PRIMARY IMMUNODEFICIENCIES GENE PANEL

Gene symbol	Depth (reads)	Coverage (avg %)	OMIM disease	Description
ACP5	96	95	200	Spondyloenchondrodysplasia with immune dysregulation
ACTB	101	17	243310	Baraitser-Winter syndrome 1
ADA	82	90	102700	Adenosine deaminase deficiency partial
AGA	121	94	208400	Aspartylglucosaminuria
AICDA	87	86	605258	Immunodeficiency with hyper-IgM type 2
AIRE	70	89	240300	Autoimmune polyendocrinopathy syndrome type I with or without reversible metaphyseal dysplasia
AK2	107	44	267500	Reticular dysgenesis
ALG13	101	85	300884	Congenital disorder of glycosylation type Is
AP3B1	109	95	608233	Hermansky-Pudlak syndrome 2
ATM	116	93	208900	Ataxia-telangiectasia
BLM	128	89	210900	Bloom syndrome
BLNK	102	91	613502	Agammaglobulinemia 4
BLOC1S6	112	88	614171	Hermansky-pudlak syndrome 9
BTK	86	90	307200	Agammaglobulinemia and isolated hormone deficiency
C1QA	107	83	613652	C1q deficiency
C1QB	106	83	613652	C1q deficiency
C1QC	117	87	613652	C1q deficiency
C1R	100	91	216950	C1r/C1s deficiency, combined
C1S	115	98	613783	C1s deficiency
C2	18	92	217000	C2 deficiency
C3	99	93	613779	C3 deficiency
C4B	23	81	614379	C4B deficiency
C5	106	90	609536	C5 deficiency
C6	123	89	612446	C6 deficiency
C7	96	89	610102	C7 deficiency
C8A	94	88	613790	C8 deficiency type I
C8B	112	82	613789	C8 deficiency type II
C9	117	100	613825	C9 deficiency with dermatomyositis

CARD11	105	88	615206	Persistent polyclonal B-cell lymphocytosis 606445 (3) Immunodeficiency primary autosomal recessive CARD11-related
CARD9	59	97	212050	Candidiasis familial 2 autosomal recessive
CASP10	98	97	603909	Autoimmune lymphoproliferative syndrome type II
CASP8	141	91	114550	Hepatocellular carcinoma somatic
CD19	83	93	613493	Immunodeficiency common variable 3
CD247	107	83	610163	Immunodeficiency due to defect in CD3-zeta
CD27	89	85	615122	Lymphoproliferative syndrome 2
CD3D	107	100	608971	Severe combined immunodeficiency T cell-negative B-cell/natural killer-cell positive
CD3E	124	87	200	-
CD3G	106	94	200	-
CD40	110	87	606843	Immunodeficiency with hyper-IgM type 3
CD40LG	89	95	308230	Immunodeficiency, X-linked, with hyper-IgM
CD46	135	36	612922	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}
CD55	124	86	613793	[Blood group Cromer]
CD59	133	93	612300	CD59 deficiency
CD79A	63	93	613501	Agammaglobulinemia 3
CD79B	128	99	612692	Agammaglobulinemia 6
CD81	70	85	613496	Immunodeficiency common variable 6
CD8A	76	94	608957	CD8 deficiency familial
CEBPE	90	97	245480	Specific granule deficiency
CFB	22	92	612924	{Hemolytic uremic syndrome, atypical, susceptibility to, 4}
CFD	53	92	613912	Complement factor D deficiency
CFH	112	70	609814	Complement factor H deficiency
CFHR1	45	20	235400	{Hemolytic uremic syndrome, atypical, susceptibility to}
CFHR3	104	67	235400	{Hemolytic uremic syndrome, atypical, susceptibility to}
CFHR5	109	80	614809	Nephropathy due to CFHR5 deficiency
CFI	141	92	610984	Complement factor I deficiency
CFP	77	82	312060	Properdin deficiency, X-linked
CHD7	129	92	214800	CHARGE syndrome
CIITA	95	91	209920	Bare lymphocyte syndrome, type II, complementation group A
CIITA	95	91	180300	{Rheumatoid arthritis, susceptibility to}
CLEC4D	99	97	200	-

