PRIMARY IMMUNODEFICIENCIES GENE PANEL DG 2.17 (393 genes)

Releasedate: 06-12-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACD	180.3	100.0%	100.0%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACP5	189.0	100.0%	99.9%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	92.6	100.0%	99.9%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADA	111.3	100.0%	99.6%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	88.7	99.8%	97.9%	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	117.8	99.7%	98.5%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAR	117.2	99.9%	99.4%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	144.3	100.0%	100.0%	Aspartylglucosaminuria, 208400
AICDA	141.1	100.0%	99.6%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	113.3	100.0%	100.0%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	100.4	98.7%	94.5%	Reticular dysgenesis, 267500
ALG13	77.7	98.6%	92.4%	Epileptic encephalopathy, early infantile, 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
AP1S3	110.7	90.5%	90.4%	No OMIM disease ID
AP3B1	108.2	99.4%	95.7%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	135.1	98.5%	97.9%	?Hermansky-Pudlak syndrome 10, 617050
APOL1	157.3	100.0%	100.0%	No OMIM disease ID
ARHGEF1	117.1	100.0%	99.6%	?Immunodeficiency 62, 618459
ARPC1B	150.6	100.0%	100.0%	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
ATM	108.4	99.6%	96.8%	Ataxia-telangiectasia, 208900 Lymphoma, mantle cell, somatic, 0 Lymphoma, B-cell non-Hodgkin, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0
ATP6AP1	113.6	99.8%	97.7%	Immunodeficiency 47, 300972

B2M	194.6	100.0%	100.0%	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
BACH2	173.7	100.0%	99.8%	Immunodeficiency 60, 618394
BCL10	127.1	100.0%	100.0%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245
BCL11B	147.9	100.0%	99.3%	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BLK	137.8	100.0%	100.0%	Maturity-onset diabetes of the young, type 11, 613375
BLM	111.3	99.9%	98.1%	Bloom syndrome, 210900
BLNK	93.3	96.9%	92.4%	?Agammaglobulinemia 4, 613502
BLOC1S6	103.0	99.3%	92.1%	?Hermansky-pudlak syndrome 9, 614171
BTK	98.6	100.0%	99.1%	Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 Agammaglobulinemia, X-linked 1, 300755
C17orf62	148.0	100.0%	99.9%	No OMIM Disease ID
C1QA	222.4	100.0%	100.0%	C1q deficiency, 613652
C1QB	178.7	100.0%	99.9%	C1q deficiency, 613652
C1QC	209.9	100.0%	100.0%	C1q deficiency, 613652
C1R	161.6	100.0%	100.0%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	101.7	99.9%	98.4%	Ehlers-Danlos syndrome, periodontal type, 2, 617174 C1s deficiency, 613783
C2	134.5	100.0%	100.0%	C2 deficiency, 217000
C3	153.6	100.0%	99.7%	C3 deficiency, 613779
C5	118.6	99.6%	97.6%	C5 deficiency, 609536
C6	139.6	100.0%	99.7%	C6 deficiency, 612446 Combined C6/C7 deficiency, 0
C7	113.0	99.9%	98.0%	C7 deficiency, 610102
C8A	108.7	100.0%	99.5%	C8 deficiency, type I, 613790
C8B	108.4	99.9%	98.7%	C8 deficiency, type II, 613789
C8G	181.2	100.0%	100.0%	No OMIM Disease ID
C9	120.8	100.0%	99.5%	C9 deficiency, 613825
CA2	141.8	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CARD11	148.2	100.0%	99.8%	Immunodeficiency 11B with atopic dermatitis, 617638 B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206
CARD14	136.0	100.0%	99.5%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	152.2	100.0%	99.9%	Candidiasis, familial, 2, autosomal recessive, 212050

CARMIL2	153.2	99.5%	98.0%	Immunodeficiency 58, 618131
CASP10	111.4	99.9%	98.9%	Lymphoma, non-Hodgkin, somatic, 605027 Gastric cancer, somatic, 613659 Autoimmune lymphoproliferative syndrome, type II, 603909
CASP8	133.1	95.6%	95.5%	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550
CAVIN1	200.3	100.0%	100.0%	Lipodystrophy, congenital generalized, type 4, 613327
CCBE1	80.9	99.8%	98.6%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD19	116.9	100.0%	100.0%	Immunodeficiency, common variable, 3, 613493
CD247	99.1	99.9%	99.2%	?Immunodeficiency 25, 610163
CD27	115.3	100.0%	99.9%	Lymphoproliferative syndrome 2, 615122
CD3D	146.3	100.0%	99.9%	Immunodeficiency 19, 615617
CD3E	126.6	100.0%	99.6%	Immunodeficiency 18, SCID variant, 615615 Immunodeficiency 18, 615615
CD3G	137.2	100.0%	100.0%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	157.9	100.0%	100.0%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	109.8	97.1%	88.0%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	126.4	100.0%	99.1%	No OMIM disease ID
CD55	133.3	95.6%	91.0%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	153.6	94.1%	86.4%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	117.0	100.0%	99.7%	Lymphoproliferative syndrome 3, 618261
CD79A	144.2	100.0%	99.1%	Agammaglobulinemia 3, 613501
CD79B	212.9	100.0%	100.0%	Agammaglobulinemia 6, 612692
CD81	174.0	100.0%	100.0%	Immunodeficiency, common variable, 6, 613496
CD8A	166.1	100.0%	100.0%	CD8 deficiency, familial, 608957
CDCA7	117.7	100.0%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDKN2B	136.2	100.0%	100.0%	No OMIM Disease ID
CEBPE	113.9	100.0%	100.0%	Specific granule deficiency, 245480
CFB	125.5	100.0%	100.0%	?Complement factor B deficiency, 615561
CFD	128.9	97.9%	92.1%	Complement factor D deficiency, 613912
CFH	148.8	99.4%	97.4%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	156.0	94.1%	92.0%	No OMIM disease ID
CFHR2	129.8	99.7%	96.4%	No OMIM disease ID
CFHR3	89.5	93.2%	91.0%	No OMIM disease ID
CFHR4	119.0	100.0%	99.9%	No OMIM disease ID
CFHR5	91.5	99.9%	97.2%	Nephropathy due to CFHR5 deficiency, 614809

CFI	137.6	99.2%	96.8%	Complement factor I deficiency, 610984
CFP	107.2	99.8%	98.3%	Properdin deficiency, X-linked, 312060
CFTR	112.9	99.4%	97.3%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF, 0
CHD7	143.6	100.0%	99.5%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CIITA	166.3	100.0%	100.0%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CLCN7	162.0	99.9%	98.9%	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600 Hypopigmentation, organomegaly, and delayed myelination and development, 618541
CLEC4D	135.0	100.0%	100.0%	No OMIM Disease ID
CLEC7A	146.7	100.0%	100.0%	Candidiasis, familial, 4, autosomal recessive, 613108
CLPB	135.3	99.7%	97.4%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COLEC11	197.8	100.0%	100.0%	3MC syndrome 2, 265050
COPA	111.2	100.0%	99.6%	No OMIM disease ID
CORO1A	166.6	100.0%	99.2%	Immunodeficiency 8, 615401
CR2	135.0	100.0%	100.0%	Immunodeficiency, common variable, 7, 614699
CREBBP	120.9	99.6%	97.3%	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CSF2RA	56.6	90.0%	88.2%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	136.0	99.9%	98.7%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	116.5	100.0%	99.2%	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	113.5	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	146.7	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type V, 616100
CTPS1	112.2	100.0%	99.9%	Immunodeficiency 24, 615897
CTSC	119.4	100.0%	100.0%	Periodontitis 1, juvenile, 170650 Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010
CXCR4	127.9	100.0%	99.9%	WHIM syndrome, 193670 Myelokathexis, isolated, 0
CYBA	119.0	97.4%	89.2%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	98.6	99.9%	99.1%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400
DCLRE1C	139.2	99.9%	98.0%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDX58	111.6	99.9%	98.4%	Singleton-Merten syndrome 2, 616298

DHFR	48.6	92.6%	80.9%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	93.9	99.7%	98.0%	Dyskeratosis congenita, X-linked, 305000
DNASE1	184.5	100.0%	100.0%	No OMIM disease ID
DNASE1L3	118.0	99.9%	99.7%	Systemic lupus erythematosus 16, 614420
DNASE2	100.7	99.3%	97.2%	No OMIM Disease ID
DNMT3B	125.5	100.0%	99.9%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	123.0	100.0%	99.5%	Immunodeficiency 40, 616433
DOCK8	115.2	100.0%	99.7%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	156.3	100.0%	99.8%	Neutropenia, severe congenital 1, autosomal dominant, 202700 Neutropenia, cyclic, 162800
ELF4	98.8	99.9%	99.2%	No OMIM Disease ID
EPG5	111.4	99.5%	98.3%	Vici syndrome, 242840
ERCC2	139.5	100.0%	99.9%	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	95.9	99.9%	98.7%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
EXTL3	200.7	100.0%	100.0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
F12	171.1	100.0%	99.7%	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
FAAP24	116.5	99.9%	97.5%	No OMIM Disease ID
FADD	201.3	100.0%	100.0%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAS	228.0	100.0%	99.9%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0
FASLG	84.8	100.0%	99.1%	Autoimmune lymphoproliferative syndrome, type IB, 601859
FAT4	195.5	100.0%	100.0%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FCGR1A	56.3	47.7%	45.9%	No OMIM Disease ID
FCGR2A	173.8	100.0%	100.0%	No OMIM Disease ID
FCGR2B	131.3	99.7%	97.4%	No OMIM Disease ID
FCGR3A	184.5	99.7%	98.1%	Immunodeficiency 20, 615707
FCGR3B	147.4	99.1%	98.5%	Neutropenia, alloimmune neonatal, 0
FCN3	135.3	100.0%	100.0%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	161.2	100.0%	99.9%	Leukocyte adhesion deficiency, type III, 612840
FOXN1	149.7	100.0%	99.8%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	126.8	99.3%	96.0%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FPR1	155.9	100.0%	99.9%	No OMIM Disease ID

G6PC	149.8	100.0%	100.0%	Glycogen storage disease Ia, 232200
G6PC3	126.1	100.0%	100.0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	126.2	99.8%	98.4%	Hemolytic anemia, G6PD deficient (favism), 300908
GATA2	128.7	100.0%	99.7%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GFI1	118.9	100.0%	99.9%	?Neutropenia, severe congenital 2, autosomal dominant, 613107 ?Neutropenia, nonimmune chronic idiopathic, of adults, 607847
GINS1	126.5	98.9%	89.5%	Immunodeficiency 55, 617827
GJC2	59.7	97.7%	86.5%	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GRHL2	119.8	100.0%	100.0%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GTF2H5	82.0	99.8%	97.4%	Trichothiodystrophy 3, photosensitive, 616395
HAX1	146.3	100.0%	100.0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HELLS	102.2	98.1%	91.0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HMOX1	153.8	98.0%	91.0%	Heme oxygenase-1 deficiency, 614034
HYOU1	140.5	100.0%	99.7%	?Immunodeficiency 59 and hypoglycemia, 233600
ICOS	154.5	100.0%	99.9%	Immunodeficiency, common variable, 1, 607594
IFIH1	110.9	99.8%	98.2%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNAR2	133.4	99.6%	97.5%	?Immunodeficiency 45, 616669
IFNGR1	145.4	100.0%	99.2%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978
IFNGR2	129.9	97.9%	94.1%	Immunodeficiency 28, mycobacteriosis, 614889
IGHM	190.2	100.0%	100.0%	Agammaglobulinemia 1, 601495
IGLL1	99.7	100.0%	99.8%	Agammaglobulinemia 2, 613500
IKBKB	116.2	99.4%	97.1%	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	64.7	90.1%	80.2%	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Immunodeficiency, isolated, 300584 Ectodermal dysplasia and immunodeficiency 1, 300291 Invasive pneumococcal disease, recurrent isolated, 2, 300640 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301

