel disease 1}, 266600 {Psoriatic arthritis, susceptibility to}, 607507
NOP10	159.6	100%	99%	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	203.3	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	NC	NC	NC	Lung disease,immunodeficiency and chromosome breakage syndrome, 617241
ORAI1	243	94%	92%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
PARN	151.5	100%	99%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX5	127.9	98%	96%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PBX1	133	99%	97%	Leukemia, acute pre-B-cell, 176310
PCCA	120.1	98%	93%	Propionicacidemia, 606054
PCCB	167.8	98%	96%	Propionicacidemia, 606054
PEPD	127	99%	98%	Prolidase deficiency, 170100
PGM3	225	100%	100%	Immunodeficiency 23, 615816
PIGA	97	94%	86%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIK3CD	159.5	99%	97%	Immunodeficiency 14, 615513
PIK3R1	158.1	99%	98%	Immunodeficiency 36, 616005 SHORT syndrome, 269880 ?Agammaglobulinemia 7, autosomal recessive, 615214
PLCG2	130.6	100%	99%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLG	138.6	87%	87%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090

PMM2	171.3	100%	99%	Congenital disorder of glycosylation, type Ia, 212065
PNP	147.3	100%	99%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POT1	125.8	99%	98%	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
PRF1	128.8	100%	99%	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKCD	204.8	100%	100%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	120.2	99%	96%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	178.9	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	102.7	100%	100%	Acne inversa, familial, 2, 613736
PSMB8	16	61%	25%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSTPIP1	104.7	99%	97%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN11	105.7	98%	93%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN22	162.4	99%	94%	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700
PTPRC	127.6	96%	89%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitic C virus, susceptibility to}, 609532
PTRF	162.4	99%	99%	Lipodystrophy, congenital generalized, type 4, 613327
RAB27A	171.6	100%	100%	Griscelli syndrome, type 2, 607624
RAC2	135.4	100%	99%	Neutrophil immunodeficiency syndrome, 608203
RAG1	208.9	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	271.4	100%	100%	Combined cellular and humoral immune defects with granulomas, 233650

				Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RASGRP2	109.7	99%	98%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	112	98%	95%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RECQL4	152.3	99%	98%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RFX5	131.9	98%	96%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	121.5	100%	99%	MHC class II deficiency, complementation group B, 209920
RFXAP	124.3	96%	94%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RHOH	143.3	100%	100%	No OMIM phenotype RHOH deficiency (Crequer (2012) J Clin Invest 122,3239)
RMRP	NC	NC	NC	Anauxetic dysplasia, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	157.1	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	128.3	98%	92%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	244.3	99%	98%	Aicardi-Goutieres syndrome 3, 610329
RNF168	293.7	100%	99%	RIDDLE syndrome, 611943
RNF31	162.3	99%	97%	No OMIM phenotype Autoinflammation, immunodeficiency, amylopectinosis and lymphangiectasia (Boisson (2015) J Exp Med 212,939)
RPSA	89	100%	99%	Asplenia, isolated congenital, 271400
RSPH9	156.2	99%	98%	Ciliary dyskinesia, primary, 12, 612650
RTEL1	137.2	99%	97%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
SAMHD1	154.9	99%	98%	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SBDS	231.2	100%	99%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SERAC1	139.3	99%	96%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	98.9	98%	94%	Angioedema, hereditary, types I and II, 106100

				Complement component 4, partial deficiency of, 120790
SH2B3	113.1	95%	84%	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SH2D1A	127.3	92%	89%	Lymphoproliferative syndrome, X-linked, 1, 308240
SKIV2L	27	81%	57%	Trichohepatoenteric syndrome 2, 614602
SLC29A3	236.3	99%	99%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	169.7	100%	99%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	244.4	99%	98%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	139.2	100%	100%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	93.7	99%	96%	Acrodermatitis enteropathica, 201100
SLC46A1	103.2	99%	95%	Folate malabsorption, hereditary, 229050
SMARCAL1	153.8	100%	99%	Schimke immunoosseous dysplasia, 242900
SOCS4	282.6	99%	98%	No OMIM phenotype Autoimmunity (Arts (2015) J Intern Med epub,epub)
SP110	148.5	100%	100%	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SPINK5	182.2	99%	98%	Atopy, 147050 Netherton syndrome, 256500
STAT1	161.2	98%	96%	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	125.3	100%	99%	Immunodeficiency 44, 616636
STAT3	132.1	100%	99%	Autoimmune disease, multisystem, infantile-onset, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT4	189.9	99%	99%	{Systemic lupus erythematosus, susceptibility to, 11}, 612253
STAT5B	140.5	99%	96%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STAT6	122.6	99%	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	144	99%	97%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1 160565

