PRIMARY IMMUNODEFICIENCIES GENE PANEL DG 2.7/DG 2.8

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACP5	224.9	100%	99%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	134.1	98%	93%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ADA	118	99%	97%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADAM17	140.9	97%	93%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAR	131.7	100%	99%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	154.1	100%	100%	Aspartylglucosaminuria, 208400
AICDA	150.7	97%	91%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	83.3	99%	93%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	117	98%	94%	Reticular dysgenesis, 267500
ALG13	107.4	98%	94%	Epileptic encephalopathy, early infantile, 36, 300884
AP3B1	111.5	97%	91%	Hermansky-Pudlak syndrome 2, 608233
APOL1	192.4	100%	100%	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
ATM	124.3	98%	93%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480
BLM	139.6	98%	95%	Bloom syndrome, 210900
BLNK	121.5	93%	90%	Agammaglobulinemia 4, 613502
BLOC1S6	106.6	98%	92%	Hermansky-pudlak syndrome 9, 614171
BTK	150.9	100%	99%	Agammaglobulinemia and isolated hormone deficiency, 307200 Agammaglobulinemia, X-linked 1, 300755
C1QA	130.2	98%	95%	C1q deficiency, 613652
C1QB	195	99%	99%	C1q deficiency, 613652

C1QC	232.3	100%	99%	C1q deficiency, 613652
C1R	168.3	100%	100%	C1r/C1s deficiency, combined, 216950
C1S	144.7	100%	99%	C1s deficiency, 613783
C2	18.5	80%	34%	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C3	170.8	100%	99%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C4A	20.7	75%	43%	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	19	72%	38%	C4B deficiency, 614379
C5	150	97%	94%	C5 deficiency, 609536 [Eculizumab, poor response to], 615749
C6	179.3	99%	99%	C6 deficiency, 612446 Combined C6/C7 deficiency
C7	153.6	98%	94%	C7 deficiency, 610102
C8A	129.8	100%	99%	C8 deficiency, type I, 613790
C8B	153.9	100%	99%	C8 deficiency, type II, 613789
C9	152	99%	98%	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591
CARD11	172.6	99%	97%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11, 615206
CARD14	120.7	99%	98%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	126.3	97%	96%	Candidiasis, familial, 2, autosomal recessive, 212050
CASP10	123.6	99%	96%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP8	179.3	100%	99%	Hepatocellular carcinoma, somatic, 114550 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 {Breast cancer, protection against}, 114480 {Lung cancer, protection against}, 211980
CD19	100.7	99%	98%	Immunodeficiency, common variable, 3, 613493
CD247	124.3	100%	100%	?Immunodeficiency 25, 610163

CD27	126.3	99%	99%	Lymphoproliferative syndrome 2, 615122
CD3D	217.5	100%	100%	Immunodeficiency 19, 615617
CD3E	172.6	100%	99%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	189	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	205.1	100%	99%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	151	96%	90%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	140.5	97%	92%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD55	154.8	91%	83%	[Blood group Cromer], 613793
CD59	234.2	92%	86%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	146.6	99%	97%	Agammaglobulinemia 3, 613501
CD79B	241.5	100%	100%	Agammaglobulinemia 6, 612692
CD81	162.7	100%	99%	Immunodeficiency, common variable, 6, 613496
CD8A	108	99%	98%	CD8 deficiency, familial, 608957
CDCA7	113.4	99%	98%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDKN2B	72.8	99%	99%	No OMIM phenotype Renal cell carcinoma (Jafri (2015) Cancer Discov 5, 723) Multiple endocrine neoplasia 1 (Agarwal (2009) J Clin Endocrinol Metab 94, 1826) ?Melanoma (Foley (2015) EBioMedicine 2,74) ?Parathyroid adenoma (Costa-Guda (2013) Horm Cancer 4, 301)
CEBPE	78.4	99%	98%	Specific granule deficiency, 245480
CECR1	111.6	99%	98%	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410
CFB	21.3	81%	45%	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFD	74.2	95%	86%	Complement factor D deficiency, 613912
CFH	193.2	98%	96%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	207	92%	90%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	107.1	85%	80%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400