IKZF1	196.9	100.0%	100.0%	Immunodeficiency, common variable, 13, 616873
IL10	100.1	99.8%	96.4%	No OMIM Disease ID
IL10RA	158.7	100.0%	99.9%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	132.0	100.0%	99.6%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL12B	99.2	99.9%	97.5%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	119.9	98.2%	95.7%	Immunodeficiency 30, 614891
IL17F	76.7	98.8%	93.0%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	167.0	100.0%	100.0%	Immunodeficiency 51, 613953
IL17RC	139.8	100.0%	100.0%	Candidiasis, familial, 9, 616445
IL1RN	145.3	100.0%	99.9%	Interleukin 1 receptor antagonist deficiency, 612852
IL2	66.1	97.9%	87.9%	No OMIM Disease ID
IL21	74.7	99.9%	94.7%	?Immunodeficiency, common variable, 11, 615767
IL21R	160.2	100.0%	100.0%	Immunodeficiency 56, 615207
IL2RA	106.8	100.0%	99.2%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL2RG	60.2	99.7%	94.3%	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL36RN	102.7	100.0%	99.9%	Psoriasis 14, pustular, 614204
IL7R	114.7	100.0%	99.7%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INO80	97.9	99.9%	98.7%	No OMIM Disease ID
INSR	123.5	99.4%	96.1%	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200
IRAK1	92.6	99.9%	98.6%	No OMIM Disease ID
IRAK4	100.7	99.7%	94.9%	IRAK4 deficiency, 607676 Invasive pneumococcal disease, recurrent isolated, 1, 610799
IRF2BP2	98.4	100.0%	99.7%	?Immunodeficiency, common variable, 14, 617765
IRF3	154.7	100.0%	99.6%	No OMIM Disease ID
IRF4	209.2	100.0%	100.0%	No OMIM Disease ID
IRF7	174.6	100.0%	99.9%	?Immunodeficiency 39, 616345
IRF8	123.3	100.0%	99.1%	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893
ISG15	207.0	100.0%	100.0%	Immunodeficiency 38, 616126
ITCH	115.3	95.5%	94.5%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGB2	171.3	100.0%	100.0%	Leukocyte adhesion deficiency, 116920
ITK	105.3	99.9%	99.3%	Lymphoproliferative syndrome 1, 613011
JAGN1	129.4	100.0%	100.0%	Neutropenia, severe congenital, 6, autosomal recessive, 616022

JAK1	109.7	100.0%	99.6%	No OMIM Disease ID
JAK2	100.6	97.1%	95.0%	Myelofibrosis, somatic, 254450 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300 Leukemia, acute myeloid, somatic, 601626 Erythrocytosis, somatic, 133100
JAK3	134.3	98.7%	97.3%	SCID, autosomal recessive, T-negative/B-positive type, 600802
KDM6A	98.1	95.6%	87.7%	Kabuki syndrome 2, 300867
KMT2D	150.7	100.0%	99.9%	Kabuki syndrome 1, 147920
LACC1	143.6	99.9%	99.2%	No OMIM Disease ID
LAMTOR2	186.6	100.0%	100.0%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAT	127.2	100.0%	99.8%	Immunodeficiency 52, 617514
LCK	163.1	99.7%	98.3%	?Immunodeficiency 22, 615758
LIG1	115.4	100.0%	99.7%	No OMIM Disease ID
LIG4	170.5	100.0%	99.9%	LIG4 syndrome, 606593
LPIN2	101.1	100.0%	99.7%	Majeed syndrome, 609628
LRBA	127.6	100.0%	99.5%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	241.2	100.0%	100.0%	?Agammaglobulinemia 5, 613506
LTBP3	166.1	100.0%	100.0%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LYST	135.6	99.3%	97.1%	Chediak-Higashi syndrome, 214500
MAGT1	95.1	98.5%	95.0%	Congenital disorder of glycosylation, type Icc, 301031 Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAL2	171.3	100.0%	100.0%	No OMIM Disease ID
MALT1	128.7	93.6%	89.3%	Immunodeficiency 12, 615468
MAN2B1	139.1	99.9%	99.1%	Mannosidosis, alpha-, types I and II, 248500
MANBA	117.1	99.7%	98.1%	Mannosidosis, beta, 248510
MAP3K14	129.6	99.3%	99.3%	No OMIM Disease ID
MASP2	129.9	100.0%	99.4%	MASP2 deficiency, 613791
MBL2	96.5	100.0%	100.0%	No OMIM Disease ID
MC2R	160.2	100.0%	99.4%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM4	141.3	100.0%	99.5%	Immunodeficiency 54, 609981
MEFV	136.9	99.0%	97.0%	Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MKL1	131.1	98.0%	96.3%	Megakaryoblastic leukemia, acute, 0
MOGS	157.9	100.0%	100.0%	Congenital disorder of glycosylation, type IIb, 606056
MRE11	48.1	97.7%	83.5%	Ataxia-telangiectasia-like disorder 1, 604391