				Stormorken syndrome, 185070
STK4	162.5	100%	99%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	348.1	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	144.3	99%	98%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TAP1	13.1	52%	18%	Bare lymphocyte syndrome, type I, 604571
TAP2	12.6	50%	20%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis
TAPBP	23.6	83%	53%	Bare lymphocyte syndrome, type I, 604571
TAZ	123.4	99%	98%	Barth syndrome, 302060
TBX1	90.6	79%	70%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TCIRG1	127.8	96%	89%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	201.5	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	150	96%	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	190.2	100%	99%	Immunodeficiency 46, 616740
THBD	129.5	99%	98%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TICAM1	110	100%	99%	{Herpes simplex encephalitic, susceptibility to, 6}, 614850
TINF2	208.7	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TIRAP	156.4	100%	100%	{Bacteremia, protection against}, 614382 {Malaria, protection against}, 611162 {Pneumococcal disease, invasive, protection against}, 610799 {Tuberculosis, protection against}, 607948
TLR3	208.7	99%	99%	{Herpes simplex encephalitis, susceptibility to, 2} 613002 {HIV1 infection, resistance to}, 609423

TLR4	161.4	99%	97%	Endotoxin hyporesponsiveness {Colorectal cancer, susceptibility to}, 114500 {Macular degeneration, age-related, 10}, 611488
TMC6	88.2	99%	97%	Epidermodysplasia verruciformis, 226400
TMC8	128.1	98%	94%	Epidermodysplasia verruciformis, 226400
TMEM173	106.3	99%	96%	STING-associated vasculopathy, infantile-onset, 615934
TNFAIP3	159.5	100%	100%	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF11A	152.6	94%	91%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF13B	115.4	100%	99%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	57.8	89%	71%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	96.7	91%	89%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFRSF4	73.6	99%	92%	?Immunodeficiency 16, 615593
TPP2	144	98%	95%	No OMIM phenotype Evans syndrome, immunodeficiency and premature immunosenescence (Stepensky (2015) Blood 125, 753)
TRAF3	139.5	99%	97%	{?Herpes simplex encephalitis, susceptibility to, 3}, 614849
TRAF3IP2	129.4	99%	97%	?Candidiasis, familial, 8, 615527 {Psoriasis susceptibility 13}, 614070
TREX1	302.9	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	123.4	98%	94%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TTC37	159.2	99%	98%	Trichohepatoenteric syndrome 1, 222470
TTC7A	131.2	99%	98%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	136.2	99%	99%	Immunodeficiency 35, 611521
UNC119	103	99%	93%	?Cone-rod dystrophy ?Immunodeficiency 13, 615518
UNC13D	111.2	99%	97%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898

UNC93B1	74	58%	55%	{Herpes simplex encephalitis, susceptibility to, 1}, 610551
UNG	84.6	99%	96%	Immunodeficiency with hyper IgM, type 5, 608106
USB1	157.7	99%	97%	Poikiloderma with neutropenia,604173
VPS13B	170.3	99%	97%	Cohen syndrome, 216550
VPS45	158.8	97%	95%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	72	89%	80%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WIPF1	88.4	100%	99%	?Wiskott-Aldrich syndrome 2, 614493
WRAP53	175.2	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	123	93%	90%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	207.8	100%	99%	Autoimmune disease,multisystem,infantile-onset,2,617006 Immunodeficiency 48,269840
ZBTB24	213.6	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 14th 2017
This list is accurate for panel version DG 2.9

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors