PRIMARY IMMUNODEFICIENCIES GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ACP5	98,7	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	61,8	100%	95%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADA	72,3	100%	96%	Severe combined immunodeficiency due to ADA deficiency, 102700 Adenosine deaminase deficiency, partial, 102700
AGA	111,6	100%	91%	Aspartylglucosaminuria, 208400
AICDA	80,4	100%	96%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	71,1	98%	87%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	70,8	78%	74%	Reticular dysgenesis, 267500
ALG13	111,6	96%	95%	Congenital disorder of glycosylation, type Is, 300884
AP3B1	104,8	100%	99%	Hermansky-Pudlak syndrome 2, 608233
APOL1	141	100%	100%	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 {End-stage renal disease, nondiabetic, susceptibility to}, 612551
ATM	111,1	99%	99%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell T-cell prolymphocytic leukemia, somatic
BLM	114,8	100%	98%	Bloom syndrome, 210900
BLNK	102,4	100%	100%	Agammaglobulinemia 4, 613502

BLOC1S6	114,4	96%	86%	Hermansky-pudlak syndrome 9, 614171
BTK	98,6	100%	99%	Agammaglobulinemia, X-linked 1, 300755 Agammaglobulinemia and isolated hormone deficiency, 307200
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	95,2	100%	97%	C1r/C1s deficiency, combined, 216950 (1)
C1S	95,5	99%	99%	C1s deficiency, 613783
C2	15,6	76%	27%	C2 deficiency, 217000 {Macular degeneration, age-related, reduced risk of}, 603075
C3	99,1	97%	93%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	1,6	4%	2%	C4a deficiency, 614380 [Blood group, Rodgers], 614374 ?Systemic lupus erythematosus, susceptibility to or protection against}, 152700 (2)
C4B	1,6	4%	2%	C4B deficiency, 614379
C5	97	100%	99%	C5 deficiency, 609536
C6	111,6	100%	99%	C6 deficiency, 612446 Combined C6/C7 deficiency
C7	90,4	99%	95%	C7 deficiency, 610102
C8A	77,2	100%	98%	C8 deficiency, type I, 613790
C8B	91,8	100%	96%	C8 deficiency, type II, 613789
C9	109,2	100%	100%	C9 deficiency, 613825 C9 deficiency with dermatomyositis, 613825

CARD11	93	100%	98%	Persistent polyclonal B-cell lymphocytosis, 606445 Immunodeficiency 11, 615206
CARD9	64,5	99%	98%	Candidiasis, familial, 2, autosomal recessive, 212050
CASP10	107,1	100%	100%	Autoimmune lymphoproliferative syndrome, type II, 603909 Non-Hodgkin lymphoma, somatic, 605027 Gastric cancer, somatic, 613659
CASP8	117,9	100%	97%	Immunodeficiency due to CASP8 deficiency, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
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	84,2	100%	91%	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 due to defect in CD3-gamma
CD40	105	95%	92%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	117,3	99%	96%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	112,1	100%	100%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 -3
CD55	103,1	89%	85%	[Blood group Cromer], 613793
CD59	113,5	100%	100%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	79,1	95%	87%	Agammaglobulinemia 3, 613501
CD79B	113,8	100%	100%	Agammaglobulinemia 6, 612692

CD81	59,4	99%	89%	Immunodeficiency, common variable, 6, 613496
CD8A	82,6	100%	99%	CD8 deficiency, familial, 608957
CEBPE	102,2	100%	100%	Specific granule deficiency, 245480
CFB	19,5	70%	37%	{Macular degeneration, age-related, reduced risk of}, 603075 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924
CFD	49,4	96%	77%	Complement factor D deficiency, 613912
CFH	105,6	95%	92%	{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 Complement factor H deficiency, 609814 {Macular degeneration, age-related, 4}, 610698 Basal laminar drusen, 126700
CFHR1	25,5	68%	51%	{Macular degeneration, age-related, reduced risk of}, 603075 {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR3	145,7	89%	80%	{Macular degeneration, age-related, reduced risk of}, 603075 {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR5	95,7	94%	86%	Nephropathy due to CFHR5 deficiency, 614809
CFI	131,3	100%	100%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFP	91,9	98%	93%	Properdin deficiency,X-linked
CHD7	117,2	100%	99%	CHARGE syndrome, 214800 {Scoliosis, idiopathic 3}, 608765 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CLEC4D	90,6	100%	99%	No OMIM phenotype
CLEC7A	103,3	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
COLEC11	118,2	100%	100%	3MC syndrome 2, 265050
CORO1A	91,8	85%	85%	Immunodeficiency 8, 615401

