PRIMARY IMMUNODEFICIENCIES GENE PANEL DGD09072015

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM ID*
ACP5	95.6	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	61.8	100%	95%	Dystonia, juvenile-onset, 607371
ADA	72.3	100%	96%	Severe combined immunodeficiency due to ADA deficiency, 102700
ADAM17	111.6	99%	97%	?Inflammatory skin and bowel disease,neonatal,1,614328
ADAR	128.1	99%	98%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	111.6	100%	91%	Aspartylglucosaminuria, 208400
AICDA	80.4	100%	96%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	73.4	99%	89%	Autoimmune polyendocrinopathy syndrome , type I, with reversible metaphyseal dysplasia, 240300
AK2	72	80%	77%	Reticular dysgenesis, 267500
ALG13	111.7	96%	95%	Congenital disorder of glycosylation, type Is, 300884
AP3B1	104.8	100%	99%	Hermansky-Pudlak syndrome 2, 608233
APOL1	137.1	100%	100%	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
ATM	111.1	99%	99%	Ataxia-telangiectasia, 208900
BLM	114.8	100%	98%	Bloom syndrome, 210900
BLNK	98.4	97%	97%	Agammaglobulinemia 4, 613502
BLOC1S6	118.7	88%	81%	Hermansky-pudlak syndrome 9, 614171
BTK	98.6	100%	99%	Agammaglobulinemia, X-linked 1, 300755
C1QA	119.1	98%	91%	C1q deficiency, 613652
C1QB	90.3	94%	87%	C1q deficiency, 613652
C1QC	121.9	84%	69%	C1q deficiency, 613652
C1R	91.7	96%	93%	C1r/C1s deficiency, combined, 216950
C1S	95.5	99%	99%	C1s deficiency, 613783
C2	17	76%	32%	C2 deficiency, 217000
C3	99.1	97%	93%	C3 deficiency, 613779
C4A	2.6	5%	3%	C4a deficiency, 614380
C4B	1.7	4%	2%	C4B deficiency, 614379
C5	97	100%	99%	C5 deficiency, 609536

C6	111.6	100%	99%	C6 deficiency, 612446
C7	90.4	99%	95%	C7 deficiency, 610102
C8A	77.2	100%	98%	C8 deficiency, type I, 613790
C8B	92.3	100%	96%	C8 deficiency, type II, 613789
C9	109.2	100%	100%	C9 deficiency, 613825
CARD11	93	100%	98%	Persistent polyclonal B-cell lymphocytosis, 606445
CARD9	64.3	99%	98%	Candidiasis, familial, 2, autosomal recessive, 212050
CASP10	97.9	100%	100%	Autoimmune lymphoproliferative syndrome, type II, 603909
CASP8	118.5	100%	97%	Immunodeficiency due to CASP8 deficiency, 607271
CD19	71.7	100%	98%	Immunodeficiency, common variable, 3, 613493
CD247	86.5	100%	100%	Immunodeficiency due to defect in CD3-zeta, 610163
CD27	74.6	100%	98%	Lymphoproliferative syndrome 2, 615122
CD3D	90.9	100%	95%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3E	93	99%	84%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3G	96.3	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	105	95%	92%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	117.3	99%	96%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	110.2	100%	100%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD55	86.5	74%	71%	[Blood group Cromer], 613793
CD59	95.4	85%	79%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	79.1	95%	87%	Agammaglobulinemia 3, 613501
CD79B	121.9	100%	100%	Agammaglobulinemia 6, 612692
CD81	81.4	100%	93%	Immunodeficiency, common variable, 6, 613496
CD8A	86.6	100%	99%	CD8 deficiency, familial, 608957
CDKN2B	114.6	100%	100%	No OMIM phenotype
CEBPE	102.2	100%	100%	Specific granule deficiency, 245480
CECR1	90.2	99%	96%	?Sneddon syndrome,182410 Polyarteritis nodosa,childhood-onset,615688
CFB	18	70%	37%	{Macular degeneration, age-related, reduced risk of}, 603075
CFD	48.6	96%	75%	Complement factor D deficiency, 613912
CFH	105.6	95%	92%	{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CFHR1	25.5	68%	51%	{Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	100.7	88%	78%	{Macular degeneration, age-related, reduced risk of}, 603075
CFHR5	96	95%	87%	Nephropathy due to CFHR5 deficiency, 614809

CFI	130	100%	100%	Complement factor I deficiency, 610984
CFP	94.8	98%	93%	Properdin deficiency,X-linked, 312060
CHD7	117.1	100%	99%	CHARGE syndrome, 214800
CIITA	88.5	98%	94%	Bare lymphocyte syndrome type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CLEC4D	90.6	100%	99%	No OMIM phenotype
CLEC7A	111.8	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108
COLEC11	109	100%	100%	3MC syndrome 2, 265050
COPA	113.5	100%	98%	{Autoimmune interstitial lung,joint and kidney disease},616414
CORO1A	98.7	92%	91%	Immunodeficiency 8, 615401
CR2	109.8	100%	100%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927
CREBBP	78	99%	97%	Rubinstein-Taybi syndrome, 180849
CSF2RA	0	0%	0%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF3R	82.5	100%	98%	Neutrophilia, hereditary, 162830
CTC1	96.7	99%	96%	Cerebroretinal microangiopathy with calcifications and cysts,612199
CTLA4	116.4	100%	99%	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	95.5	100%	99%	Papillon-Lefevre syndrome, 245000
CXCR4	193.2	100%	100%	WHIM syndrome, 193670
CYBA	25.7	64%	48%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	94.4	92%	88%	Chronic granulomatous disease, X-linked, 306400
DCLRE1C	97.2	90%	90%	Severe combined immunodeficiency, Athabascan type, 602450
DDX58	112.7	100%	99%	Singleton-Merten syndrome 2,616298
DHFR	50.6	79%	63%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	91.1	100%	98%	Dyskeratosis congenita, X-linked, 305000
DNMT3B	91.4	100%	97%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	103	100%	99%	Immunodeficiency 40,616433
DOCK8	85.9	100%	98%	Mental retardation, autosomal dominant 2, 614113
ELANE	109.7	99%	92%	Neutropenia, cyclic, 162800
ELF4	106.8	100%	100%	No OMIM phenotype Hypogammaglobulinaemia (Stewart (2005) Curr Opin Allergy Clin Immunol 5, 510)

EPG5	87.2	100%	99%	Vici syndrome, 242840
ERCC2	87.2	99%	93%	Xeroderma pigmentosum, group D, 278730
ERCC3	121.5	100%	100%	Xeroderma pigmentosum, group B, 610651
F12	96.7	100%	99%	Factor XII deficiency, 234000
FADD	100.9	100%	98%	Infections, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759
FAS	200.3	100%	99%	{Autoimmune lymphoproliferative syndrome}, 601859
FASLG	87.1	99%	95%	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FCGR1A	0	0%	0%	[IgG receptor I, phagocytic, familial deficiency of]
FCGR3A	45.1	52%	46%	Immunodeficiency 20, 615707
FCN3	106.4	98%	96%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	101.5	100%	97%	Leukocyte adhesion deficiency, type III, 612840
FOXN1	113.5	100%	96%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	84.9	99%	94%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FPR1	160.8	100%	100%	No OMIM phenotype Periodontitis, aggressive, association with (Gunji (2007) Biochem Biophys Res Commun 364,7) Periodontitis, juvenile, association with (Gwinn (1999) J Periodontol 70,1194)
G6PC	134.5	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	115.8	100%	99%	Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	104.9	95%	95%	Favism, 134700 Hemolytic anemia due to G6PD deficiency, 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GATA2	105	96%	91%	Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency, 614172
GFI1	71.2	100%	98%	Neutropenia, severe congenital 2, autosomal dominant, 613107
GJC2	52.1	92%	82%	Leukodystrophy, hypomyelinating, 2, 608804
GTF2H5	89.9	100%	100%	Trichothiodystrophy, complementation group A, 601675
HAX1	128.2	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
ICOS	125.5	100%	100%	Immunodeficiency, common variable, 1, 607594
IFIH1	127.2	100%	99%	Aicardi-Goutieres syndrome 7,615846 Singleton-Merten syndrome 1,182250
IFNGR1	140.9	100%	100%	Mycobacterial infection, atypical, familial disseminated, 209950
IFNGR2	108.9	93%	92%	{Mycobacterial infection, atypical, familial disseminated},209950
IGLL1	21.9	67%	41%	Agammaglobulinemia 2, 613500
IKBKB	93.7	97%	94%	Immunodeficiency 15,615592

IKBKG	25.9	26%	26%	Incontinentia pigmenti, type II, 308300
IKZF1	108.7	100%	99%	Leukemia,acute lymphoblastic Systemic lupus erythematosus, association with (Han (2009) Nat Genet 41,1234)
IL10RA	97.5	100%	98%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	111.8	100%	94%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL12B	85.1	100%	100%	BCG and salmonella infection, disseminated, 209950
IL12RB1	61.2	100%	89%	{Mycobacterial and salmonella infections, susceptibility to}, 209950
IL17F	91.9	100%	95%	Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	87.2	98%	89%	Candidiasis, familial, 5, autosomal recessive, 613953
IL1RN	110.9	100%	100%	{Gastric cancer risk after H. pylori infection}, 137215
IL2	93.9	100%	100%	Severe combined immunodeficiency due to IL2 deficiency Allergic disorders, association with (Christinsen (2006) Eur J Hum Genet 14,227)
IL21R	107.1	100%	98%	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 [IgE, elevated level of], 147050
IL2RA	98.1	100%	100%	Interleukin-2 receptor, alpha chain, deficiency of, 606367
IL2RG	93.2	100%	95%	Severe combined immunodeficiency, X-linked, 300400
IL36RN	89	100%	100%	Psoriasis, generalized pustular, 614204
IL7R	92.7	100%	95%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INSR	120.5	98%	95%	Leprechaunism, 246200
IRAK4	109.6	100%	100%	IRAK4 deficiency, 607676
IRF7	89.3	100%	99%	?Immunodeficiency 39,616345
IRF8	65.9	100%	98%	Monocyte and dendritic cell deficiency, recessive, 614894
ISG15	95.1	100%	100%	Immunodeficiency 38,616126
ITCH	103.5	95%	95%	Autoimmune disease, syndromic multisystem, 613385
ITGB2	82.6	99%	96%	Leukocyte adhesion deficiency, 116920
ITK	100.4	100%	100%	Lymphoproliferative syndrome 1, 613011
JAK2	109.7	100%	99%	Erythrocytosis,somatic,133100 Leukemia,acute myelogenous,601626 Myelofibrosis,somatic,254450 Polycythemia vera,263300 Thrombocythemia 3,614521 {Budd-Chiari syndrome},600880
JAK3	85.5	98%	94%	SCID, autosomal recessive, T-negative/B-positive type, 600802

KMT2D	102	99%	98%	Kabuki syndrome 1,147920
KRAS	65.1	95%	89%	Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myelogenous Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic,
LAMTOR2	73.4	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LCK	91.9	92%	82%	?Immunodeficiency 22, 615758
LIG1	74.5	98%	91%	DNA ligase I deficiency
LIG4	177.2	100%	100%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LPIN2	75.8	99%	95%	Majeed syndrome, 609628
LRBA	102.9	100%	97%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	129.6	100%	100%	Agammaglobulinemia 5, 613506
LYST	116.3	99%	97%	Chediak-Higashi syndrome, 214500
MAGT1	96.3	98%	98%	Mental retardation, X-linked 95, 300716
MAL	97	94%	88%	Meleda disease,248300
MAN2B1	81.3	99%	92%	Mannosidosis, alpha-, types I and II, 248500
MANBA	87.6	100%	99%	Mannosidosis, beta, 248510
MASP2	114.6	99%	96%	MASP2 deficiency, 613791
MBL2	118.3	100%	98%	{Chronic infections, due to MBL deficiency}, 614372
MC2R	126.8	100%	99%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM4	98.2	100%	97%	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MEFV	113.7	96%	95%	Familial Mediterranean fever, AR, 249100
MKL1	69.1	97%	91%	Megakaryoblastic leukemia,acute
MPO	86.2	100%	98%	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers} Cardiomyopathy, dilated, 1T, 613740
MRE11A	90.5	99%	99%	Ataxia-telangiectasia-like disorder, 604391
MS4A1	130.7	100%	100%	Immunodeficiency, common variable, 5, 613495