				{Macular degeneration, age-related, reduced risk of}, 603075
CFHR5	112.2	98%	94%	Nephropathy due to CFHR5 deficiency, 614809
CFI	174.8	97%	96%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFP	103.5	98%	93%	Properdin deficiency,X-linked, 312060
CHD7	161	99%	98%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CIITA	133.9	100%	99%	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CLEC4D	151.1	100%	100%	No OMIM phenotype
CLEC7A	170.2	100%	99%	Candidiasis, familial, 4, autosomal recessive, 613108 {Aspergillosis, susceptibility to}, 614079
COLEC11	220.9	100%	100%	3MC syndrome 2, 265050
COPA	147.5	100%	99%	{Autoimmune interstitial lung, joint, and kidney disease}, 616414
CORO1A	172.4	98%	94%	Immunodeficiency 8, 615401
CR2	183.5	100%	99%	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927
CREBBP	147.6	99%	96%	Rubinstein-Taybi syndrome, 180849
CSF2RA	61.2	89%	86%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF3R	100.3	98%	95%	?Neutrophilia, hereditary, 162830
CTC1	118.9	99%	99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	227.1	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	148.6	100%	100%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CXCR4	210.4	100%	99%	Myelokathexis, isolated WHIM syndrome, 193670
CYBA	99.1	86%	79%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	143.1	100%	99%	Chronic granulomatous disease, X-linked, 306400

				Immunodeficiency 34, mycobacteriosis, X-linked, 300645
DCLRE1C	144.2	97%	94%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDX58	146.4	98%	96%	Singleton-Merten syndrome 2, 616298
DHFR	55	90%	73%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	138.1	99%	98%	Dyskeratosis congenita, X-linked, 305000
DNMT3B	141	100%	99%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	161.1	100%	99%	Immunodeficiency 40, 616433
DOCK8	151.8	100%	99%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	103.8	99%	97%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	93.1	99%	97%	No OMIM phenotype
EPG5	144.7	99%	97%	Vici syndrome, 242840
ERCC2	143.5	100%	99%	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	117.6	99%	98%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
F12	123	99%	99%	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
FADD	142.2	99%	98%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759
FAS	303	99%	99%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic {Autoimmune lymphoproliferative syndrome}, 601859
FASLG	97.9	100%	99%	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FCGR1A	69.2	47%	42%	[IgG receptor I, phagocytic, familial deficiency of]
FCGR3A	226.3	100%	99%	Immunodeficiency 20, 615707
FCN3	143.3	99%	99%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	132.4	99%	99%	Leukocyte adhesion deficiency,type III,612840
FOXN1	110.9	99%	98%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	136.8	98%	91%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100

FPR1	228.6	100%	99%	No OMIM phenotype
G6PC	219.4	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	138.4	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	138	99%	98%	Favism, 134700 Hemolytic anemia due to G6PD deficiency, 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GATA2	145.6	100%	99%	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GFI1	99.2	98%	92%	Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GJC2	52.2	85%	68%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GTF2H5	149.4	100%	99%	Trichothiodystrophy 3, photosensitive, 616395
HAX1	148	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HELLS	106.6	91%	86%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4,616911
ICOS	168.7	100%	99%	Immunodeficiency, common variable, 1, 607594
IFIH1	130	98%	96%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNGR1	153.4	99%	97%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948
IFNGR2	169.1	94%	93%	Immunodeficiency 28, mycobacteriosis, 614889
IGLL1	73	98%	92%	Agammaglobulinemia 2, 613500
IKBKB	135.8	98%	94%	Immunodeficiency 15, 615592
IKBKG	56.2	84%	70%	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636

				Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IKZF1	188.9	100%	99%	Immunodeficiency,common variable, 1,616873
IL10RA	156.7	100%	99%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	214.3	98%	96%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 {Hepatitis B virus, susceptibility to}, 610424
IL12B	125.9	100%	99%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	137.3	97%	95%	Immunodeficiency 30, 614891
IL17F	89.2	99%	95%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	136.4	99%	96%	?Candidiasis, familial, 5, autosomal recessive, 613953
IL1RN	186.9	100%	99%	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL2	73.6	94%	85%	Severe combined immunodeficiency due to IL2 deficiency
IL21R	144.1	100%	99%	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 [IgE, elevated level of], 147050
IL2RA	143.5	100%	99%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RG	78.6	100%	98%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL36RN	106.9	100%	99%	Psoriasis 14, pustular, 614204
IL7R	154.9	100%	99%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INSR	146.5	96%	94%	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
IRAK4	105	97%	88%	Invasive pneumococcal disease, recurrent isolated, 1, 610799 IRAK4 deficiency, 607676
IRF7	101.2	99%	99%	?Immunodeficiency 39, 616345
IRF8	124.3	99%	97%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 614894
ISG15	167.8	100%	100%	Immunodeficiency 38, 616126
ITCH	136.2	95%	94%	Autoimmune disease, multisystem, with facial dysmorphism, 613385

ITGB2	174.4	100%	99%	Leukocyte adhesion deficiency, 116920
ITK	141	99%	98%	Lymphoproliferative syndrome 1, 613011
JAK2	103	95%	92%	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600800
JAK3	111.6	98%	94%	SCID, autosomal recessive, T-negative/B-positive type, 600802
KMT2D	162.1	99%	99%	Kabuki syndrome 1, 147920
KRAS	72.1	99%	96%	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-Mims syndrome, somatic mosaic, 163200
LAMTOR2	175.9	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LCK	176.3	97%	95%	?Immunodeficiency 22, 615758
LIG1	102.6	99%	99%	DNA ligase I deficiency
LIG4	181.8	100%	99%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LPIN2	129.6	99%	99%	Majeed syndrome, 609628
LRBA	150	98%	97%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	290.6	99%	99%	Agammaglobulinemia 5, 613506
LTBP3	126.4	98%	96%	Dental anomalies and short stature, 601216
LYST	151.3	97%	94%	Chediak-Higashi syndrome, 214500
MAGT1	130.4	98%	95%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAL	158.4	100%	100%	No OMIM phenotype
MAN2B1	137.2	98%	96%	Mannosidosis, alpha-, types I and II, 248500

MANBA	144.2	99%	95%	Mannosidosis, beta, 248510
MASP2	165.4	99%	98%	MASP2 deficiency, 613791
MBL2	137.1	99%	99%	{Chronic infections, due to MBL deficiency}, 614372
MC2R	233.7	100%	99%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM4	182.8	99%	98%	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981
MEFV	131.1	95%	91%	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MKL1	119.5	98%	95%	Megakaryoblastic leukemia, acute
MPO	164.8	100%	99%	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}
MRE11A	57.6	95%	85%	Ataxia-telangiectasia-like disorder, 604391
MS4A1	138	99%	95%	Immunodeficiency, common variable, 5, 613495
MTHFD1	157.8	99%	97%	{Abruptio placentae, susceptibility to} {Spina bifida, folate-sensitive, susceptibility to}, 601634
MVK	146.1	100%	99%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYD88	207.3	99%	99%	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
NBN	89.5	98%	94%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NCF1	21.9	25%	22%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	138.1	99%	98%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	174.6	100%	99%	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	127.9	100%	99%	Acne inversa, familial, 1, 142690
NDNL2	139	98%	95%	No OMIM phenotype
NFKB1	122.7	99%	95%	Immunodeficiency, common variable, 12, 616576
NFKB2	139.3	98%	95%	Immunodeficiency, common variable, 10, 615577
NFKBIA	117.7	98%	94%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NHEJ1	87.6	100%	98%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	87.3	99%	98%	Dyskeratosis congenita, autosomal recessive 2, 613987