CR2	108,9	100%	100%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 Immunodeficiency, common variable, 7, 614699
CREBBP	77,5	99%	97%	Rubinstein-Taybi syndrome, 180849
CSF2RA	0	0%	0%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF3R	82,8	100%	98%	Neutrophilia, hereditary, 162830
CTSC	94,7	100%	99%	Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010 Periodontitis 1, juvenile, 170650
CXCR4	193,2	100%	100%	WHIM syndrome, 193670 Myelokathexis, isolated
CYBA	38,2	92%	71%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	94,4	92%	88%	Chronic granulomatous disease, X-linked, 306400 Atypical mycobacteriosis, familial, X-linked 2, 300645
DCLRE1C	104,2	97%	97%	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DHFR	50,6	79%	63%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	91,1	100%	98%	Dyskeratosis congenita, X-linked, 305000
DNMT3B	93,3	100%	97%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK8	84	100%	98%	Mental retardation, autosomal dominant 2, 614113 Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	109,7	99%	92%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	106,8	100%	100%	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 Trichothiodystrophy, 601675 Cerebrooculofacioskeletal syndrome 2, 610756

ERCC3	121,5	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy, 601675
F12	96,7	100%	99%	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
FADD	100,9	100%	98%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759
FAS	189,9	100%	99%	{Autoimmune lymphoproliferative syndrome}, 601859 Squamous cell carcinoma, burn scar-related, somatic Autoimmune lymphoproliferative syndrome, type IA, 601859
FCGR1A	0	0%	0%	[IgG receptor I, phagocytic, familial deficiency of]
FCGR3A	54	56%	50%	{Viral infections, recurrent}
FCN3	99,7	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	69,7	98%	91%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FPR1	160,4	100%	100%	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 Dursun syndrome, 612541
G6PD	100,2	95%	95%	Hemolytic anemia due to G6PD deficiency Favism, 134700 {Resistance to malaria due to G6PD deficiency}, 611162
GATA2	96,6	96%	91%	Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency, 614172 Emberger syndrome, 614038 {Myelodysplastic syndrome, susceptibility to}, 614286 {Leukemia, acute myeloid, susceptibility to}, 601626
GFI1	71,2	100%	98%	Neutropenia, severe congenital 2, autosomal dominant, 613107 Neutropenia, nonimmune chronic idiopathic, of adults, 607847