MTHFD1	90.6	100%	96%	{Abrubtio placentae, susceptibility to} {Spina bifida, folate-sensitive, susceptibility to}, 601634
MVK	86.1	100%	99%	Mevalonic aciduria, 610377
MYD88	165.3	100%	99%	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
NBN	115.9	98%	97%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCF1	0.5	0%	0%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	92.5	100%	98%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	90.1	98%	97%	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	79.2	97%	92%	Acne inversa, familial, 1, 142690
NFKB1	73	100%	98%	Immunodeficiency,common variable,12,616576
NFKB2	89.2	100%	97%	Immunodeficiency, common variable, 10, 615577
NFKBIA	95.9	100%	99%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NHEJ1	88	100%	94%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	47	100%	93%	Dyskeratosis congenita, autosomal recessive 2, 613987
NKX2-5	125.3	100%	99%	Atrial septal defect 7, with or without AV conduction defects, 108900
NLRP1	105.9	99%	98%	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia,615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1},606579
NLRP12	103.6	99%	98%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	116.6	100%	99%	Cold-induced autoinflammatory syndrome, familial, 120100
NOD2	91.7	100%	98%	{Inflammatory bowel disease 1}, 266600
NOP10	169.5	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	135	100%	100%	Autoimmune lymphoproliferative syndrome type IV, 614470
ORAI1	77.2	92%	88%	Immunodeficiency 9, 612782
PARN	105.1	100%	100%	Dyskeratosiscongenita,autosomal recessive 6,616353
PBX1	71.9	99%	86%	Leukemia,acute pre-B cell,176310
PCCA	95.4	98%	95%	Propionicacidemia, 606054
PCCB	101.6	99%	97%	pccB complementation group Propionicacidemia, 606054
PEPD	63.9	100%	90%	Prolidase deficiency, 170100
PGM3	102.7	96%	88%	Immunodeficiency 23, 615816
PIGA	134.6	100%	99%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIK3CD	85.5	98%	92%	Immunodeficiency 14, 615513

PIK3R1	140.4	100%	100%	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PLCG2	105.4	100%	99%	Familial cold autoinflammatory syndrome 3, 614468
PLG	64.4	75%	68%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PMM2	85.6	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNP	117.2	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PRF1	84.5	100%	97%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553
PRKDC	86.3	99%	96%	Immunodeficiency 16 with or without neurologic abnormalities, 615966
PRPS1	129.3	100%	100%	Gout, PRPS-related, 300661
PSENEN	117.2	100%	100%	Acne inversa, familial, 2, 613736
PSMB8	10	38%	7%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSTPIP1	57	96%	89%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN11	41.7	83%	68%	Noonan syndrome 1, 163950
PTPRC	103	97%	94%	{Hepatitic C virus, susceptibility to}, 609532 Severe combined immunodeficiency,T cell-negative,B-cell/natural killer-cell positive,608971
PTRF	146.8	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
RAB27A	119.7	100%	100%	Griscelli syndrome, type 2, 607624
RAC2	59.8	99%	96%	Neutrophil immunodeficiency syndrome, 608203
RAG1	132.2	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457
RAG2	187.4	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457
RASGRP2	80.9	100%	97%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	78.4	95%	90%	Polyglucosan body myopathy, early-onset, with or without immunodeficiency, 615895
RECQL4	91.5	98%	96%	Rothmund-Thomson syndrome, 268400
RFX5	106.5	99%	99%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	87.8	99%	96%	MHC class II deficiency, complementation group B, 209920
RFXAP	87.4	92%	86%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RHOH	146.4	100%	100%	No OMIM phenotype RHOH deficiency (Crequer (2012) J Clin Invest 122, 3239)
RNASEH2A	96.1	99%	94%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	102.3	99%	97%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	133.2	100%	100%	Aicardi-Goutieres syndrome 3, 610329

