Whole Exome Sequencing
Gene package Primary Immunodeficiency Disorders, version 6.1, 31-1-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered 30x
ACD	?Dyskeratosis congenita 6, 616553?Dyskeratosis congenita 7, 616553	609377	139	100	100	100
ACP5	Spondyloenchondrodysplasia with immune dysregulation, 607944	171640	143	100	100	100
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	192	100	100	100
ADA	Adenosine deaminase deficiency, partial, 102700, Somatic mosaicism Severe combined immunodeficiency due to ADA deficiency, 102700, Somatic mosaicism	608958	85	100	100	96
ADA2	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688	607575	96	100	100	99
ADAM17	?Inflammatory skin and bowel disease, neonatal, 1, 614328	603639	67	100	99	94
ADAMTS3	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154	605011	62	100	99	95
ADAR	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400	146920	90	100	100	100
AGA	Aspartylglucosaminuria, 208400	613228	73	100	100	96
AICDA	Immunodeficiency with hyper-IgM, type 2, 605258	605257	134	100	100	95
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300	607358	100	100	100	100

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AK2	Reticular dysgenesis, 267500	103020	68	100	100	97
ALG13	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884	300776	50	100	97	83
AP1S3	{Psoriasis 15, pustular, susceptibility to}, 616106	615781	49	100	100	93
AP3B1	Hermansky-Pudlak syndrome 2, 608233	603401	66	100	98	87
AP3D1	?Hermansky-Pudlak syndrome 10, 617050	607246	103	99	98	97
APOL1	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551	603743	113	100	100	100
ARHGEF1	?Immunodeficiency 62, 618459	601855	97	100	100	98
ARPC1B	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718	604223	107	100	100	100
ATM	Ataxia-telangiectasia, 208900 {Breast cancer, susceptibility to}, 114480 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic	607585	75	100	99	95
ATP6AP1	Immunodeficiency 47, 300972	300197	76	100	99	95
B2M	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600	109700	94	100	100	100
BACH2	Immunodeficiency 60, 618394	605394	118	100	100	100
BCL10	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Male germ cell tumor, somatic}, 273300 {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic}	603517	66	100	100	96
BCL11B	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092	606558	106	100	97	92
BLK	Maturity-onset diabetes of the young, type 11, 613375	191305	113	100	100	100
BLM	Bloom syndrome, 210900	604610	82	100	100	96
BLNK	?Agammaglobulinemia 4, 613502	604515	60	100	98	88
BLOC1S6	?Hermansky-pudlak syndrome 9, 614171	604310	46	100	100	84
BTK	Agammaglobulinemia 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200	300300	50	100	98	83
C1QA	C1q deficiency, 613652	120550	155	100	100	100

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C1QB	C1q deficiency, 613652	120570	121	100	100	100
C1QC	C1q deficiency, 613652	120575	150	100	100	100
C1S	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174	120580	96	100	100	98
C2	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489	613927	106	100	100	100
C3	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378	120700	127	100	100	99
C5	C5 deficiency, 609536 [Eculizumab, poor response to], 615749	120900	72	100	98	94
C6	C6 deficiency, 612446 Combined C6/C7 deficiency	217050	65	100	99	94
C7	C7 deficiency, 610102	217070	79	100	99	96
C8A	C8 deficiency, type I, 613790	120950	69	100	100	98
C8B	C8 deficiency, type II, 613789	120960	100	100	100	98
C8G	No OMIM phenotype	120930	151	100	100	100
C9	C9 deficiency, 613825 {Macular degeneration, age-related, 15, susceptibility to}, 615591	120940	66	100	100	97
CA2	Osteopetrosis 3, with renal tubular acidosis, 259730	611492	99	100	100	100
CARD11	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206 Immunodeficiency 11B with atopic dermatitis, 617638	607210	108	100	100	99
CARD14	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723	607211	112	100	99	99
CARD9	Candidiasis, familial, 2, 212050	607212	122	100	100	99
CARMIL2	Immunodeficiency 58, 618131	610859	125	100	96	94
CASP10	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027	601762	75	100	100	97