GJC2	52,1	92%	82%	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480
GTF2H5	89,9	100%	100%	Trichothiodystrophy, complementation group A, 601675
HAX1	129,3	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
ICOS	125,5	100%	100%	Immunodeficiency, common variable, 1, 607594
IFNGR1	141,9	100%	100%	Mycobacterial infection, atypical, familial disseminated, 209950 BCG infection, generalized familial, 209950 {H. pylori infection, susceptibility to}, 600263 {Tuberculosis, susceptibility to}, 607948 {Mycobacterium tuberculosis infection, protection against}, 607948 {Hepatitis B virus infection, susceptibility to}, 610424
IFNGR2	109,2	92%	92%	{Mycobacterial infection, atypical, familial disseminated},209950
IGLL1	20,7	72%	35%	Agammaglobulinemia 2, 613500
IKBKG	18,8	20%	20%	Incontinentia pigmenti, type II, 308300 Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency, isolated, 300584 {Atypical mycobacteriosis, familial}, 300636 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IKZF1	104	100%	99%	Leukemia, acute lymphoblastic
IL10RA	97	100%	98%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL12B	85,1	100%	100%	BCG and salmonella infection, disseminated, 209950 {Asthma, susceptibility to}, 600807
IL12RB1	60,4	100%	90%	{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	106	100%	100%	{Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628 Interleukin 1 receptor antagonist deficiency, 612852
IL2	93,9	100%	100%	Severe combined immunodeficiency due to IL2 deficiency (1)
IL21R	110,7	100%	99%	[IgE, elevated level of], 147050 Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207
IL2RA	98,1	100%	100%	Interleukin-2 receptor, alpha chain, deficiency of, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	93,2	100%	95%	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL36RN	89	100%	100%	Psoriasis, generalized pustular, 614204
IL7R	88,9	100%	95%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INSR	117,8	96%	92%	Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
IRAK4	109,6	100%	100%	IRAK4 deficiency, 607676 Invasive pneumococcal disease, recurrent isolated, 1, 610799
IRF8	66,2	100%	98%	Monocyte and dendritic cell deficiency, recessive, 614894 CD11C+/CD1C+ dendritic cell deficiency, dominant, 614893
ITCH	108,3	100%	99%	Autoimmune disease, syndromic multisystem, 613385
ITGB2	87,2	100%	99%	Leukocyte adhesion deficiency, 116920
ITK	100,4	100%	100%	Lymphoproliferative syndrome 1, 613011

JAK2	109,7	100%	99%	Polycythemia vera, 263300 Thrombocythemia 3, 614521 Myelofibrosis, somatic, 254450 {Budd-Chiari syndrome}, 600880 Leukemia, acute myelogenous, 601626 Erythrocytosis, somatic, 133100
JAK3	84,9	98%	94%	SCID, autosomal recessive, T-negative/B-positive type, 600802
KRAS	63,6	98%	93%	Lung cancer, somatic, 211980 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Gastric cancer, somatic, 137215 Leukemia, acute myelogenous Noonan syndrome 3, 609942 Cardiofaciocutaneous syndrome 2, 615278 Breast cancer, somatic, 114480 SFM syndrome, somatic mosaic, 163200
LAMTOR2	73,8	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798 -3
LCK	83,6	91%	80%	SCID due to LCK deficiency (1)
LIG1	74,5	98%	91%	DNA ligase I deficiency
LIG4	177,1	100%	100%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500 Severe combined immunodeficiency with sensitivity to ionizing radiation, 602450
LPIN2	75,8	99%	95%	Majeed syndrome, 609628
LRBA	103,8	100%	97%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	122,9	100%	100%	Agammaglobulinemia 5, 613506
LYST	116,3	99%	97%	Chediak-Higashi syndrome, 214500
MAGT1	97,6	100%	100%	Mental retardation, X-linked 95, 300716 Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853

MAL	85,6	89%	79%	No OMIM phenotype
MAN2B1	81,3	99%	92%	Mannosidosis, alpha-, types I and II, 248500
MANBA	87,6	100%	99%	Mannosidosis, beta, 248510
MASP2	114,4	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	97,8	100%	97%	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MEFV	117,3	100%	99%	Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MPO	88,6	100%	98%	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}
MPO	88,6	100%	98%	Cardiomyopathy, dilated, 1T, 613740
MRE11A	90,5	100%	100%	Ataxia-telangiectasia-like disorder, 604391
MS4A1	127,7	100%	100%	Immunodeficiency, common variable, 5, 613495
MVK	86,1	100%	99%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
MYD88	165,1	100%	99%	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 Macroglobulinemia, Waldenstrom, somatic, 153600
NCF1	0,5	0%	0%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	95,2	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	78,4	97%	92%	Acne inversa, familial, 1, 142690