MS4A1	128.7	100.0%	99.1%	Immunodeficiency, common variable, 5, 613495
MSN	69.6	98.2%	92.3%	Immunodeficiency 50, 300988
MTHFD1	119.4	99.9%	98.3%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MVK	130.3	90.5%	90.4%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYD88	219.1	100.0%	99.9%	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 Macroglobulinemia, Waldenstrom, somatic, 153600
MYSM1	110.1	99.8%	98.6%	Bone marrow failure syndrome 4, 618116
NBAS	138.4	99.9%	99.2%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBN	90.6	100.0%	98.2%	Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065
NCF1	24.7	28.6%	22.6%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	112.5	100.0%	98.6%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	160.6	100.0%	100.0%	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	97.9	100.0%	99.7%	Acne inversa, familial, 1, 142690
NFAT5	176.5	99.8%	98.7%	No OMIM Disease ID
NFKB1	93.3	100.0%	98.6%	Immunodeficiency, common variable, 12, 616576
NFKB2	149.9	99.3%	97.1%	Immunodeficiency, common variable, 10, 615577
NFKBIA	148.5	96.7%	90.7%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHEJ1	60.2	99.4%	94.3%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	135.0	100.0%	99.8%	Dyskeratosis congenita, autosomal recessive 2, 613987
NLRC4	168.2	100.0%	99.9%	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
NLRP1	126.8	99.5%	97.9%	Palmoplantar carcinoma, multiple self-healing, 615225 Autoinflammation with arthritis and dyskeratosis, 617388
NLRP12	176.4	100.0%	100.0%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	146.0	100.0%	99.9%	Familial cold inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Keratoendothelitis fugax hereditaria, 148200
NLRP7	131.5	99.9%	98.9%	Hydatidiform mole, recurrent, 1, 231090
NOD2	136.5	100.0%	100.0%	Blau syndrome, 186580

NOP10	124.6	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	145.3	100.0%	100.0%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
NSMCE3	213.7	100.0%	100.0%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
ORAI1	226.3	100.0%	99.0%	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782
OSTM1	113.8	97.8%	92.1%	Osteopetrosis, autosomal recessive 5, 259720
OTULIN	137.5	99.1%	96.1%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
PARN	127.9	100.0%	99.6%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
PAX5	114.1	99.2%	96.0%	No OMIM disease ID
PBX1	115.4	100.0%	98.3%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	97.7	99.1%	95.4%	Propionicacidemia, 606054
PCCB	114.9	99.5%	97.1%	Propionicacidemia, 606054
PEPD	126.5	100.0%	99.9%	Prolidase deficiency, 170100
PGM3	148.4	100.0%	99.9%	Immunodeficiency 23, 615816
PIGA	72.9	93.0%	83.4%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIK3CD	173.9	99.8%	98.1%	Immunodeficiency 14, 615513
PIK3R1	125.7	99.9%	98.8%	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PLCG2	110.7	100.0%	99.5%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLEKHM1	139.5	100.0%	100.0%	Osteopetrosis, autosomal dominant 3, 618107 ?Osteopetrosis, autosomal recessive 6, 611497
PLG	95.7	87.8%	86.7%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PMM2	130.3	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PNP	113.1	100.0%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179

POLA1	104.7	99.1%	94.7%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLE2	66.8	98.0%	85.2%	No OMIM Disease ID
POMP	122.5	99.9%	95.8%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POT1	93.9	100.0%	98.7%	No OMIM disease ID
PRF1	154.3	91.2%	90.7%	Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027 Hemophagocytic lymphohistiocytosis, familial, 2, 603553
PRKCD	177.4	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	98.5	99.4%	96.4%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	113.2	100.0%	99.9%	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500 Arts syndrome, 301835 Gout, PRPS-related, 300661
PSENEN	98.3	100.0%	100.0%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMA3	69.3	99.8%	94.6%	No OMIM Disease ID
PSMB4	124.8	100.0%	99.8%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	119.3	100.0%	99.3%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	85.2	100.0%	99.1%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMG2	118.8	100.0%	99.3%	No OMIM Disease ID
PSTPIP1	114.3	99.9%	99.3%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN22	124.5	99.5%	95.6%	No OMIM disease ID
PTPRC	98.4	98.3%	93.6%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
RAB27A	123.3	100.0%	99.6%	Griscelli syndrome, type 2, 607624
RAC2	109.4	100.0%	99.4%	Neutrophil immunodeficiency syndrome, 608203
RAG1	158.7	100.0%	100.0%	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 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
RAG2	188.7	100.0%	100.0%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RANBP2	105.4	50.3%	49.4%	No OMIM disease ID
RASGRP1	112.5	100.0%	99.7%	Immunodeficiency 64, 618534

RASGRP2	111.5	100.0%	99.8%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	118.4	100.0%	99.4%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RECQL4	181.4	100.0%	100.0%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
RELB	120.3	99.7%	96.5%	?Immunodeficiency 53, 617585
RFX5	117.8	99.9%	98.2%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	133.2	100.0%	99.9%	MHC class II deficiency, complementation group B, 209920
RFXAP	127.3	100.0%	100.0%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RHOH	131.3	100.0%	100.0%	No OMIM disease ID
RIPK1	106.9	99.9%	99.1%	Immunodeficiency 57, 618108
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	143.0	100.0%	100.0%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	98.0	99.8%	96.3%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	314.2	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
RNF168	187.1	100.0%	99.4%	RIDDLE syndrome, 611943
RNF31	160.5	100.0%	99.9%	No OMIM Disease ID
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
RORC	132.1	100.0%	100.0%	Immunodeficiency 42, 616622
RPSA	68.7	100.0%	99.6%	Asplenia, isolated congenital, 271400
RSPH9	143.1	99.9%	98.0%	Ciliary dyskinesia, primary, 12, 612650
RTEL1	145.6	99.8%	98.2%	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
SAMD9	161.7	100.0%	100.0%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	170.6	100.0%	100.0%	Ataxia-pancytopenia syndrome, 159550
SAMHD1	135.4	100.0%	98.7%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SBDS	167.5	100.0%	100.0%	Shwachman-Diamond syndrome, 260400
SEMA3E	131.0	100.0%	99.7%	?CHARGE syndrome, 214800
SERAC1	110.4	100.0%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739

SERPING1	101.0	99.6%	97.5%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SH2B3	122.5	99.8%	98.8%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SH2D1A	108.5	95.7%	90.7%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	154.6	92.4%	91.4%	Cherubism, 118400
SKIV2L	150.1	100.0%	99.9%	Trichohepatoenteric syndrome 2, 614602
SLC29A3	190.1	100.0%	99.7%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	123.5	100.0%	99.8%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	209.1	100.0%	99.9%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	122.0	100.0%	99.7%	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC39A4	130.4	100.0%	99.7%	Acrodermatitis enteropathica, 201100
SLC46A1	121.7	100.0%	98.0%	Folate malabsorption, hereditary, 229050
SMARCAL1	119.6	100.0%	99.8%	Schimke immunoosseous dysplasia, 242900
SMARCD2	99.5	87.5%	86.1%	Specific granule deficiency 2, 617475
SNX10	124.3	96.2%	95.5%	Osteopetrosis, autosomal recessive 8, 615085
SOCS4	219.7	99.9%	99.3%	No OMIM Disease ID
SP110	112.3	100.0%	99.9%	Hepatic venoocclusive disease with immunodeficiency, 235550
SPINK5	128.1	100.0%	99.2%	Netherton syndrome, 256500
SPPL2A	56.3	84.3%	67.4%	No OMIM Disease ID
STAT1	116.6	99.2%	97.2%	Immunodeficiency 31C, autosomal dominant, 614162 Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796
STAT2	117.1	100.0%	99.9%	Immunodeficiency 44, 616636
STAT3	106.9	100.0%	99.4%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT4	140.0	100.0%	99.6%	No OMIM disease ID
STAT5B	119.7	99.9%	98.8%	Leukemia, acute promyelocytic, somatic, 102578 Growth hormone insensitivity with immunodeficiency, 245590
STAT6	121.5	100.0%	99.9%	No OMIM Disease ID
STIM1	129.2	99.8%	97.1%	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STK4	124.5	100.0%	99.7%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	330.7	100.0%	100.0%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552