NKX2-5	94.7	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	191.4	100%	99%	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
NLRP1	137	99%	96%	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP12	168.8	99%	99%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	153.2	100%	99%	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900
NOD2	148.9	100%	99%	Blau syndrome, 186580 Sarcoidosis, early-onset, 609464 {Inflammatory bowel disease 1}, 266600 {Psoriatic arthritis, susceptibility to}, 607507
NOP10	189.4	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	205.7	100%	100%	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 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470
ORAI1	223.9	93%	91%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
PARN	140.4	99%	97%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PBX1	123.1	98%	95%	Leukemia, acute pre-B-cell, 176310
PCCA	116.6	96%	91%	Propionicacidemia, 606054
PCCB	160.5	97%	95%	Propionicacidemia, 606054
PEPD	121.4	99%	98%	Prolidase deficiency, 170100

PGM3	212.5	100%	99%	Immunodeficiency 23, 615816
PIGA	102.1	92%	84%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIK3CD	149.1	99%	97%	Immunodeficiency 14, 615513
PIK3R1	144	99%	96%	Immunodeficiency 36, 616005 SHORT syndrome, 269880 ?Agammaglobulinemia 7, autosomal recessive, 615214
PLCG2	133.6	100%	99%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLG	133.1	87%	87%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PMM2	178.4	99%	99%	Congenital disorder of glycosylation, type Ia, 212065
PNP	148.5	100%	99%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PRF1	130.4	100%	99%	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKDC	120.4	97%	93%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	201.5	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	93.7	99%	99%	Acne inversa, familial, 2, 613736
PSMB8	16.1	57%	25%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSTPIP1	99.8	99%	95%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN11	101.2	96%	90%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPRC	115.8	93%	86%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 {Hepatitic C virus, susceptibility to}, 609532
PTRF	142.9	99%	98%	Lipodystrophy, congenital generalized, type 4, 613327
RAB27A	178	100%	98%	Griscelli syndrome, type 2, 607624
RAC2	130.5	100%	99%	Neutrophil immunodeficiency syndrome, 608203

RAG1	230.7	100%	100%	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 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAG2	258.8	100%	99%	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RASGRP2	111.2	99%	97%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	110.1	97%	93%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RECQL4	150	98%	97%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RFX5	121.3	98%	97%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	116.6	100%	100%	MHC class II deficiency, complementation group B, 209920
RFXAP	130.5	95%	92%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RHOH	142.5	100%	100%	No OMIM phenotype
RMRP	NC	NC	NC	Anauxetic dysplasia, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	149.3	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	125.1	94%	84%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	207.6	99%	97%	Aicardi-Goutieres syndrome 3, 610329
RNF168	261.8	100%	98%	RIDDLE syndrome, 611943
RNF31	165.8	99%	97%	No OMIM phenotype Autoinflammation, immunodeficiency, amylopectinosis and lymphangiectasia (Boisson (2015) J Exp Med 212,939)
RPSA	89.8	100%	99%	Asplenia, isolated congenital, 271400
RSPH9	150.8	99%	98%	Ciliary dyskinesia, primary, 12, 612650
RTEL1	127.4	99%	96%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
SAMHD1	149.9	99%	98%	Aicardi-Goutieres syndrome 5, 612952

				?Chilblain lupus 2, 614415
SBDS	210.4	99%	99%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SERAC1	125.5	98%	94%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	103.7	98%	94%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SH2D1A	137.5	90%	89%	Lymphoproliferative syndrome, X-linked, 1, 308240
SKIV2L	26.9	80%	57%	Trichohepatoenteric syndrome 2, 614602
SLC29A3	226.4	99%	99%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	146	99%	97%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	230	99%	97%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	141	100%	99%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC39A4	88.4	99%	96%	Acrodermatitis enteropathica, 201100
SLC46A1	105.4	98%	94%	Folate malabsorption, hereditary, 229050
SMARCAL1	148.2	100%	99%	Schimke immunoosseous dysplasia, 242900
SOCS4	303.4	99%	97%	No OMIM phenotype
SP110	147.2	99%	99%	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948
SPINK5	170.1	99%	96%	Atopy, 147050 Netherton syndrome, 256500
STAT1	151.4	97%	95%	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 ncy 31B, mycobacterial and viral infections, autosomal recessive, 613796 Immunodeficiency 31C, autosomal dominant, 614162
STAT2	126.6	100%	99%	Immunodeficiency 44, 616636
STAT3	141	99%	98%	Autoimmune disease, multisystem, infantile-onset, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT4	178.7	99%	97%	{Systemic lupus erythematosus, susceptibility to, 11}, 612253
STAT5B	141.7	99%	96%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STAT6	128.1	100%	99%	No OMIM phenotype {Schistosomiasis infection, association with} (He (2008) Genes Immun 9, 195) {Atopic asthma, association with} (Gao (2004) J Med Genet 41,535)
STIM1	143.1	99%	96%	Immunodeficiency 10, 612783

