PRIMARY IMMUNODEFICIENCIES GENE PANEL DG 2.5/2.6

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ACP5	187.9	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	135.8	100%	89%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADA	104.1	99%	97%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADAM17	134.1	96%	93%	?Inflammatory skin and bowel disease,neonatal,1,614328
ADAR	122.2	100%	99%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	146	100%	100%	Aspartylglucosaminuria, 208400
AICDA	137.8	99%	97%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	79	99%	96%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	98.7	99%	91%	Reticular dysgenesis, 267500
ALG13	125	100%	99%	Congenital disorder of glycosylation, type Is, 300884
AP3B1	103.7	95%	89%	Hermansky-Pudlak syndrome 2, 608233
APOL1	181.9	100%	100%	{End-stage renal disease,nondiabetic,susceptibility to},612551 {Glomerulosclerosis,focal segmental 4,susceptibility to},612551
ATM	118.9	99%	94%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480
BLM	130.3	98%	94%	Bloom syndrome, 210900
BLNK	118.7	94%	91%	Agammaglobulinemia 4, 613502
BLOC1S6	98	99%	98%	Hermansky-pudlak syndrome 9, 614171
BTK	183.8	100%	100%	Agammaglobulinemia, X-linked 1, 300755
C1QA	121	100%	92%	C1q deficiency, 613652
C1QB	169.6	100%	100%	C1q deficiency, 613652
C1QC	205.8	100%	100%	C1q deficiency, 613652

C1R	147.8	100%	100%	C1r/C1s deficiency, combined,216950
C1S	133.9	100%	99%	C1s deficiency, 613783
C2	17.9	76%	32%	C2 deficiency, 217000
C3	161.5	100%	99%	C3 deficiency, 613779
C4A	14.1	58%	20%	C4a deficiency, 614380
C4B	12.7	54%	16%	C4B deficiency, 614379
C5	136.4	97%	95%	C5 deficiency, 609536
C6	169.3	100%	99%	C6 deficiency, 612446
C7	147.6	94%	90%	C7 deficiency, 610102
C8A	118.3	100%	100%	C8 deficiency, type I, 613790
C8B	132.4	100%	99%	C8 deficiency, type II, 613789
C9	144.3	100%	99%	C9 deficiency, 613825
CARD11	164.2	98%	97%	Persistent polyclonal B-cell lymphocytosis, 606445
CARD9	118.1	96%	95%	Candidiasis, familial, 2, autosomal recessive, 212050
CASP10	108.7	99%	97%	Autoimmune lymphoproliferative syndrome, type II, 603909
CASP8	162.9	100%	99%	Immunodeficiency due to CASP8 deficiency, 607271
CD19	90.7	100%	96%	Immunodeficiency, common variable, 3, 613493
CD247	116.9	100%	100%	Immunodeficiency due to defect in CD3-zeta, 610163
CD27	124.5	100%	100%	Lymphoproliferative syndrome 2, 615122
CD3D	198.6	100%	100%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3E	167.5	100%	99%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3G	177.4	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	186.7	100%	100%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	172.1	99%	95%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	132.1	98%	89%	{Hemolytic uremic syndrome, atypical,susceptibility to},612922
CD55	143.8	86%	79%	[Blood group Cromer],613793
CD59	222.5	86%	79%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	144.3	100%	96%	Agammaglobulinemia 3, 613501
CD79B	230.5	100%	100%	Agammaglobulinemia 6, 612692
CD81	155.2	100%	100%	Immunodeficiency, common variable, 6, 613496
CD8A	92.4	100%	100%	CD8 deficiency, familial, 608957
CDCA7	109.8	99%	97%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDKN2B	66.1	100%	100%	No OMIM disease
CEBPE	70.3	100%	99%	Specific granule deficiency, 245480