CASP8	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 {Breast cancer, protection against}, 114480 Hepatocellular carcinoma, somatic, 114550 {Lung cancer, protection against}, 211980	601763	87	100	100	100

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CAVIN1	Lipodystrophy, congenital generalized, type 4, 613327	603198	172	100	100	100
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510	612753	87	100	99	95
CD19	Immunodeficiency, common variable, 3, 613493	107265	107	100	100	99
CD247	?Immunodeficiency 25, 610163	186780	74	100	100	100
CD27	Lymphoproliferative syndrome 2, 615122	186711	95	100	100	100
CD3D	Immunodeficiency 19, 615617	186790	100	100	100	99
CD3E	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615	186830	113	100	100	100
CD3G	Immunodeficiency 17, CD3 gamma deficient, 615607	186740	76	100	100	99
CD40	Immunodeficiency with hyper-IgM, type 3, 606843	109535	104	100	100	100
CD40LG	Immunodeficiency, with hyper-IgM, 308230	300386	67	100	100	97
CD46	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922	120920	88	100	100	95
CD55	[Blood group Cromer], 613793 Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300	125240	55	91	85	79
CD59	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300	107271	86	100	100	100
CD70	Lymphoproliferative syndrome 3, 618261	602840	100	100	100	100
CD79A	Agammaglobulinemia 3, 613501	112205	83	100	91	85
CD79B	Agammaglobulinemia 6, 612692	147245	146	100	100	100
CD81	Immunodeficiency, common variable, 6, 613496	186845	146	97	90	90
CD8A	CD8 deficiency, familial, 608957	186910	100	100	100	97
CDCA7	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910	609937	61	100	99	92
CDKN2B	No OMIM phenotype	600431	105	100	100	100
CEBPE	Specific granule deficiency, 245480	600749	57	100	100	99
CFB	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489	138470	106	100	100	100
CFD	Complement factor D deficiency, 613912	134350	99	100	98	90
CFH	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698	134370	78	100	100	96
CFHR1	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075	134371	65	93	93	83

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CFHR3	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075	605336	55	100	93	87
CFHR5	Nephropathy due to CFHR5 deficiency, 614809	608593	75	100	100	97
CFI	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439	217030	67	100	99	93
CFP	Properdin deficiency, 312060	300383	68	100	95	86
CFTR	{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 Congenital bilateral absence of vas deferens, 277180 Cystic fibrosis, 219700 {Hypertrypsinemia, neonatal} {Pancreatitis, hereditary}, 167800 Sweat chloride elevation without CF	602421	93	100	100	97
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	87	100	100	97
CIITA	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300	600005	117	100	100	99
CLCN7	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis 2, 166600 Osteopetrosis 4, 611490	602727	128	100	100	99
CLEC4D	No OMIM phenotype	609964	52	100	100	93
CLEC7A	{Aspergillosis, susceptibility to}, 614079 Candidiasis, familial, 4, 613108	606264	64	100	100	91
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271	616254	114	100	100	100
COG6	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328	606977	65	100	99	90
COPA	{Autoimmune interstitial lung, joint, and kidney disease}, 616414	601924	75	100	100	99
CORO1A	Immunodeficiency 8, 615401	605000	122	92	92	91
CR2	Immunodeficiency, common variable, 7, 614699 {Systemic lupus erythematosus, susceptibility to, 9}, 610927	120650	63	100	100	97
CREBBP	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849	600140	85	100	99	94
CSF2RA	Surfactant metabolism dysfunction, pulmonary, 4, 300770	306250	0	0	0	0
CSF2RB	Surfactant metabolism dysfunction, pulmonary, 5, 614370	138981	132	100	100	100

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CSF3R	Neutropenia, severe congenital, 7, 617014	138971	98	100	100	100
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, 612199	613129	100	100	100	100
CTLA4	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	123890	90	100	100	100
CTPS1	Immunodeficiency 24, 615897	123860	70	100	100	97