STXBP2	110.2	84.1%	80.8%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TAP1	133.9	99.9%	97.7%	Bare lymphocyte syndrome, type I, 604571
TAP2	101.4	99.6%	98.7%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	130.6	96.6%	96.6%	Bare lymphocyte syndrome, type I, 604571
TAZ	125.3	99.3%	96.2%	Barth syndrome, 302060
TBX1	114.2	93.7%	88.3%	Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Conotruncal anomaly face syndrome, 217095
TCF3	100.2	99.4%	96.9%	Agammaglobulinemia 8, autosomal dominant, 616941
TCIRG1	149.6	99.6%	98.0%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	157.6	100.0%	100.0%	Transcobalamin II deficiency, 275350
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	160.1	99.9%	99.0%	No OMIM disease ID
TFRC	133.1	99.9%	99.0%	Immunodeficiency 46, 616740
THBD	208.2	100.0%	100.0%	Thrombophilia due to thrombomodulin defect, 614486
TICAM1	133.7	100.0%	100.0%	No OMIM disease ID
TINF2	190.9	100.0%	100.0%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TIRAP	151.5	100.0%	100.0%	No OMIM disease ID
TLR3	169.0	100.0%	99.3%	No OMIM disease ID
TLR4	133.7	100.0%	99.8%	No OMIM Disease ID
TMC6	102.1	100.0%	99.8%	Epidermodysplasia verruciformis, 226400
TMC8	148.5	100.0%	99.9%	Epidermodysplasia verruciformis 2, 618231
TMEM173	104.9	99.6%	94.7%	STING-associated vasculopathy, infantile-onset, 615934
TNFAIP3	158.7	100.0%	100.0%	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF11A	139.5	96.4%	95.6%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF13B	109.3	100.0%	99.8%	Immunoglobulin A deficiency 2, 609529 Immunodeficiency, common variable, 2, 240500
TNFRSF13C	115.9	96.9%	86.5%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	121.5	92.8%	91.4%	Periodic fever, familial, 142680
TNFRSF4	89.9	99.9%	98.2%	?Immunodeficiency 16, 615593
TNFSF11	133.0	100.0%	100.0%	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	101.5	100.0%	99.7%	No OMIM Disease ID
TPP2	113.5	99.3%	96.5%	No OMIM Disease ID
TRAC	132.2	100.0%	100.0%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387

TRAF3	117.1	99.9%	98.9%	No OMIM disease ID
TRAF3IP2	118.0	100.0%	98.4%	?Candidiasis, familial, 8, 615527
TREX1	261.9	100.0%	100.0%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRNT1	100.7	99.2%	95.3%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TTC37	131.5	99.9%	98.9%	Trichohepatoenteric syndrome 1, 222470
TTC7A	123.2	99.9%	98.9%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	142.1	100.0%	99.7%	Immunodeficiency 35, 611521
UNC13D	120.2	99.9%	99.2%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	68.6	61.7%	60.4%	No OMIM disease ID
UNG	127.2	98.9%	95.1%	Immunodeficiency with hyper IgM, type 5, 608106
USB1	122.0	99.8%	98.2%	Poikiloderma with neutropenia, 604173
USP18	151.7	95.9%	95.9%	Pseudo-TORCH syndrome 2, 617397
VAV1	111.8	98.5%	97.0%	No OMIM Disease ID
VPS13B	135.9	99.4%	97.8%	Cohen syndrome, 216550
VPS45	127.2	97.3%	94.1%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	75.4	95.3%	84.4%	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
WDR1	106.8	99.9%	98.9%	No OMIM Disease ID
WIPF1	95.7	100.0%	99.3%	?Wiskott-Aldrich syndrome 2, 614493
WRAP53	178.7	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	94.7	93.1%	88.3%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	206.3	100.0%	99.9%	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB24	160.7	100.0%	100.0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions :December 11th , 2019.

This list is accurate for panel version DG 2.17

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