CLEC7A	106	99	613108	Candidiasis familial 4 autosomal recessive
COLEC11	108	96	265050	3MC syndrome 2
CORO1A	94	94	615401	Immunodeficiency 8
CR2	127	92	614699	Immunodeficiency common variable 7
CREBBP	90	92	180849	Rubinstein-Taybi syndrome
CSF3R	96	90	162830	Neutrophilia hereditary
CTSC	99	96	245010	Haim-Munk syndrome
CXCR4	198	100	200	Myelokathexis
CYBA	43	85	233690	Chronic granulomatous disease autosomal due to deficiency of CYBA
CYBB	80	85	300645	Atypical mycobacteriosis familial X-linked 2
DCLRE1C	117	98	603554	Omenn syndrome
DHFR	57	32	613839	Megaloblastic anemia due to dihydrofolate reductase deficiency
DKC1	84	94	305000	Dyskeratosis congenita X-linked
DNMT3B	93	82	242860	Immunodeficiency-centromeric instability-facial anomalies syndrome 1
DOCK8	97	84	243700	Hyper-IgE recurrent infection syndrome autosomal recessive
ELANE	124	83	162800	Neutropenia cyclic
ELF4	73	94	200	-
EPG5	97	87	242840	Vici syndrome
ERCC2	88	89	610756	Cerebrooculofacioskeletal syndrome 2
ERCC3	134	93	601675	Trichothiodystrophy
F12	107	88	610618	Angioedema hereditary type III
FADD	98	94	613759	Infections recurrent with encephalopathy hepatic dysfunction and cardiovasuclar malformations
FAS	185	88	601859	Autoimmune lymphoproliferative syndrome, type IA
FASLG	91	94	601859	Autoimmune lymphoproliferative syndrome, type IB
FASLG	91	94	211980	{Lung cancer, susceptibility to}
FCGR3A	133	29	200	{Viral infections, recurrent}
FCN3	101	88	613860	Immunodeficiency due to ficolin 3 deficiency
FERMT3	89	94	612840	Leukocyte adhesion deficiency, type III
FOXN1	116	92	601705	T-cell immunodeficiency congenital alopecia and nail dystrophy
FOXP3	69	86	304790	Immunodysregulation polyendocrinopathy and enteropathy X-linked
FPR1	184	100	200	-
G6PC	137	91	232200	Glycogen storage disease Ia
G6PC3	123	86	612541	Dursun syndrome

G6PD	76	94	134700	Favism
GATA2	98	92	614172	Dendritic cell monocyte B lymphocyte and natural killer lymphocyte deficiency
GFI1	67	86	607847	Neutropenia nonimmune chronic idiopathic of adults
GJC2	34	99	608804	Leukodystrophy hypomyelinating 2
GTF2H5	101	37	601675	Trichothiodystrophy complementation group A
HAX1	157	100	610738	Neutropenia severe congenital 3 autosomal recessive
ICOS	133	100	607594	Immunodeficiency common variable 1
IFNGR1	139	97	209950	BCG infection generalized familial
IFNGR2	133	84	209950	{Mycobacterial infection, atypical, familial disseminated}
IGLL1	47	68	613500	Agammaglobulinemia 2
IKBKG	84	79	300291	Ectodermal dysplasia hypohidrotic with immune deficiency
IKZF1	116	92	200	Leukemia
IL10RA	105	89	613148	Inflammatory bowel disease 28 early onset autosomal recessive
IL10RB	114	88	612567	Inflammatory bowel disease 25, early onset, autosomal recessive
IL10RB	114	88	610424	{Hepatitis B virus, susceptibility to}
IL12B	107	80	209950	BCG and salmonella infection, disseminated
IL12B	107	80	600807	{Asthma, susceptibility to}
IL12RB1	74	83	209950	{Mycobacterial and salmonella infections, susceptibility to}
IL17F	101	86	613956	Candidiasis familial 6 autosomal dominant
IL17RA	91	94	613953	Candidiasis familial 5 autosomal recessive
IL1RN	100	91	612852	Interleukin 1 receptor antagonist deficiency
IL2	101	99	200	Severe combined immunodeficiency due to IL2 deficiency
IL21R	115	91	615207	Immunodeficiency primary autosomal recessive IL21R-related
IL2RA	107	89	606367	Interleukin-2 receptor alpha chain deficiency of
IL2RG	80	79	312863	Combined immunodeficiency X-linked moderate
IL36RN	83	79	614204	Psoriasis generalized pustular
IL7R	118	93	608971	Severe combined immunodeficiency T-cell negative B-cell/natural killer cell-positive type
INSR	130	88	610549	Diabetes mellitus insulin-resistant with acanthosis nigricans
IRAK4	105	95	610799	Invasive pneumococcal disease recurrent isolated 1
IRF8	80	88	614893	CD11C+/CD1C+ dendritic cell deficiency dominant
ITCH	116	95	613385	Autoimmune disease syndromic multisystem
ITGB2	85	89	116920	Leukocyte adhesion deficiency
ITK	113	87	613011	Lymphoproliferative syndrome 1