RNF168	193.1	100%	100%	RIDDLE syndrome, 611943
RPSA	22.4	77%	49%	Asplenia, isolated congenital, 271400
RTEL1	73.3	99%	92%	Dyskeratosis congenita, autosomal recessive 5, 615190
SAMHD1	112.6	100%	98%	Aicardi-Goutieres syndrome 5, 612952
SBDS	88.2	98%	93%	Shwachman-Bodian-Diamond syndrome, 260400
SERAC1	88.2	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	120.3	96%	89%	Angioedema, hereditary, types I and II, 106100
SH2D1A	87	99%	99%	Lymphoproliferative syndrome, X-linked, 308240
SKIV2L	17.3	68%	33%	Trichohepatoenteric syndrome 2, 614602
SLC29A3	147.5	100%	99%	Histiocytosis-lymphadenopathy plus syndrome,602782
SLC35A1	98.6	100%	98%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	97.4	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	81.5	100%	97%	Glycogen storage disease Ib, 232220
SLC39A4	69.9	100%	97%	Acrodermatitis enteropathica, 201100
SLC46A1	84	100%	98%	Folate malabsorption, hereditary, 229050
SMARCAL1	118.1	99%	97%	Schimke immunoosseous dysplasia, 242900
SOCS4	187.1	100%	100%	No OMIM phenotype
SP110	96.2	100%	100%	Hepatic venoocclusive disease with immunodeficiency, 235550
SPINK5	97.3	100%	99%	Netherton syndrome, 256500
STAT1	86.7	100%	98%	Mycobacterial infection, atypical, familial disseminated, 209950
STAT2	130.4	100%	100%	Immunodeficiency 44,616636
STAT3	85	100%	96%	Hyper-IgE recurrent infection syndrome, 147060
STAT4	106.1	100%	98%	{Systemic lupus erythematosus, susceptitbility to, 11}, 612253
STAT5B	72.5	83%	75%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, STAT5B/RARA type
STIM1	81.6	99%	96%	Immunodeficiency 10, 612783
STK4	99.2	100%	99%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	169.7	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	84.4	100%	95%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TAP1	14	60%	22%	Bare lymphocyte syndrome, type I, 604571
TAP2	10.9	39%	15%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	17.6	63%	30%	Bare lymphocyte syndrome, type I, 604571
TAZ	100	100%	100%	Barth syndrome, 302060
TBX1	66.3	72%	66%	Conotruncal anomaly face syndrome, 217095

TCIRG1	73.7	92%	84%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	104.1	100%	97%	linked to P1 Transcobalamin II deficiency, 275350
TERT	103.7	100%	99%	{Bone marrow failure, telomere-related, 1}, 614742 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Dyskeratosis congenita, autosomal dominant 2}, 613989 {Coronary artery disease} {Pulmonary fibrosis, telomere-related, 1}, 614742
THBD	74.2	100%	100%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TICAM1	80.1	100%	100%	{Encephalopathy, acute, infection-induced, susceptibility to, 6}, 614850
TINF2	176.7	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990
TLR3	160.4	100%	100%	{Herpes simplex encephalitis, susceptibility to, 2} 613002
TMC6	58.8	99%	94%	Epidermodysplasia verruciformis, 226400
TMC8	76.1	99%	94%	Epidermodysplasia verruciformis, 226400
TMEM173	57.9	100%	94%	STING-associated vasculopathy,infantile onset,615934
TNFRSF11A	100.8	95%	93%	Osteolysis, familial expansile, 174810
TNFRSF13B	57.8	99%	94%	Immunoglobulin A deficiency 2, 609529
TNFRSF13C	54.1	100%	81%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	67.7	93%	85%	Periodic fever, familial, 142680
TNFRSF4	58.7	98%	87%	?Immunodeficiency 16, 615593
TPP2	98.7	99%	93%	No OMIM phenotype
TRAF3	111.4	100%	97%	{Herpes simplex encephalitis, susceptibility to, 3}, 614849
TRAF3IP2	110.8	100%	99%	?Candidiasis,familial,8,615527 {Psoriasis susceptibility 13},614070
TREX1	134.4	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750
TRNT1	107.8	100%	100%	Sideroblastic anemia with B-cell immunodeficiency,periodic fevers,and developmental delay,616084
TTC37	111.4	100%	100%	Trichohepatoenteric syndrome 1, 222470
TTC7A	62.3	96%	92%	Intestinal atresia, multiple, 243150
TYK2	88.9	99%	95%	Tyrosine kinase 2 deficiency, 611521
UNC119	118.9	100%	100%	?Cone-rod dystrophy ?Immunodeficiency 13,615518
UNC13D	61.7	95%	91%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	40	54%	53%	s simplex encephalitis, susceptibility to, 1, 610551
UNG	63.6	91%	83%	Immunodeficiency with hyper IgM, type 5, 608106

USB1	52.1	91%	83%	Poikiloderma with neutropenia, 604173
VPS13B	104.4	99%	98%	Cohen syndrome, 216550
VPS45	100.1	94%	93%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	60.1	100%	90%	Wiskott-Aldrich syndrome, 301000
WIPF1	98.2	97%	94%	Wiskott-Aldrich syndrome 2, 614493
WRAP53	142.8	100%	98%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	130.3	90%	82%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	92.3	98%	93%	Selective T-cell defect, 269840
ZBTB24	148.3	100%	99%	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
OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015
This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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