CECR1	105.5	100%	97%	?Sneddon syndrome,182410 Polyarteritis nodosa,childhood-onset,615688
CFB	21.1	83%	47%	?Complement factor B deficiency,615561 {Hemolytic uremic syndrome,atypical,susceptibility to,4},612924 {Macular degeneration,age-related,14,reduced risk of},615489
CFD	65.7	87%	82%	Complement factor D deficiency, 613912
CFH	186.8	97%	95%	{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CFHR1	226.2	99%	99%	{Hemolytic uremic syndrome,atypical,susceptibility to},235400 {Macular degeneration,age-related,reduced risk of},603075
CFHR3	142.8	97%	92%	{Hemolytic uremic syndrome,atypical,susceptibility to},235400 {Macular degeneration,age-related,reduced risk of},603075
CFHR5	104.5	99%	91%	Nephropathy due to CFHR5 deficiency, 614809
CFI	160.9	97%	96%	Complement factor I deficiency, 610984
CFP	126.3	100%	97%	Properdin deficiency,X-linked, 312060
CHD7	144.8	100%	98%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CIITA	121.5	100%	100%	Bare lymphocyte syndrome type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CLEC4D	151.1	100%	100%	No OMIM disease
CLEC7A	161.4	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108
COLEC11	225.5	100%	100%	3MC syndrome 2, 265050
COPA	141.8	100%	99%	{Autoimmune interstitial lung,joint and kidney disease},616414
CORO1A	171.8	99%	94%	Immunodeficiency 8, 615401
CR2	169.5	99%	99%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927
CREBBP	140	98%	95%	Rubinstein-Taybi syndrome, 180849
CSF2RA	52	90%	85%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF3R	92.1	96%	94%	Neutrophilia, hereditary, 162830
CTC1	106.8	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	204.9	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

CTSC	131.9	100%	100%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CXCR4	203.6	100%	100%	WHIM syndrome, 193670
CYBA	85	82%	74%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	165.6	100%	98%	Chronic granulomatous disease, X-linked, 306400
DCLRE1C	119.7	99%	90%	Severe combined immunodeficiency, Athabascan type, 602450
DDX58	137.7	98%	95%	Singeleton-Merten syndrome 2,616298
DHFR	48	95%	70%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	160.6	100%	100%	Dyskeratosis congenita, X-linked, 305000
DNMT3B	133.2	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	149.5	100%	99%	Immunodeficiency 40,616433
DOCK8	140.3	100%	99%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	96.6	100%	95%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	100.1	100%	100%	No OMIM disease
EPG5	132.8	99%	97%	Vici syndrome, 242840
ERCC2	139.5	100%	100%	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	106.8	99%	98%	Trichothiodystrophy, 601675 Xeroderma pigmentosum, group B, 610651
F12	102.9	99%	96%	Factor XII deficiency, 234000
FADD	140.3	100%	97%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759
FAS	283.2	100%	100%	{Autoimmune lymphoproliferative syndrome}, 601859
FASLG	96.3	100%	99%	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FCGR1A	60.8	47%	41%	[IgG receptor I,phagocytic,familial deficiency of]
FCGR3A	201.3	100%	98%	Immunodeficiency 20, 615707
FCN3	134.9	100%	100%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	126.1	100%	99%	Leukocyte adhesion deficiency, type III, 612840
FOXN1	103.6	100%	99%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	170	100%	98%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790

FPR1	205.5	100%	100%	No OMIM disease
G6PC	202.4	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	132	100%	100%	Dursun syndrome,612541 Neutropenia,severe congenital 4,autosomal recessive,612541
G6PD	180.9	100%	100%	Favism, 134700 Hemolytic anemia due to G6PD deficiency,300908 Resistance to malaria due to G6PD deficiency, 611162
GATA2	137.7	100%	99%	Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency, 614172
GFI1	89.6	96%	89%	Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GJC2	49.1	88%	59%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GTF2H5	180.5	100%	99%	Trichothiodystrophy, complementation group A, 601675
HAX1	125.5	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HELLS	101.8	95%	88%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4,616911
ICOS	154.4	100%	100%	Immunodeficiency, common variable, 1, 607594
IFIH1	116.7	99%	97%	Aicardi-Goutieres syndrome 7,615846 Singleton-Merten syndrome 1,182250
IFNGR1	139.1	99%	94%	Mycobacterial infection, atypical, familial disseminated, 209950
IFNGR2	160.5	93%	93%	Immunodeficiency 28,mycobacteriosis,614889
IGLL1	54.3	99%	88%	Agammaglobulinemia 2, 613500
IKBKB	126.7	97%	94%	Immunodeficiency 15,615592
IKBKG	61	91%	80%	Ectodermal dysplasia,hypohidrotic,with immune deficiency,300291 Ectodermal,dysplasia,anhidrotic,lymphedema and immunodeficiency,300301 Immunodeficiency 33,300636 Immunodeficiency,isolated,300584 Uncontinentia pigmenti,308300 Invasive pneumococcal disease,recurrent isolated,2,300640
IKZF1	172.4	100%	100%	Leukemia,acute lymphoblastic Systemic lupus erythematosus, association with (Han (2009) Nat Genet 41,1234)
IL10RA	144	100%	99%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	215	97%	95%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424