JAK2	112	95	133100	Erythrocytosis somatic
JAK3	97	85	600802	SCID autosomal recessive T-negative/B-positive type
KMT2D	111	96	147920	Kabuki syndrome 1
KRAS	68	74	109800	Bladder cancer somatic
LAMTOR2	90	84	610798	Immunodeficiency due to defect in MAPBP-interacting protein
LCK	88	90	200	SCID due to LCK deficiency
LIG1	90	83	200	-
LIG4	186	100	606593	LIG4 syndrome
LPIN2	89	85	609628	Majeed syndrome
LRBA	110	95	614700	Immunodeficiency common variable 8 with autoimmunity
LRRC8A	127	98	613506	Agammaglobulinemia 5
LYST	124	94	214500	Chediak-Higashi syndrome
MAGT1	86	94	300853	Immunodeficiency X-linked with magnesium defect Epstein-Barr virus infection and neoplasia
MAL	88	83	200	-
MAN2B1	89	85	248500	Mannosidosis alpha- types I and II
MANBA	98	92	248510	Mannosidosis beta
MASP2	114	92	613791	MASP2 deficiency
MBL2	125	88	614372	{Chronic infections, due to MBL deficiency}
MC2R	126	98	202200	Glucocorticoid deficiency due to ACTH unresponsiveness
MCM4	121	80	609981	Natural killer cell and glucocorticoid deficiency with DNA repair defect
MEFV	108	96	134610	Familial Mediterranean fever AD
MPO	86	84	254600	Myeloperoxidase deficiency
MRE11A	99	79	604391	Ataxia-telangiectasia-like disorder
MS4A1	161	94	613495	Immunodeficiency common variable 5
MTHFD1	115	76	200	{Abruptio placentae, susceptibility to}
MTHFD1	115	76	601634	{Spina bifida, folate-sensitive, susceptibility to}
MVK	101	78	260920	Hyper-IgD syndrome
MYD88	148	90	153600	Macroglobulinemia Waldenstrom somatic
NBN	127	94	609135	Aplastic anemia
NBN	127	94	613065	Leukemia, acute lymphoblastic
NBN	127	94	251260	Nijmegen breakage syndrome
NCF2	119	89	233710	Chronic granulomatous disease due to deficiency of NCF-2
NCF4	98	84	613960	Granulomatous disease chronic autosomal recessive cytochrome b-positive type III

NCSTN	105	75	142690	Acne inversa familial 1
NDNL2	106	100	200	-
NFKBIA	115	88	612132	Ectodermal dysplasia anhidrotic with T-cell immunodeficiency
NHEJ1	90	80	611291	Severe combined immunodeficiency with microcephaly growth retardation and sensitivity to ionizing radiation
NHP2	74	37	613987	Dyskeratosis congenita, autosomal recessive 2
NKX2-5	82	100	271400	Asplenia isolated congenital
NLRP12	110	92	611762	Familial cold autoinflammatory syndrome 2
NLRP3	128	96	607115	CINCA syndrome
NOD2	95	94	186580	Blau syndrome
NOP10	141	100	224230	Dyskeratosis congenita, autosomal recessive 1
NRAS	128	94	614470	Autoimmune lymphoproliferative syndrome type IV
ORAI1	82	92	612782	Immune dysfunction with T-cell inactivation due to calcium entry defect 1
PCCA	96	90	606054	Propionicacidemia
PCCB	116	84	606054	Propionicacidemia
PEPD	78	89	170100	Prolidase deficiency
PIGA	110	89	300868	Multiple congenital anomalies-hypotonia-seizures syndrome 2
PIK3CD	96	86	615513	activated PI3K-delta syndrome
PIK3R1	162	93	615214	Agammaglobulinemia 7 autosomal recessive
PLCG2	125	83	614878	Autoinflammation antibody deficiency and immune dysregulation syndrome
PLG	116	75	217090	Conjunctivitis ligneous
PMM2	118	89	212065	Congenital disorder of glycosylation type Ia
PNP	138	97	613179	Immunodeficiency due to purine nucleoside phosphorylase deficiency
PRF1	82	100	603553	Hemophagocytic lymphohistiocytosis familial 2
PRKDC	99	86	200	-
PRPS1	95	64	301835	Arts syndrome
PSENEN	145	77	613736	Acne inversa familial 2
PSMB8	12	80	256040	Autoinflammation lipodystrophy and dermatosis syndrome
PSTPIP1	58	94	604416	Pyogenic sterile arthritis pyoderma gangrenosum and acne
PTPN11	90	25	151100	LEOPARD syndrome 1
PTPRC	110	89	608971	Severe combined immunodeficiency T cell-negative B-cell/natural killer-cell positive
PTRF	135	97	613327	Lipodystrophy congenital generalized type 4
RAB27A	137	91	607624	Griscelli syndrome type 2

RAC2	53	85	608203	Neutrophil immunodeficiency syndrome
RAG1	159	100	609889	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion severe cytomegalovirus infection and autoimmunity
RAG2	201	100	233650	Combined cellular and humoral immune defects with granulomas
RASGRP2	82	82	200	-
RBCK1	85	83	200	-
RECQL4	88	97	218600	Baller-Gerold syndrome
RFX5	128	93	209920	Bare lymphocyte syndrome type II complementation group C
RFXANK	83	54	209920	MHC class II deficiency complementation group B
RFXAP	81	100	209920	Bare lymphocyte syndrome type II complementation group D
RHOH	142	100	200	-
RNASEH2A	103	87	610333	Aicardi-Goutieres syndrome 4
RNASEH2B	94	95	610181	Aicardi-Goutieres syndrome 2
RNASEH2C	134	100	610329	Aicardi-Goutieres syndrome 3
RNF168	192	95	611943	RIDDLE syndrome
RPSA	83	16	604400	Arrhythmogenic right ventricular dysplasia 5
RPSA	83	16	271400	isolated congenital asplenia (ICAS)
RTEL1	80	84	615190	Dyskeratosis congenita autosomal recessive 5
SAMHD1	127	89	612952	Aicardi-Goutieres syndrome 5
SBDS	84	70	260400	Shwachman-Bodian-Diamond syndrome
SERAC1	95	95	614739	3-methylglutaconic aciduria with deafness encephalopathy and Leigh-like syndrome
SERPING1	144	93	106100	Angioedema, hereditary, types I and II
SERPING1	144	93	120790	Complement component 4, partial deficiency of
SH2D1A	74	87	308240	Lymphoproliferative syndrome X-linked
SKIV2L	19	91	614602	Trichohepatoenteric syndrome 2
SLC35A1	106	97	603585	Congenital disorder of glycosylation type IIf
SLC35C1	83	100	266265	Congenital disorder of glycosylation type IIc
SLC37A4	94	91	232220	Glycogen storage disease Ib
SLC39A4	73	90	201100	Acrodermatitis enteropathica
SLC46A1	94	91	229050	Folate malabsorption hereditary
SMARCAL1	137	89	242900	Schimke immunoosseous dysplasia
SP110	95	82	235550	Hepatic venoocclusive disease with immunodeficiency
SPINK5	113	89	147050	Atopy