CTSC	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650	602365	75	100	100	97
CXCR4	Myelokathexis, isolated WHIM syndrome, 193670	162643	82	83	83	82
CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690	608508	106	94	81	66
CYBB	Chronic granulomatous disease, 306400 Immunodeficiency 34, mycobacteriosis, 300645	300481	49	100	98	91
CYBC1	No OMIM phenotype	618334	No coverage data			
DCLRE1B	No OMIM phenotype	609683	95	100	100	98
DCLRE1C	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450	605988	64	100	100	92
DDX58	Singleton-Merten syndrome 2, 616298	609631	68	100	100	96
DGAT1	?Diarrhea 7, protein-losing enteropathy type, 615863	604900	116	96	91	85
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839	126060	76	100	100	98
DKC1	Dyskeratosis congenita, 305000	300126	53	100	98	89
DNAJC21	Bone marrow failure syndrome 3, 617052	617048	62	100	96	89
DNASE1	{Systemic lupus erythematosus, susceptibility to}, 152700	125505	132	100	100	100
DNASE1L3	Systemic lupus erythematosus 16, 614420	602244	67	100	100	98
DNASE2	No OMIM phenotype	126350	95	100	100	100
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860	602900	115	100	100	99
DOCK2	Immunodeficiency 40, 616433	603122	82	100	100	97
DOCK8	Hyper-IgE recurrent infection syndrome, 243700	611432	74	100	99	94
EFL1	Shwachman-Diamond syndrome 2, 617941	617538	60	99	97	92
ELANE	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, 202700	130130	127	100	100	100

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ELF4	No OMIM phenotype	300775	67	100	100	100
EPG5	Vici syndrome, 242840	615068	69	100	100	97
ERCC2	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730	126340	97	100	99	98
ERCC3	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651	133510	82	100	100	99
ERCC6L2	Bone marrow failure syndrome 2, 615715	615667	57	100	98	93
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425	605744	150	100	100	100
F12	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000	610619	135	100	100	100
FAAP24	No OMIM phenotype	610884	97	100	100	100
FADD	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759	602457	143	100	100	100
FAS	Autoimmune lymphoproliferative syndrome, type IA, 601859 {Autoimmune lymphoproliferative syndrome}, 601859 Squamous cell carcinoma, burn scar-related, somatic	134637	264	100	100	96
FASLG	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980	134638	65	100	100	100
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546	612411	90	100	100	99
FCGR1A	[IgG receptor I, phagocytic, familial deficiency of]	146760	83	97	97	93
FCGR2A	{Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162 {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700	146790	150	100	100	100
FCGR2B	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700	604590	99	84	82	78
FCGR3A	Immunodeficiency 20, 615707	146740	244	100	100	98
FCGR3B	Neutropenia, alloimmune neonatal	610665	210	100	100	100
FCN3	Immunodeficiency due to ficolin 3 deficiency, 613860	604973	103	100	100	97
FERMT3	Leukocyte adhesion deficiency, type III, 612840	607901	114	100	100	97
FNIP1	No OMIM phenotype	610594	59	100	99	94
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705	600838	136	100	100	100
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, 304790	300292	69	100	99	88
FPR1	No OMIM phenotype	136537	129	100	100	100

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G6PC	Glycogen storage disease Ia, 232200	613742	113	100	100	100
G6PC3	Dursun syndrome, 612541 Neutropenia, severe congenital 4, 612541	611045	103	100	100	100
G6PD	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162	305900	96	100	100	100
GATA2	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286	137295	100	100	100	100
GFI1	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 ?Neutropenia, severe congenital 2, 613107	600871	136	100	100	100
GINS1	Immunodeficiency 55, 617827	610608	65	100	100	91
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, 613206	608803	85	96	86	76
GRHL2	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029	608576	81	100	100	99
GTF2H5	Trichothiodystrophy 3, photosensitive, 616395	608780	53	100	100	100
HAX1	Neutropenia, severe congenital 3, 610738	605998	99	100	100	99