NDNL2	110,6	100%	100%	No OMIM phenotype
NFKBIA	95,9	100%	99%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NHEJ1	80,9	100%	94%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	43,2	100%	95%	Dyskeratosis congenita, autosomal recessive 2, 613987
NKX2-5	125,3	100%	99%	Atrial septal defect 7, with or without AV conduction defects, 108900 Tetrology of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Ventricular septal defect 3, 614432 Hypoplastic left heart syndrome 2, 614435 Conotruncal heart malformations, variable, 217095
NLRP12	103,8	100%	99%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	117,3	100%	99%	Cold-induced autoinflammatory syndrome, familial, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115
NOD2	91,7	100%	98%	{Inflammatory bowel disease 1}, 266600 Blau syndrome, 186580 {Psoriatic arthritis, susceptibility to}, 607507 Sarcoidosis, early-onset, 609464
NOP10	169,5	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	135	100%	100%	Autoimmune lymphoproliferative syndrome type IV, 614470 Noonan syndrome 6, 613224 Epidermal nevus, somatic, 162900 Thyroid carcinoma, follicular, somatic, 188470 Colorectal cancer, somatic, 114500
ORAI1	77,2	92%	88%	Immunodeficiency 9, 612782
PCCA	95,3	98%	95%	Propionicacidemia, 606054
PCCB	106,8	100%	100%	pccB complementation group Propionicacidemia, 606054

PEPD	63,3	100%	89%	Prolidase deficiency, 170100
PIGA	134,6	100%	99%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIK3CD	89	99%	93%	?Agammaglobulinemia 7, autosomal recessive, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880
PIK3R1	144,8	100%	100%	Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PLCG2	105,4	100%	99%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLG	64,4	75%	68%	Plasminogen Tochigi disease Thrombophilia, dysplasminogenemic (1) Plasminogen deficiency, types I and II (1) Conjunctivitis, ligneous, 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 Lymphoma, non-Hodgkin, 605027
PRKDC	85,8	99%	96%	Immunodeficiency 16 with or without neurologic abnormalities, 615966
PRPS1	131,2	100%	100%	Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Arts syndrome, 301835 Deafness, X-linked 1, 304500
PSENEN	119,6	100%	100%	Acne inversa, familial, 2, 613736
PSMB8	9,4	40%	3%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSTPIP1	58	96%	89%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 -3

PTPN11	41,7	83%	68%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250
PTPRC	106,1	99%	98%	{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	114,6	100%	100%	Griscelli syndrome, type 2, 607624
RAC2	59,1	99%	95%	Neutrophil immunodeficiency syndrome, 608203
RAG1	132,2	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457 Omenn syndrome, 603554 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	187,2	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457 Omenn syndrome, 603554 Combined cellular and humoral immune defects with granulomas, 233650
RASGRP2	80,2	100%	96%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	77,3	94%	90%	Polyglucosan body myopathy, early-onset, with or without immunodeficiency, 615895
RECQL4	91,5	98%	96%	Rothmund-Thomson syndrome, 268400 RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600
RFX5	106,5	99%	99%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	89,6	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%	non-Hodgkin lymphoma
RNASEH2A	96,1	99%	94%	Aicardi-Goutieres syndrome 4, 610333

RNASEH2B	102,3	99%	97%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	133	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNF168	193,1	100%	100%	RIDDLE syndrome, 611943
RPSA	22,6	77%	50%	Asplenia, isolated congenital
RTEL1	82,1	99%	95%	Dyskeratosis congenita, autosomal recessive 5, 615190 Dyskeratosis congenita, autosomal dominant 4, 615190
SAMHD1	112,7	100%	98%	Aicardi-Goutieres syndrome 5, 612952 Chilblain lupus 2, 614415 -3
SBDS	88,2	98%	93%	Shwachman-Bodian-Diamond syndrome, 260400
SERAC1	88,8	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	119,2	97%	95%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SH2D1A	87	99%	99%	Lymphoproliferative syndrome, X-linked, 308240
SKIV2L	17,7	68%	36%	Trichohepatoenteric syndrome 2, 614602
SLC35A1	103,6	100%	99%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	96,6	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	84,9	100%	97%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	69,9	100%	97%	Acrodermatitis enteropathica, 201100
SLC46A1	80,3	100%	98%	Folate malabsorption, hereditary, 229050
SMARCAL1	118,1	99%	97%	Schimke immunoosseous dysplasia, 242900
SP110	96,2	100%	100%	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948