				Myopathy, tubular aggregate, 1 160565 Stormorken syndrome, 185070
STK4	161.5	99%	98%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	303	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	134.6	99%	98%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TAP1	12.3	51%	15%	Bare lymphocyte syndrome, type I, 604571
TAP2	15	58%	25%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis
TAPBP	22.6	80%	52%	Bare lymphocyte syndrome, type I, 604571
TAZ	126.3	100%	98%	Barth syndrome, 302060
TBX1	86.7	77%	66%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TCIRG1	125.6	95%	88%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	192.4	100%	100%	Transcobalamin II deficiency, 275350
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERT	148.3	96%	91%	{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	177.7	99%	99%	Immunodeficiency 46, 616740
THBD	123	99%	97%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TICAM1	113.2	99%	99%	{Herpes simplex encephalitic, susceptibility to, 6}, 614850
TINF2	196.8	100%	99%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TLR3	189.9	98%	97%	{Herpes simplex encephalitis, susceptibility to, 2} 613002 {HIV1 infection, resistance to}, 609423
TLR4	147.5	99%	98%	Endotoxin hyporesponsiveness {Colorectal cancer, susceptibility to}, 114500

				{Macular degeneration, age-related, 10}, 611488
TMC6	88	99%	96%	Epidermodysplasia verruciformis, 226400
TMC8	119	97%	92%	Epidermodysplasia verruciformis, 226400
TMEM173	102.7	99%	96%	STING-associated vasculopathy, infantile-onset, 615934
TNFRSF11A	144.8	93%	91%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF13B	115	100%	99%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	59.2	93%	75%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	104.1	91%	88%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFRSF4	68.3	98%	91%	?Immunodeficiency 16, 615593
TPP2	143.4	98%	94%	No OMIM phenotype
TRAF3	142.2	99%	98%	{?Herpes simplex encephalitis, susceptibility to, 3}, 614849
TRAF3IP2	135	99%	96%	?Candidiasis, familial, 8, 615527 {Psoriasis susceptibility 13}, 614070
TREX1	272.2	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	111.2	95%	90%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TTC37	139	99%	97%	Trichohepatoenteric syndrome 1, 222470
TTC7A	122.3	99%	97%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	127.5	99%	98%	Immunodeficiency 35, 611521
UNC119	105.1	97%	89%	?Cone-rod dystrophy ?Immunodeficiency 13, 615518
UNC13D	104.1	99%	97%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	71.1	57%	54%	{Herpes simplex encephalitis, susceptibility to, 1}, 610551
UNG	76.6	99%	95%	Immunodeficiency with hyper IgM, type 5, 608106
USB1	144.5	99%	97%	Poikiloderma with neutropenia,604173
VPS13B	156.8	98%	96%	Cohen syndrome, 216550
VPS45	143.4	96%	94%	Neutropenia, severe congenital, 5, autosomal recessive, 615285

WAS	68.7	87%	78%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WIPF1	94	99%	98%	?Wiskott-Aldrich syndrome 2, 614493
WRAP53	164.3	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	126.2	91%	87%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	212.4	99%	99%	Autoimmune disease,multisystem,infantile-onset,2,617006 Immunodeficiency 48,269840
ZBTB24	191.3	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 : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 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