HELLS	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911	603946	61	100	96	84
HMOX1	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963	141250	112	100	100	99
HYOU1	?Immunodeficiency 59 and hypoglycemia, 233600	601746	112	100	100	98
ICOS	Immunodeficiency, common variable, 1, 607594	604558	57	100	100	95
ICOSLG	No OMIM phenotype	605717	93	100	97	95
IFIH1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250	606951	86	100	100	96
IFNAR2	{Hepatitis B virus, susceptibility to}, 610424 ?Immunodeficiency 45, 616669	602376	93	100	100	97

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IFNGR1	{H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424 Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948	107470	68	100	100	95
IFNGR2	Immunodeficiency 28, mycobacteriosis, 614889	147569	64	97	92	90
IGLL1	Agammaglobulinemia 2, 613500	146770	132	100	100	100
IKBKB	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592	603258	86	100	100	97
IKBKG	Ectodermal dysplasia and immunodeficiency 1, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640	300248	24	38	33	31
IKZF1	Immunodeficiency, common variable, 13, 616873	603023	166	100	100	100
IL10	{Graft-versus-host disease, protection against}, 614395 {HIV-1, susceptibility to}, 609423 {Rheumatoid arthritis, progression of}, 180300	124092	120	100	100	99
IL10RA	Inflammatory bowel disease 28, early onset, 613148	146933	125	100	100	99
IL10RB	{Hepatitis B virus, susceptibility to}, 610424 Inflammatory bowel disease 25, early onset, 612567	123889	69	100	100	97
IL12B	Immunodeficiency 29, mycobacteriosis, 614890	161561	77	100	100	98
IL12RB1	Immunodeficiency 30, 614891	601604	116	100	100	100
IL17F	?Candidiasis, familial, 6, 613956	606496	79	100	100	100
IL17RA	Immunodeficiency 51, 613953	605461	125	100	100	100
IL17RC	Candidiasis, familial, 9, 616445	610925	109	100	100	99
IL1RN	{Gastric cancer risk after H. pylori infection}, 137215 Interleukin 1 receptor antagonist deficiency, 612852 {Microvascular complications of diabetes 4}, 612628	147679	76	100	100	90
IL2	No OMIM phenotype	147680	42	100	88	77
IL21	?Immunodeficiency, common variable, 11, 615767	605384	69	100	99	89

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IL21R	[IgE, elevated level of], 147050 Immunodeficiency 56, 615207	605383	113	100	100	100
IL2RA	{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367	147730	96	100	100	100
IL2RB	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495	146710	84	100	100	99
IL2RG	Combined immunodeficiency, moderate, 312863 Severe combined immunodeficiency, 300400	308380	54	100	99	91
IL36RN	Psoriasis 14, pustular, 614204	605507	82	100	100	100
IL6R	[Interleukin 6, serum level of, QTL], 614752 [Interleukin-6 receptor, soluble, serum level of, QTL], 614689	147880	132	100	100	99
IL6ST	Hyper-IgE recurrent infection syndrome 4, 618523	600694	64	100	96	83
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971	146661	60	100	100	99
INO80	No OMIM phenotype	610169	73	100	99	92
INSR	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190	147670	115	100	98	95
IRAK1	No OMIM phenotype	300283	62	100	97	86
IRAK4	IRAK4 deficiency, 607676 Invasive pneumococcal disease, recurrent isolated, 1, 610799	606883	72	100	99	91
IRF2BP2	?Immunodeficiency, common variable, 14, 617765	615332	82	100	100	91
IRF3	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532	603734	132	100	100	97
IRF7	?Immunodeficiency 39, 616345	605047	128	100	99	98
IRF8	Immunodeficiency 32A, mycobacteriosis, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, 226990	601565	136	100	100	99
IRF9	No OMIM phenotype	147574	123	100	100	99
ISG15	Immunodeficiency 38, 616126	147571	144	100	100	100
ITCH	Autoimmune disease, multisystem, with facial dysmorphism, 613385	606409	61	96	95	90
ITGB2	Leukocyte adhesion deficiency, 116920	600065	140	100	100	100
ITK	Lymphoproliferative syndrome 1, 613011	186973	76	100	100	95
JAGN1	Neutropenia, severe congenital, 6, 616022	616012	91	100	100	100
JAK1	No OMIM phenotype	147795	71	100	98	93

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JAK2	{Budd-Chiari syndrome, somatic}, 600880 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521	147796	63	100	99	93
