PRIMARY IMMUNODEFICIENCIES GENE PANEL DG 2.3.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
ACP5	95,4	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	60,1	97%	90%	Dystonia, juvenile-onset, 607371
ADA	70,7	100%	95%	Severe combined immunodeficiency due to ADA deficiency, 102700
ADAR	133,6	99%	98%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	117,2	100%	89%	Aspartylglucosaminuria, 208400
AICDA	81	100%	99%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	72,5	98%	92%	Autoimmune polyendocrinopathy syndrome, with/without reversible metaphyseal dysplasia, 240300
AK2	75,1	79%	77%	Reticular dysgenesis, 267500
ALG13	57,9	93%	84%	Congenital disorder of glycosylation, type Is, 300884
AP3B1	109,5	100%	99%	Hermansky-Pudlak syndrome 2, 608233
APOL1	149,4	100%	100%	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
ATM	118	100%	99%	Ataxia-telangiectasia, 208900
BLM	118,7	100%	100%	Bloom syndrome, 210900
BLNK	97,3	97%	97%	Agammaglobulinemia 4, 613502
BLOC1S6	121,5	92%	86%	Hermansky-pudlak syndrome 9, 614171
BTK	53,4	99%	95%	Agammaglobulinemia, X-linked 1, 300755
C1QA	126,3	100%	98%	C1q deficiency, 613652
C1QB	101,5	96%	86%	C1q deficiency, 613652
C1QC	137,1	94%	80%	C1q deficiency, 613652
C1R	84,9	97%	93%	C1r/C1s deficiency, combined, 216950
C1S	105,2	100%	99%	C1s deficiency, 613783
C2	18,6	80%	40%	C2 deficiency, 217000
C3	99,1	99%	95%	C3 deficiency, 613779
C4A	2,2	5%	3%	C4a deficiency, 614380
C4B	1	3%	1%	C4B deficiency, 614379
C5	100,4	100%	99%	C5 deficiency, 609536
C6	114,8	100%	99%	C6 deficiency, 612446

C7	95,3	98%	93%	C7 deficiency, 610102
C8A	88,6	100%	99%	C8 deficiency, type I, 613790
C8B	96,3	100%	97%	C8 deficiency, type II, 613789
C9	112,3	100%	100%	C9 deficiency, 613825
CARD11	97,9	100%	98%	Persistent polyclonal B-cell lymphocytosis, 606445
CARD9	60,9	100%	97%	Candidiasis, familial, 2, autosomal recessive, 212050
CASP10	103,3	100%	100%	Autoimmune lymphoproliferative syndrome, type II, 603909
CASP8	121	100%	98%	Immunodeficiency due to CASP8 deficiency, 607271
CD19	78,8	100%	98%	Immunodeficiency, common variable, 3, 613493
CD247	87,8	100%	100%	Immunodeficiency due to defect in CD3-zeta, 610163
CD27	80,9	100%	98%	Lymphoproliferative syndrome 2, 615122
CD3D	89,5	100%	99%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3E	93,8	95%	86%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
CD3G	87,9	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	103,3	95%	91%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	63	91%	83%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	112,3	100%	100%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD55	100,6	75%	70%	[Blood group Cromer], 613793
CD59	103,9	85%	79%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	75,4	98%	85%	Agammaglobulinemia 3, 613501
CD79B	136,1	100%	99%	Agammaglobulinemia 6, 612692
CD81	84,4	98%	91%	Immunodeficiency, common variable, 6, 613496
CD8A	84,5	95%	93%	CD8 deficiency, familial, 608957
CEBPE	101,1	100%	100%	Specific granule deficiency, 245480
CFB	19,6	77%	40%	{Macular degeneration, age-related, reduced risk of}, 603075
CFD	49,9	97%	86%	Complement factor D deficiency, 613912
CFH	107,3	95%	92%	{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CFHR1	21,4	67%	44%	{Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	111,5	88%	74%	{Macular degeneration, age-related, reduced risk of}, 603075
CFHR5	108,2	94%	92%	Nephropathy due to CFHR5 deficiency, 614809
CFI	136	100%	100%	Complement factor I deficiency, 610984
CFP	50,1	97%	87%	Properdin deficiency,X-linked, 312060
CHD7	121,3	100%	99%	CHARGE syndrome, 214800