IL12B	121.4	100%	100%	Immunodeficiency 29,mycobacteriosis,614890
IL12RB1	129	96%	94%	Immunodeficiency 30,614891
IL17F	83.2	95%	88%	Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	124.7	100%	94%	Candidiasis, familial, 5, autosomal recessive, 613953
IL1RN	188.6	100%	100%	{Gastric cancer risk after H. pylori infection}, 137215
IL2	77.4	87%	86%	Severe combined immunodeficiency due to IL2 deficiency
IL21R	135	100%	100%	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 [IgE, elevated level of], 147050
IL2RA	131.1	100%	100%	Interleukin-2 receptor, alpha chain, deficiency of, 606367
IL2RG	93.4	100%	100%	Severe combined immunodeficiency, X-linked, 300400
IL36RN	93.1	100%	100%	Psoriasis, generalized pustular, 614204
IL7R	147.1	100%	99%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INSR	129.9	96%	93%	Leprechaunism, 246200
IRAK4	94.7	87%	84%	IRAK4 deficiency, 607676
IRF7	88.7	100%	99%	?Immunodeficiency 39,616345
IRF8	128.1	100%	96%	Monocyte and dendritic cell deficiency, recessive, 614894
ISG15	172.1	100%	100%	Immunodeficiency 38,616126
ITCH	127.2	95%	95%	Autoimmune disease, syndromic multisystem, 613385
ITGB2	163.2	100%	99%	Leukocyte adhesion deficiency, 116920
ITK	131.5	100%	99%	Lymphoproliferative syndrome 1, 613011
JAK2	96	95%	93%	Erythrocytosis,somatic,133100 Leukemia,acute myelogenous,601626 Myelofibrosis,somatic,254450 Polycythemia vera,263300 Thrombocythemia 3,614521 {Budd-Chiari syndrome},600880
JAK3	108.1	98%	96%	SCID, autosomal recessive, T-negative/B-positive type, 600802
KMT2D	147.5	100%	99%	Kabuki syndrome 1, 147920

KRAS	77.3	100%	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-Mins syndrome,somatic mosaic,163200
LAMTOR2	154.7	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LCK	164.6	95%	93%	?Immunodeficiency 22,615758
LIG1	94.3	100%	98%	DNA ligase I deficiency
LIG4	176.1	100%	100%	LIG4 syndrome, 606593 Severe combined immunodeficiency with sensitivity to ionizing radiation, 602450 {Multiple myeloma, resistance to}, 254500
LPIN2	127.5	100%	100%	Majeed syndrome, 609628
LRBA	136.6	98%	97%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	287.7	100%	100%	Agammaglobulinemia 5, 613506
LTBP3	117.3	98%	97%	Dental anomalies and short stature,601216
LYST	136	98%	93%	Chediak-Higashi syndrome,214500
MAGT1	148.9	98%	98%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 Mental retardation, X-linked 95, 300716
MAL	144	100%	100%	No OMIM disease
MAN2B1	123.1	98%	94%	Mannosidosis, alpha-, types I and II, 248500
MANBA	129.3	100%	98%	Mannosidosis, beta, 248510
MASP2	148	99%	99%	MASP2 deficiency, 613791
MBL2	118.3	99%	89%	{Chronic infections,due to MBL deficiency},614372
MC2R	239.8	100%	100%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM4	174.7	100%	96%	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MEFV	119.6	94%	90%	Familial Mediterranean fever, AR, 249100
MKL1	113	98%	95%	Megakaryoblastic leukemia,acute

MPO	165.3	100%	100%	Cardiomyopathy, dilated, 1T, 613740 Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}
MRE11A	52.7	93%	81%	Ataxia-telangiectasia-like disorder, 604391
MS4A1	122.7	98%	89%	Immunodeficiency, common variable, 5, 613495
MTHFD1	148.2	99%	96%	{Abruptio placentae,susceptibility to} {Spina bifida,folate-sensitive,susceptibility to},601634
MVK	126.1	100%	100%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, disseminated superficial actinic, 175900
MYD88	212.3	100%	98%	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
NBN	84.7	100%	97%	Aplastic anemia,609135 Leukemia,acute lymphoblastic,613065 Nijmegen breakage syndrome,251260
NCF1	22.9	25%	25%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	133.3	100%	98%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	158.8	100%	100%	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	116.1	100%	99%	Acne inversa, familial, 1, 142690
NDNL2	115.2	100%	91%	No OMIM disease
NFKB1	117.2	98%	93%	Immunodeficiency,common variable, 12,616576
NFKB2	123.3	97%	94%	Immunodeficiency, common variable, 10, 615577
NFKBIA	108.7	97%	94%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NHEJ1	76.6	100%	97%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	69.5	100%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987
NKX2-5	71.4	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	173.2	100%	100%	?Familial cold autoinflammatory syndrome 4,616115 Autoinflammation with infantile enterocolitis,616050

NLRP1	128.9	98%	94%	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia,615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1},606579
NLRP12	151.5	99%	99%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	134.9	100%	100%	CINCA syndrome, 607115 Cold-induced autoinflammatory syndrome, familial, 120100 Muckle-Wells syndrome, 191900
NOD2	136.3	100%	99%	{Inflammatory bowel disease 1}, 266600
NOP10	183.4	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	195	100%	100%	?RAS-associated autoimmune lymphoproliferative syndrome type IV,somatic,611470 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
ORAI1	219.7	92%	90%	Immunodeficiency 9,612782 Myopathy,tubular aggregate, 2,615883
PARN	128.2	100%	98%	Dyskeratosis congenita,autosomal recessive 6,616353 Pulmonary fibrosis and/or bone marrow failure,telomere-related,4,616371
PBX1	113.4	99%	96%	Leukemia,acute pre-B-cell,176310
PCCA	112.5	94%	92%	Propionicacidemia, 606054
PCCB	142.5	95%	94%	Propionicacidemia, 606054
PEPD	114	98%	98%	Prolidase deficiency, 170100
PGM3	202.2	100%	100%	Immunodeficiency 23,615816
PIGA	120.2	96%	92%	Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868 Paroxysmal nocturnal hemoglobinuria,somatic,300818
PIK3CD	137.4	99%	97%	Immunodeficiency 14, 615513
PIK3R1	131.8	99%	96%	Agammaglobulinemia 7, autosomal recessive, 615214
PLCG2	123.4	100%	99%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLG	118.8	87%	87%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PMM2	163.7	100%	100%	Congenital disorder of glycosylation, type Ia, 212065

PNP	144.4	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PRF1	120.6	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKDC	107.8	97%	92%	Immunodeficiency 26,with or without neurologic abnormalities,615966
PRPS1	254.1	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	91.7	100%	100%	Acne inversa, familial, 2, 613736
PSMB8	15.7	54%	24%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSTPIP1	98.3	99%	96%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN11	92.6	94%	86%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPRC	108.6	93%	87%	{Hepatitic C virus, susceptibility to}, 609532 Severe combined immunodeficiency,T cell-negative,B-cell/natural killer-cell positive,608971
PTRF	133.8	100%	98%	Lipodystrophy, congenital generalized, type 4, 613327
RAB27A	169.8	100%	100%	Griscelli syndrome, type 2, 607624
RAC2	130.8	100%	100%	Neutrophil immunodeficiency syndrome, 608203
RAG1	201.2	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457
RAG2	221.6	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457
RASGRP2	100.1	100%	98%	?Bleeding disorder,platelet-type,18,615888
RBCK1	102.6	96%	91%	Polyglucosan body myopathy 1 with or without immunodeficiency,615895
RECQL4	131.6	98%	96%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RFX5	109.4	99%	97%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	110.9	100%	100%	MHC class II deficiency, complementation group B, 209920
RFXAP	124.6	94%	92%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RHOH	135.3	100%	100%	No OMIM disease

RMRP				Anauxetic dysplasia,607095 Cartilage-hair hypoplasia,250250 Metaphyseal dysplasia without hypotrichosis,250460
RNASEH2A	138.8	100%	100%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	115.7	93%	81%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	179.3	100%	99%	Aicardi-Goutieres syndrome 3, 610329
RNF168	236.8	100%	96%	RIDDLE syndrome, 611943
RNF31	153.4	98%	96%	No OMIM disease
RPSA	79.1	100%	100%	Asplenia, isolated congenital, 271400
RSPH9	135.5	98%	96%	Ciliary dyskinesia, primary, 12, 612650
RTEL1	118.1	99%	97%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
SAMHD1	141.3	99%	95%	Aicardi-Goutieres syndrome 5, 612952
SBDS	195.9	99%	96%	Shwachman-Bodian-Diamond syndrome, 260400
SERAC1	115	100%	99%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	88.7	96%	92%	Angioedema, hereditary, types I and II, 106100
SH2D1A	184.3	89%	89%	Lymphoproliferative syndrome, X-linked, 308240
SKIV2L	24.8	79%	55%	Trichohepatoenteric syndrome 2, 614602
SLC29A3	224	100%	99%	Histiocytosis-lymphadenopathy plus syndrome,602782
SLC35A1	130.2	100%	97%	Congenital disorder of glycosylation, type 2f, 603585
SLC35C1	238.9	99%	97%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	135.6	100%	100%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	87.6	98%	94%	Acrodermatitis enteropathica, 201100
SLC46A1	89	97%	95%	Folate malabsorption, hereditary, 229050
SMARCAL1	134.8	99%	98%	Schimke immunoosseous dysplasia, 242900
SOCS4	293.2	100%	98%	No OMIM disease
SP110	133.6	100%	99%	Hepatic venoocclusive disease with immunodeficiency, 235550
SPINK5	160.1	99%	96%	Netherton syndrome, 256500
STAT1	133.2	96%	95%	Mycobacterial infection, atypical, familial disseminated, 209950
STAT2	118.7	100%	100%	Immunodeficiency 44,616636
STAT3	126.7	100%	96%	Hyper-IgE recurrent infection syndrome, 147060
STAT4	166.3	98%	96%	{Systemic lupus erythematosus,susceptibility to,11},612253

STAT5B	130	98%	92%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, STAT5B/RARA type
STIM1	126.2	99%	95%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STK4	153.5	100%	99%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	274.8	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	126.6	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TAP1	11.1	44%	9%	Bare lymphocyte syndrome, type I, 604571
TAP2	11.4	43%	14%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	20.8	80%	49%	Bare lymphocyte syndrome, type I, 604571
TAZ	158.3	100%	100%	Barth syndrome, 302060
TBX1	82.7	75%	64%	Conotruncal anomaly face syndrome, 217095
TCIRG1	126.6	96%	87%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	173.3	100%	100%	Transcobalamin II deficiency, 275350
TERC				Dyskeratosis congenita,autosomal dominant 1,127550 {Aplastic anemia},614743
TERT	141.2	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	171	100%	100%	Immunodeficiency 46,616740
THBD	119.1	100%	98%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TICAM1	109	100%	100%	{Herpes-simplex encephalitic,susceptibility to,6},614850
TINF2	178.8	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TLR3	172.3	99%	97%	{Herpes simplex encephalitis, susceptibility to,2},613002 {HIV1 infection,resistance to},609423
TLR4	126.3	99%	97%	Endotoxin hyporesponsiveness {Colorectal cancer,susceptibility to},114500 {Macular degeneration,age-related,10},611488
TMC6	81.1	99%	95%	Epidermodysplasia verruciformis, 226400

TMC8	106	95%	85%	Epidermodysplasia verruciformis, 226400
TMEM173	97.5	99%	91%	STING-associated vasculopathy,infantile-onset,615934
TNFRSF11A	126	93%	89%	Osteolysis, familial expansile, 174810 Paget disease of bone, 602080 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF13B	108.1	100%	99%	Immunoglobulin A deficiency 2, 609529
TNFRSF13C	58.7	93%	75%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	104.8	90%	87%	Periodic fever, familial, 142680
TNFRSF4	70.4	99%	93%	?Immunodeficiency 16,615593
TPP2	136.1	96%	92%	No OMIM disease
TRAF3	134.4	99%	97%	[?Herpes simplex encephalitis,susceptibility to,3],614849
TRAF3IP2	115.4	99%	97%	?Candidiasis,familial,8,615527 {Psoriasis susceptibility 13},614070
TREX1	250.5	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	104.4	94%	86%	Retinitis pigmentosa and erythrocytic microcytosis,616959 Sideroblastic anemia with B-cell immunodeficiency,periodic fevers and developmental delay,616084
TTC37	129.7	100%	96%	Trichohepatoenteric syndrome 1, 222470
TTC7A	114.1	99%	97%	Intestinal atresia, multiple, 243150
TYK2	117.4	100%	98%	Tyrosine kinase 2 deficiency, 611521
UNC119	110.7	98%	92%	?Cone-rod dystrophy ?Immunodeficiency 13,615518
UNC13D	100.8	98%	96%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	65.3	58%	55%	Herpes simplex encephalitis, susceptibility to, 1, 610551
UNG	69.2	97%	91%	Immunodeficiency with hyper IgM, type 5, 608106
USB1	121.6	97%	93%	Poikiloderma with neutropenia, 604173
VPS13B	146.1	98%	96%	Cohen syndrome, 216550
VPS45	137.2	94%	93%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	84.6	92%	83%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000

WIPF1	77.5	99%	98%	Wiskott-Aldrich syndrome 2, 614493
WRAP53	146.4	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	136.5	94%	88%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	203.2	100%	98%	Selective T-cell defect, 269840
ZBTB24	171.7	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 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

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