JAK3	SCID, T-negative/B-positive type, 600802	600173	104	100	100	99
KDM6A	Kabuki syndrome 2, 300867	300128	52	100	95	80
KMT2A	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130	159555	71	100	100	98
KMT2D	Kabuki syndrome 1, 147920	602113	114	100	100	99
LACC1	No OMIM phenotype	613409	62	100	98	93
LAMTOR2	Immunodeficiency due to defect in MAPBP-interacting protein, 610798	610389	108	100	100	100
LAT	Immunodeficiency 52, 617514	602354	91	100	100	97
LCK	?Immunodeficiency 22, 615758	153390	160	100	100	100
LIG1	No OMIM phenotype	126391	94	100	100	99
LIG4	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500	601837	71	100	100	100
LPIN2	Majeed syndrome, 609628	605519	77	100	100	98
LRBA	Immunodeficiency, common variable, 8, with autoimmunity, 614700	606453	65	100	99	94
LRRC8A	?Agammaglobulinemia 5, 613506	608360	174	100	100	100
LTBP3	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809	602090	130	100	99	96
LYST	Chediak-Higashi syndrome, 214500	606897	72	100	99	95
MAGT1	Immunodeficiency, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853	300715	47	100	99	85
MAL	No OMIM phenotype	188860	102	100	100	98
MALT1	Immunodeficiency 12, 615468	604860	68	97	93	89
MAN2B1	Mannosidosis, alpha-, types I and II, 248500	609458	115	100	100	100
MANBA	Mannosidosis, beta, 248510	609489	85	100	100	95
MAP3K14	No OMIM phenotype	604655	113	100	100	100
MASP2	MASP2 deficiency, 613791	605102	85	100	100	97
MBL2	{Chronic infections, due to MBL deficiency}, 614372	154545	122	100	100	100
MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200	607397	102	100	100	100
MCM4	Immunodeficiency 54, 609981	602638	88	100	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered 30x
MEFV	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100	608107	105	100	100	100
MOGS	Congenital disorder of glycosylation, type IIb, 606056	601336	130	100	100	100
MRE11	Ataxia-telangiectasia-like disorder 1, 604391	600814	55	100	97	83
MRTFA	Megakaryoblastic leukemia, acute	606078	94	98	97	92
MS4A1	Immunodeficiency, common variable, 5, 613495	112210	80	100	100	98
MSN	Immunodeficiency 50, 300988	309845	51	99	92	78
MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	172460	76	100	100	96
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	100	100	100	100
MYD88	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260	602170	157	100	100	100
MYSM1	Bone marrow failure syndrome 4, 618116	612176	55	100	98	90
NBAS	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800	608025	66	100	99	94
NBN	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260	602667	68	100	99	88
NCF1	Chronic granulomatous disease due to deficiency of NCF-1, 233700	608512	108	68	65	61
NCF2	Chronic granulomatous disease due to deficiency of NCF-2, 233710	608515	94	100	100	98
NCF4	?Granulomatous disease, chronic, cytochrome b-positive, type III, 613960	601488	121	100	100	100
NCSTN	Acne inversa, familial, 1, 142690	605254	91	100	100	98
NFAT5	No OMIM phenotype	604708	64	100	99	96
NFKB1	Immunodeficiency, common variable, 12, 616576	164011	56	100	98	89
NFKB2	Immunodeficiency, common variable, 10, 615577	164012	143	100	100	96
NFKBIA	Ectodermal dysplasia and immunodeficiency 2, 612132	164008	131	100	100	100
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291	611290	55	100	100	96
NHP2	Dyskeratosis congenita 2, 613987	606470	122	100	100	100
NLRC4	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115	606831	83	100	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered 30x
NLRP1	Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579	606636	103	100	100	100
NLRP12	Familial cold autoinflammatory syndrome 2, 611762	609648	135	100	100	100
NLRP3	CINCA syndrome, 607115 Deafness 34, with or without inflammation, 617772 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Muckle-Wells syndrome, 191900	606416	125	100	100	100
NOD2	Blau syndrome, 186580 {Inflammatory bowel disease 1, Crohn disease}, 266600 {Psoriatic arthritis, susceptibility to}, 607507 {Yao syndrome}, 617321	605956	113	100	100	99
NOP10	Dyskeratosis congenita 1, 224230	606471	147	100	100	100
NRAS	Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470	164790	63	100	100	98
NSMCE3	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241	608243	161	100	100	100
ORAI1	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883	610277	175	99	99	97
OSTM1	Osteopetrosis 5, 259720	607649	79	100	100	97
OTULIN	Autoinflammation, panniculitis, and dermatosis syndrome, 617099	615712	70	97	91	82
PARN	Dyskeratosis congenita 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371	604212	59	100	99	92
PAX5	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545	167414	106	100	100	95
PBX1	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641	176310	72	100	100	98
PCCA	Propionicacidemia, 606054	232000	68	100	100	95
PCCB	Propionicacidemia, 606054	232050	72	100	98	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered 30x
PEPD	Prolidase deficiency, 170100	613230	100	100	100	98
PGM3	Immunodeficiency 23, 615816	172100	88	100	100	98
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818	311770	67	100	100	97
PIK3CD	Immunodeficiency 14, 615513	602839	137	100	99	98
PIK3R1	?Agammaglobulinemia 7, 615214 Immunodeficiency 36, 616005 SHORT syndrome, 269880	171833	80	100	100	97
PLCG2	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468	600220	107	100	100	99
PLEKHM1	Osteopetrosis 3, 618107 ?Osteopetrosis 6, 611497	611466	174	100	100	99
PLG	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090	173350	81	100	100	98
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	72	100	100	97
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	164050	70	100	100	97
POLA1	Pigmentary disorder, reticulate, with systemic manifestations, 301220 Van Esch-O'Driscoll syndrome, 301030	312040	44	99	92	77
POLE2	No OMIM phenotype	602670	40	99	92	69
POMP	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 Proteasome-associated autoinflammatory syndrome 2, 618048	613386	45	100	100	87
POT1	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848	606478	71	100	99	94
PRF1	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027	170280	120	100	100	100
PRKCD	Autoimmune lymphoproliferative syndrome, type III, 615559	176977	115	100	100	100
PRKDC	Immunodeficiency 26, with or without neurologic abnormalities, 615966	600899	73	100	99	94
PRPS1	Arts syndrome, 301835 Charcot-Marie-Tooth disease recessive, 5, 311070 Deafness 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661	311850	50	100	99	90
PSENEN	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736	607632	88	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered 30x
PSMA3	No OMIM phenotype	176843	53	100	99	87
PSMB4	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591	602177	73	100	100	100
PSMB8	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040	177046	124	100	100	100
PSMB9	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591	177045	59	100	90	90
PSMG2	No OMIM phenotype	609702	58	100	99	86
PSTPIP1	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416	606347	109	100	100	99
PTPN22	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700	600716	54	100	99	92
PTPRC	{Hepatitis C virus, susceptibility to}, 609532 Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971	151460	73	100	99	95
RAB27A	Griscelli syndrome, type 2, 607624	603868	54	100	100	91
RAC2	Neutrophil immunodeficiency syndrome, 608203	602049	99	100	100	100
RAG1	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	179615	101	100	100	100
RAG2	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457	179616	71	100	100	100
RANBP2	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033	601181	112	100	100	99
RASGRP1	Immunodeficiency 64, 618534	603962	70	100	100	98
RASGRP2	?Bleeding disorder, platelet-type, 18, 615888	605577	93	100	100	100
RBCK1	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895	610924	135	100	100	99
RECQL4	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400	603780	148	100	100	99
RELB	?Immunodeficiency 53, 617585	604758	88	100	98	93
RFX5	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920	601863	100	100	100	98
RFXANK	MHC class II deficiency, complementation group B, 209920	603200	118	100	100	100
RFXAP	Bare lymphocyte syndrome, type II, complementation group D, 209920	601861	148	100	100	100
RHOH	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307	602037	101	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered 30x
RIPK1	Immunodeficiency 57, 618108	603453	54	100	97	91
RMRP	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460	157660	No coverage data			
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	109	100	100	100
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	62	100	98	88
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	323	100	100	100
RNF168	RIDDLE syndrome, 611943	612688	93	100	100	99
RNF31	No OMIM phenotype	612487	121	100	100	99
RNU4ATAC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651	601428	No coverage data			
RORC	Immunodeficiency 42, 616622	602943	97	100	100	99
RPSA	Asplenia, isolated congenital, 271400	150370	84	100	100	100
RSPH9	Ciliary dyskinesia, primary, 12, 612650	612648	125	100	100	100
RTEL1	Dyskeratosis congenita 4, 615190 Dyskeratosis congenita 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373	608833	131	100	100	99
SAMD9	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455	610456	68	100	100	100
SAMD9L	Ataxia-pancytopenia syndrome, 159550	611170	63	100	100	99
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	64	100	99	88
SBDS	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400	607444	79	100	100	99
SEMA3E	?CHARGE syndrome, 214800	608166	64	100	100	94
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	65	100	99	86
SERPING1	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790	606860	112	100	100	99
SH2B3	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950	605093	115	100	100	100
SH2D1A	Lymphoproliferative syndrome, 1, 308240	300490	56	100	98	83
SH3BP2	Cherubism, 118400	602104	127	91	91	91
SKIV2L	Trichohepatoenteric syndrome 2, 614602	600478	124	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered 30x
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome, 602782	612373	152	100	99	99
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585	605634	66	100	100	97
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	146	100	100	100
SLC37A4	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240	602671	96	100	100	95
SLC39A4	Acrodermatitis enteropathica, 201100	607059	125	100	100	100
SLC46A1	Folate malabsorption, hereditary, 229050	611672	121	100	100	98
SMARCAL1	Schimke immunoosseous dysplasia, 242900	606622	88	100	100	98
SMARCD2	Specific granule deficiency 2, 617475	601736	77	87	86	84
SNX10	Osteopetrosis 8, 615085	614780	77	100	100	98
SOCS4	No OMIM phenotype	616337	78	100	100	100
SP110	Hepatic venoocclusive disease with immunodeficiency, 235550 {Mycobacterium tuberculosis, susceptibility to}, 607948	604457	61	100	100	96
SPINK5	Netherton syndrome, 256500	605010	66	100	99	95
SPPL2A	No OMIM phenotype	608238	32	92	71	46
SRP54	No OMIM phenotype	604857	56	100	97	89
STAT1	Immunodeficiency 31A, mycobacteriosis, 614892 Immunodeficiency 31B, mycobacterial and viral infections, 613796 Immunodeficiency 31C, 614162	600555	64	100	99	94
STAT2	Immunodeficiency 44, 616636	600556	87	100	100	97
STAT3	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060	102582	90	100	100	99
STAT4	{Systemic lupus erythematosus, susceptibility to, 11}, 612253	600558	63	100	99	93
STAT5B	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578	604260	107	100	100	97
STAT6	No OMIM phenotype	601512	95	100	100	98
STIM1	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070	605921	95	100	100	99
STING1	STING-associated vasculopathy, infantile-onset, 615934	612374	122	100	100	100
STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868	604965	82	100	100	99
STN1	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341	613128	62	100	97	90
STX11	Hemophagocytic lymphohistiocytosis, familial, 4, 603552	605014	308	100	100	100
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5, 613101	601717	121	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered 30x
TAP1	Bare lymphocyte syndrome, type I, 604571	170260	125	100	100	100
TAP2	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571	170261	106	100	100	100
TAPBP	Bare lymphocyte syndrome, type I, 604571	601962	106	100	100	100
TAZ	Barth syndrome, 302060	300394	97	100	98	90
TBX1	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430	602054	94	94	85	80
TCF3	Agammaglobulinemia 8, 616941	147141	81	100	100	96
TCIRG1	Osteopetrosis 1, 259700	604592	117	100	100	97
TCN2	Transcobalamin II deficiency, 275350	613441	118	100	100	100
TERC	{Aplastic anemia}, 614743 Dyskeratosis congenita 1, 127550 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743	602322	No coverage data			
TERT	{Dyskeratosis congenita 2}, 613989 {Dyskeratosis congenita 4}, 613989 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742	187270	136	100	100	99
TFRC	Immunodeficiency 46, 616740	190010	58	100	99	93
THBD	{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 Thrombophilia due to thrombomodulin defect, 614486	188040	195	100	100	100
TICAM1	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850	607601	127	100	100	98
TINF2	Dyskeratosis congenita 3, 613990 Revesz syndrome, 268130	604319	155	100	100	100
TIRAP	{Bacteremia, protection against}, 614382 {Malaria, protection against}, 611162 {Pneumococcal disease, invasive, protection against}, 610799 {Tuberculosis, protection against}, 607948	606252	158	100	100	100
TLR3	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002 {HIV1 infection, resistance to}, 609423	603029	66	100	100	99
TLR4	No OMIM phenotype	603030	61	100	100	99
TMC6	Epidermodysplasia verruciformis, 226400	605828	86	100	100	97
TMC8	Epidermodysplasia verruciformis 2, 618231	605829	118	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered 30x
TNFAIP3	Autoinflammatory syndrome, familial, Behcet-like, 616744	191163	106	100	99	96
TNFRSF11A	Osteolysis, familial expansile, 174810 Osteopetrosis 7, 612301 {Paget disease of bone 2, early-onset}, 602080	603499	97	95	95	95
TNFRSF13B	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529	604907	106	100	100	100
TNFRSF13C	Immunodeficiency, common variable, 4, 613494	606269	78	100	91	73
TNFRSF1A	{Multiple sclerosis, susceptibility to, 5}, 614810 Periodic fever, familial, 142680	191190	97	100	98	94
TNFRSF4	?Immunodeficiency 16, 615593	600315	61	99	89	82
TNFSF11	Osteopetrosis 2, 259710	602642	56	100	100	94
TNFSF12	No OMIM phenotype	602695	68	100	97	92
TOM1	No OMIM phenotype	604700	115	100	100	100
TPP2	No OMIM phenotype	190470	58	100	99	91
TRAF3	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849	601896	102	100	100	100
TRAF3IP2	?Candidiasis, familial, 8, 615527 {Psoriasis susceptibility 13}, 614070	607043	94	100	100	99
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 {Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315	606609	234	100	100	100
TRNT1	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084	612907	68	100	99	92
TTC37	Trichohepatoenteric syndrome 1, 222470	614589	58	100	99	93
TTC7A	Gastrointestinal defects and immunodeficiency syndrome, 243150	609332	110	100	100	99
TYK2	Immunodeficiency 35, 611521	176941	128	100	100	99
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3, 608898	608897	100	100	100	100
UNC93B1	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551	608204	69	96	87	76
UNG	Immunodeficiency with hyper IgM, type 5, 608106	191525	104	100	100	100
USB1	Poikiloderma with neutropenia, 604173	613276	93	100	100	99
USP18	Pseudo-TORCH syndrome 2, 617397	607057	117	95	95	93
VAV1	No OMIM phenotype	164875	98	100	100	95
VPS13B	Cohen syndrome, 216550	607817	75	100	99	96
VPS45	Neutropenia, severe congenital, 5, 615285	610035	74	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered 30x
WAS	Neutropenia, severe congenital, 300299 Thrombocytopenia, 313900 Thrombocytopenia, intermittent, 313900 Wiskott-Aldrich syndrome, 301000	300392	66	97	84	76
WDR1	No OMIM phenotype	604734	77	100	99	94
WIPF1	?Wiskott-Aldrich syndrome 2, 614493	602357	73	100	100	96
WRAP53	Dyskeratosis congenita 3, 613988	612661	148	100	100	100
XIAP	Lymphoproliferative syndrome, 2, 300635	300079	47	100	95	80
ZAP70	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840	176947	151	100	100	100
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069	614064	73	100	100	100

- Gene symbols according HGCN
- OMIM release used: 8-9-2019
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 100 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x or 30x