STAT1	103	87	614162	Candidiasis familial 7
STAT2	137	92	200	-
STAT3	103	87	147060	Hyper-IgE recurrent infection syndrome
STAT4	115	92	612253	{Systemic lupus erythematosus, susceptitbility to, 11}
STAT5B	93	79	245590	Growth hormone insensitivity with immunodeficiency
STIM1	84	91	612783	Immune dysfunction with T-cell inactivation due to calcium entry defect 2
STK4	111	83	614868	T-cell immunodeficiency recurrent infections autoimmunity and cardiac malformations
STX11	133	100	603552	Hemophagocytic lymphohistiocytosis familial 4
STXBP2	81	96	613101	Hemophagocytic lymphohistiocytosis familial 5
TAP1	16	86	604571	Bare lymphocyte syndrome type I
TAP2	13	90	604571	Bare lymphocyte syndrome type I due to TAP2 deficiency
TAPBP	21	88	604571	Bare lymphocyte syndrome type I
TAZ	68	97	302060	Barth syndrome
TBX1	80	86	217095	Conotruncal anomaly face syndrome
TCIRG1	76	91	259700	Osteopetrosis autosomal recessive 1
TCN2	112	85	275350	Transcobalamin II deficiency
TERC	64	100	127550	Dyskeratosis congenita autosomal dominant 1
TERT	104	90	614742	{Bone marrow failure, telomere-related, 1}
TERT	104	90	613989	{Dyskeratosis congenita}
TERT	104	90	601626	{Leukemia, acute myeloid}
THBD	71	100	614486	Thrombophilia due to thrombomodulin defect
TICAM1	71	100	614850	{Encephalopathy, acute, infection-induced, susceptibility to, 6}
TINF2	206	92	613990	Dyskeratosis congenita autosomal dominant 3
TLR3	177	94	613002	{Herpes simplex encephalitis, susceptibility to, 2}
TLR3	177	94	609423	{HIV1 infection, resistance to}
TMC6	68	90	226400	Epidermodysplasia verruciformis
TMC8	92	87	226400	Epidermodysplasia verruciformis
TNFRSF11A	104	95	174810	Osteolysis familial expansile
TNFRSF13B	70	86	240500	Immunodeficiency common variable 2
TNFRSF13C	67	80	613494	Immunodeficiency common variable 4
TNFRSF1A	72	89	142680	Periodic fever familial
TRAF3	122	91	614849	{Herpes simplex encephalitis, susceptibility to, 3}
TREX1	122	100	225750	Aicardi-Goutieres syndrome 1 dominant and recessive

TTC37	115	97	222470	Trichohepatoenteric syndrome 1
TYK2	85	89	611521	Tyrosine kinase 2 deficiency
UNC119	106	92	200	-
UNC13D	60	88	608898	Hemophagocytic lymphohistiocytosis familial 3
UNC93B1	77	88	610551	Herpes simplex encephalitis susceptibility to 1
UNG	100	80	608106	Immunodeficiency with hyper IgM type 5
USB1	53	83	604173	Poikiloderma with neutropenia
VPS13B	117	92	216550	Cohen syndrome
WAS	56	85	300299	Neutropenia severe congenital X-linked
WIPF1	105	85	614493	Wiskott-Aldrich syndrome 2
WRAP53	129	89	613988	Dyskeratosis congenita autosomal recessive 3
XIAP	120	86	300635	Lymphoproliferative syndrome X-linked 2
ZAP70	76	93	269840	Selective T-cell defect
ZBTB24	155	98	614069	Immunodeficiency-centromeric instability-facial anomalies syndrome-2

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Depth describes the average number of reads seen across 50 exomes
Coverage describes the average coverage of a gene across 50 exomes in percentiles
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013

Ad 1. OMIM identifier 200 signifies a gene without a current OMIM association

Ad 2. OMIM phenotype descriptions between {} signify risk factors