CIITA	98,1	98%	95%	Bare lymphocyte syndrome type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CLEC4D	102,3	100%	100%	No OMIM phenotype
CLEC7A	104,6	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108
COLEC11	117,5	100%	100%	3MC syndrome 2, 265050
CORO1A	95,2	92%	90%	Immunodeficiency 8, 615401
CR2	120,4	100%	100%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927
CREBBP	83,6	99%	97%	Rubinstein-Taybi syndrome, 180849
CSF2RA	0	0%	0%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF3R	80,4	100%	98%	Neutrophilia, hereditary, 162830
CTC1	95,5	100%	99%	Other Well-defined Immunodeficiency Syndromes;
CTSC	88,2	100%	100%	Papillon-Lefevre syndrome, 245000
CXCR4	226,9	100%	100%	WHIM syndrome, 193670
CYBA	29,7	65%	58%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	53,3	88%	81%	Chronic granulomatous disease, X-linked, 306400
DCLRE1C	99	90%	90%	Severe combined immunodeficiency, Athabascan type, 602450
DDX58	116,6	99%	97%	Singleton-Merten syndrome 2,616298
DHFR	51,9	81%	58%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	50	99%	91%	Dyskeratosis congenita, X-linked, 305000
DNMT3B	91,5	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK8	85,9	100%	98%	Mental retardation, autosomal dominant 2, 614113
ELANE	113,7	97%	80%	Neutropenia, cyclic, 162800
ELF4	51,9	99%	97%	No OMIM phenotype Hypogammaglobulinaemia (Stewart (2005) Curr Opin Allergy Clin Immunol 5, 510)
EPG5	89,4	100%	99%	Vici syndrome, 242840
ERCC2	88,6	99%	96%	Xeroderma pigmentosum, group D, 278730
ERCC3	123,3	100%	100%	Xeroderma pigmentosum, group B, 610651
F12	94,8	100%	96%	Factor XII deficiency, 234000
FADD	110,3	100%	100%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759
FAS	195,1	100%	100%	{Autoimmune lymphoproliferative syndrome}, 601859
FASLG	83,8	97%	92%	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FCGR1A	0	0%	0%	[IgG receptor I, phagocytic, familial deficiency of]

FCGR3A	45	45%	44%	Immunodeficiency 20, 615707
FCN3	112,7	99%	95%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	104,3	100%	97%	Leukocyte adhesion deficiency, type III, 612840
FOXN1	108,5	100%	99%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	38,4	92%	76%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FPR1	179,3	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	131,2	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	116,3	100%	100%	Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	58,3	95%	92%	Favism, 134700 Hemolytic anemia due to G6PD deficiency, 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GATA2	103,5	99%	92%	Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency, 614172
GFI1	61,8	100%	93%	Neutropenia, severe congenital 2, autosomal dominant, 613107
GJC2	57	99%	83%	Leukodystrophy, hypomyelinating, 2, 608804
GTF2H5	97	100%	100%	Trichothiodystrophy, complementation group A, 601675
HAX1	140,1	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
ICOS	133,8	100%	100%	Immunodeficiency, common variable, 1, 607594
IFIH1	134,4	100%	100%	Diseases of Immune Dysregulation;syndromaal;
IFNGR1	144,3	100%	100%	Mycobacterial infection, atypical, familial disseminated, 209950
IFNGR2	121,2	93%	93%	{Mycobacterial infection, atypical, familial disseminated},209950
IGLL1	25,5	74%	48%	Agammaglobulinemia 2, 613500
IKBKB	90,2	95%	92%	Combined T-cell and B-cell immunodeficiencies
IKBKG	12,6	26%	24%	Incontinentia pigmenti, type II, 308300
IKZF1	105,9	100%	99%	Leukemia,acute lymphoblastic Systemic lupus erythematosus, association with (Han (2009) Nat Genet 41,1234)
IL10RA	99,7	100%	100%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	111,2	98%	94%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL12B	93,1	100%	100%	BCG and salmonella infection, disseminated, 209950
IL12RB1	58,5	99%	91%	{Mycobacterial and salmonella infections, susceptibility to}, 209950
IL17F	90	97%	93%	Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	90,8	100%	92%	Candidiasis, familial, 5, autosomal recessive, 613953

IL1RN	113,6	100%	100%	{Gastric cancer risk after H. pylori infection}, 137215
IL2	102,6	100%	100%	Severe combined immunodeficiency due to IL2 deficiency Allergic disorders, association with (Christinsen (2006) Eur J Hum Genet 14,227)
IL21R	115,6	100%	99%	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 [IgE, elevated level of], 147050
IL2RA	101,4	100%	99%	Interleukin-2 receptor, alpha chain, deficiency of, 606367
IL2RG	46,6	98%	87%	Severe combined immunodeficiency, X-linked, 300400
IL36RN	98,7	100%	100%	Psoriasis, generalized pustular, 614204
IL7R	103,1	100%	100%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INSR	123,5	98%	95%	Leprechaunism, 246200
IRAK4	103,3	100%	100%	IRAK4 deficiency, 607676
IRF8	68,3	100%	98%	Monocyte and dendritic cell deficiency, recessive, 614894
ITCH	100,8	95%	94%	Autoimmune disease, syndromic multisystem, 613385
ITGB2	80	99%	96%	Leukocyte adhesion deficiency, 116920
ITK	100,2	100%	100%	Lymphoproliferative syndrome 1, 613011
JAK2	115,9	99%	98%	Erythrocytosis,somatic,133100 Leukemia,acute myelogenous,601626 Myelofibrosis,somatic,254450 Polycythemia vera,263300 Thrombocythemia 3,614521 {Budd-Chiari syndrome},600880
JAK3	90,1	99%	96%	SCID, autosomal recessive, T-negative/B-positive type, 600802
KMT2D	100,9	99%	98%	Other Well-defined Immunodeficiency Syndromes;syndromaal;
KRAS	67,2	97%	86%	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, 260350 SFM syndrome, somatic mosaic, 163200
LAMTOR2	79,1	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LCK	98,6	91%	83%	?Immunodeficiency 22, 615758

LIG1	77,8	99%	93%	DNA ligase I deficiency
LIG4	190,1	100%	100%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LPIN2	75,5	100%	97%	Majeed syndrome, 609628
LRBA	111,2	99%	98%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	126,9	100%	100%	Agammaglobulinemia 5, 613506
LYST	120,5	99%	96%	Chediak-Higashi syndrome, 214500
MAGT1	53,3	98%	98%	Mental retardation, X-linked 95, 300716
MAL	92,5	99%	88%	Meleda disease,248300
MAN2B1	80,2	97%	92%	Mannosidosis, alpha-, types I and II, 248500
MANBA	92,3	100%	99%	Mannosidosis, beta, 248510
MASP2	111,9	98%	93%	MASP2 deficiency, 613791
MBL2	128,2	100%	97%	{Chronic infections, due to MBL deficiency}, 614372
MC2R	149,2	100%	100%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM4	104,8	99%	97%	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MEFV	112,2	96%	96%	Familial Mediterranean fever, AR, 249100
MPO	89,1	100%	98%	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers} Cardiomyopathy, dilated, 1T, 613740
MRE11A	97,1	99%	99%	Ataxia-telangiectasia-like disorder, 604391
MS4A1	151,6	100%	100%	Immunodeficiency, common variable, 5, 613495
MTHFD1	98,7	99%	95%	{Abrubtio placentae, susceptibility to} {Spina bifida, folate-sensitive, susceptibility to}, 601634
MVK	88,3	100%	97%	Mevalonic aciduria, 610377
MYD88	167,1	100%	96%	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
NBN	121,4	98%	97%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCF1	0,9	0%	0%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	100,6	100%	98%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	79,4	97%	96%	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	80,7	97%	90%	Acne inversa, familial, 1, 142690
NDNL2	131	100%	100%	No OMIM phenotype

NFKB2	90,2	100%	98%	Immunodeficiency, common variable, 10, 615577
NFKBIA	100,8	100%	100%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NHEJ1	81,4	100%	99%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	52,1	100%	95%	Dyskeratosis congenita, autosomal recessive 2, 613987
NKX2-5	113	100%	99%	Atrial septal defect 7, with or without AV conduction defects, 108900
NLRP12	102,6	99%	98%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	124,1	100%	99%	Cold-induced autoinflammatory syndrome, familial, 120100
NOD2	96,2	100%	98%	{Inflammatory bowel disease 1}, 266600
NOP10	193,8	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	129,6	100%	100%	Autoimmune lymphoproliferative syndrome type IV, 614470
ORAI1	79,1	99%	87%	Immunodeficiency 9, 612782
PCCA	95,7	96%	94%	Propionicacidemia, 606054
PCCB	106,5	100%	98%	pccB complementation group Propionicacidemia, 606054
PEPD	66,9	97%	89%	Prolidase deficiency, 170100
PGM3	104,4	99%	94%	Immunodeficiency 23, 615816
PIGA	66,6	99%	98%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIK3CD	88,3	97%	90%	Immunodeficiency 14, 615513
PIK3R1	141,9	100%	100%	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PLCG2	107,3	99%	98%	Familial cold autoinflammatory syndrome 3, 614468
PLG	65,3	74%	70%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PMM2	92,4	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNP	124,9	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PRF1	94,1	100%	98%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553
PRKDC	89	100%	98%	Immunodeficiency 16 with or without neurologic abnormalities, 615966
PRPS1	62,3	99%	97%	Gout, PRPS-related, 300661
PSENEN	123,7	100%	100%	Acne inversa, familial, 2, 613736
PSMB8	10,1	40%	9%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSTPIP1	60,5	99%	85%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN11	46,9	88%	66%	Noonan syndrome 1, 163950
PTPRC	100	98%	93%	{Hepatitic C virus, susceptibility to}, 609532

PTRF	151,4	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
RAB27A	120,4	100%	100%	Griscelli syndrome, type 2, 607624
RAC2	53,9	100%	94%	Neutrophil immunodeficiency syndrome, 608203
RAG1	149,1	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457
RAG2	206,1	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457
RASGRP2	74,3	100%	96%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	80,8	90%	90%	Polyglucosan body myopathy, early-onset, with or without immunodeficiency, 615895
RECQL4	87,7	98%	95%	Rothmund-Thomson syndrome, 268400
RFX5	116	99%	97%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	94,1	97%	95%	MHC class II deficiency, complementation group B, 209920
RFXAP	86,6	93%	88%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RHOH	141,6	100%	100%	No OMIM phenotype RHOH deficiency (Crequer (2012) J Clin Invest 122, 3239)
RNASEH2A	90,2	100%	88%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	115	100%	100%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	162,6	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNF168	204,4	100%	100%	RIDDLE syndrome, 611943
RPSA	25,9	76%	54%	Asplenia, isolated congenital, 271400
RTEL1	77,2	98%	90%	Dyskeratosis congenita, autosomal recessive 5, 615190
SAMHD1	122,4	100%	99%	Aicardi-Goutieres syndrome 5, 612952
SBDS	93	99%	95%	Shwachman-Bodian-Diamond syndrome, 260400
SERAC1	89,3	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	120,7	96%	91%	Angioedema, hereditary, types I and II, 106100
SH2D1A	52,7	94%	83%	Lymphoproliferative syndrome, X-linked, 308240
SKIV2L	17,7	72%	36%	Trichohepatoenteric syndrome 2, 614602
SLC35A1	111,1	100%	100%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	100,2	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	83,8	99%	97%	Glycogen storage disease Ib, 232220
SLC39A4	65,8	100%	98%	Acrodermatitis enteropathica, 201100
SLC46A1	83,3	100%	97%	Folate malabsorption, hereditary, 229050
SMARCAL1	121,3	99%	96%	Schimke immunoosseous dysplasia, 242900
SP110	93,7	100%	100%	Hepatic venoocclusive disease with immunodeficiency, 235550

SPINK5	99,5	100%	97%	Netherton syndrome, 256500
STAT1	89,4	100%	99%	Mycobacterial infection, atypical, familial disseminated, 209950
STAT2	132,7	100%	100%	No OMIM phenotype STAT2 deficiency (Hambleton (2013) Proc Natl Acad Sci USA 110,3053) Autism (Neale (2012) Nature 485, 242) Psoriasis susceptibility, association with (Tsoi (2012) Nat Genet 44, 1341)
STAT3	84,3	98%	93%	Hyper-IgE recurrent infection syndrome, 147060
STAT4	110,6	100%	98%	{Systemic lupus erythematosus, susceptitbility to, 11}, 612253
STAT5B	73,1	84%	74%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, STAT5B/RARA type
STIM1	87,9	98%	95%	Immunodeficiency 10, 612783
STK4	97,2	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	173,6	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	81,3	100%	96%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TAP1	13,3	61%	17%	Bare lymphocyte syndrome, type I, 604571
TAP2	11,2	34%	16%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	16,5	63%	26%	Bare lymphocyte syndrome, type I, 604571
TAZ	47,3	100%	98%	Barth syndrome, 302060
TBX1	64,9	77%	72%	Conotruncal anomaly face syndrome, 217095
TCIRG1	76,7	95%	84%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	103,4	100%	97%	linked to P1 Transcobalamin II deficiency, 275350
TERT	109,7	99%	96%	{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 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134
THBD	75,2	100%	100%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TICAM1	76,1	100%	100%	{Encephalopathy, acute, infection-induced, susceptibility to, 6}, 614850
TINF2	187,6	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990
TLR3	179,9	100%	100%	{Herpes simplex encephalitis, susceptibility to, 2} 613002
TMC6	59,4	100%	93%	Epidermodysplasia verruciformis, 226400

TMC8	79,4	100%	98%	Epidermodysplasia verruciformis, 226400
TNFRSF11A	106,8	95%	93%	Osteolysis, familial expansile, 174810
TNFRSF13B	60,1	100%	93%	Immunoglobulin A deficiency 2, 609529
TNFRSF13C	51,2	100%	73%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	65,4	92%	86%	Periodic fever, familial, 142680
TNFRSF4	55,6	99%	86%	?Immunodeficiency 16, 615593
TRAF3	122,5	100%	100%	{Herpes simplex encephalitis, susceptibility to, 3}, 614849
TREX1	120,7	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750
TTC37	116,5	100%	100%	Trichohepatoenteric syndrome 1, 222470
TTC7A	65,2	95%	94%	Intestinal atresia, multiple, 243150
TYK2	85,9	99%	96%	Tyrosine kinase 2 deficiency, 611521
UNC119	113	100%	100%	CD4 lymphopenia, idiopathic (Gorska (2012) Blood 119, 1399) Cone-rod dystrophy, late-onset (Kobayashi (2000) Invest Ophthalmol Vis Sci 41, 3268) Cone-rod dystrophy (Huang (2013) Mol Med Rep epub) ?Immunodeficiency 13, 615518
UNC13D	60,5	97%	90%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	40,9	55%	54%	s simplex encephalitis, susceptibility to, 1, 610551
UNG	73,7	93%	91%	Immunodeficiency with hyper IgM, type 5, 608106
USB1	52	88%	83%	Poikiloderma with neutropenia, 604173
VPS13B	113,3	99%	98%	Cohen syndrome, 216550
VPS45	99,4	97%	94%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	27,3	89%	69%	Wiskott-Aldrich syndrome, 301000
WIPF1	99,6	97%	95%	Wiskott-Aldrich syndrome 2, 614493
WRAP53	133,2	100%	99%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	69,4	84%	76%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	94,2	93%	93%	Selective T-cell defect, 269840
ZBTB24	150,5	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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 : June 30th, 2015

This list is accurate for all panel versions starting with DG 2.3. (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