SPINK5	97,3	100%	99%	Netherton syndrome, 256500 Atopy, 147050
STAT1	85,9	100%	98%	Mycobacterial infection, atypical, familial disseminated, 209950 Mycobacterial and viral infections, susceptibility to, autosomal recessive, 613796 Candidiasis, familial, 7, 614162
STAT2	128,6	100%	100%	Autism (Neale (2012) Nature 485,242) Psoriasis susceptibility, association with (Tsoi (2012) Nat Genet 44, 1341) STAT2 deficiency (Hambleton (2013) Proc Natl Acad Sci USA 110,3053)
STAT3	85	100%	96%	Hyper-IgE recurrent infection syndrome, 147060
STAT4	107,8	100%	98%	{Systemic lupus erythematosus, susceptitbility to, 11}, 612253
STAT5B	72,5	83%	75%	Leukemia, acute promyelocytic, STAT5B/RARA type Growth hormone insensitivity with immunodeficiency, 245590
STIM1	80,7	99%	96%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 160565
STK4	99,9	100%	99%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	169,7	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	84,6	100%	94%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TAP1	11,1	47%	9%	Bare lymphocyte syndrome, type I, 604571
TAP2	10,3	29%	13%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis
TAPBP	19	67%	31%	Bare lymphocyte syndrome, type I, 604571
TAZ	102,3	100%	100%	Barth syndrome, 302060
TBX1	66,3	72%	66%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Velocardiofacial syndrome, 192430 Tetralogy of Fallot, 187500
TCIRG1	74,8	92%	84%	Osteopetrosis, autosomal recessive 1, 259700

TCN2	104,1	100%	97%	linked to P1 Transcobalamin II deficiency, 275350
TERT	99,5	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 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134
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	170,7	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TLR3	160,4	100%	100%	{Herpes simplex encephalitis, susceptibility to, 2} 613002 {HIV1 infection, resistance to}, 609423
TMC6	57,9	99%	94%	Epidermodysplasia verruciformis, 226400
TMC8	76,1	99%	94%	Epidermodysplasia verruciformis, 226400
TNFRSF11A	100,8	95%	93%	Osteolysis, familial expansile, 174810 Paget disease of bone, 602080 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF13B	59	99%	93%	Immunoglobulin A deficiency 2, 609529 Immunodeficiency, common variable, 2, 240500
TNFRSF13C	54,1	100%	81%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	72	96%	91%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TRAF3	113,1	100%	98%	{Herpes simplex encephalitis, susceptibility to, 3}, 614849
TREX1	135,6	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

TTC37	111,4	100%	100%	Trichohepatoenteric syndrome 1, 222470
TYK2	86,4	99%	95%	Tyrosine kinase 2 deficiency, 611521
UNC119	112,9	100%	100%	Cone-rod dystrophy
UNC13D	60,8	95%	91%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	40,3	54%	53%	Herpes simplex encephalitis, susceptibility to, 1, 610551
UNG	67,5	92%	85%	Immunodeficiency with hyper IgM, type 5, 608106
VPS13B	104,4	99%	98%	Cohen syndrome, 216550
WAS	60,1	100%	90%	Wiskott-Aldrich syndrome, 301000 Thrombocytopenia, X-linked, 313900 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900
WIPF1	92,2	97%	96%	Wiskott-Aldrich syndrome 2, 614493
WRAP53	133,9	100%	99%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	130,3	90%	82%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	86,7	96%	87%	Selective T-cell defect, 269840
ZBTB24	162,4	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding region that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding region that is covered at least 20x
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors