PRIMARY IMMUNODEFICIENCY GENE PANEL DG 3.00 (436 genes)

Releasedate: 02-12-2020

Gene	Agilent V5 covered > 10x	Agilent V5 covered > 20x	TWIST covered > 10x	TWIST covered 20x	Associated Phenotype description and OMIM disease ID
ACD	100	99,9	100	100	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACP5	99,8	98,3	100	100	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	99,7	96,1	100	100	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADA	100	99,7	100	100	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	100	99	100	100	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 ?Sneddon syndrome, 182410
ADAM17	99,9	99	100	100	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAR	100	99,8	100	100	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
AGA	100	100	100	100	Aspartylglucosaminuria, 208400
AICDA	100	100	100	100	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	100	99,8	100	100	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	98,9	94,9	100	99,9	Reticular dysgenesis, 267500
ALG13	98,4	92,6	100	99,6	Developmental and epileptic encephalopathy 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
ALPI	100	99,5	100	100	No OMIM disease ID
AP1S3	90,4	90,1	90,5	90,5	{Psoriasis 15, pustular, susceptibility to}, 616106
AP3B1	99,2	95,8	100	100	Hermansky-Pudlak syndrome 2, 608233
AP3D1	99,8	98,6	100	100	?Hermansky-Pudlak syndrome 10, 617050
APOL1	100	100	100	100	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551

ARHGEF1	99,9	98,4	100	100	?Immunodeficiency 62, 618459
ARPC1B	100	100	100	100	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ATM	99,8	98,1	100	100	Ataxia-telangiectasia, 208900 {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell, somatic, 0 Lymphoma, B-cell non-Hodgkin, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0
ATP6AP1	98,2	92,1	100	100	Immunodeficiency 47, 300972
B2M	100	100	100	100	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
BACH2	100	100	100	100	Immunodeficiency 60, 618394
BCL10	100	100	100	100	?Immunodeficiency 37, 616098 {Male germ cell tumor, somatic}, 273300 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic}, 0
BCL11B	99,1	95,6	98,8	97,3	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BLK	100	100	100	100	Maturity-onset diabetes of the young, type 11, 613375
BLM	99,8	98,3	100	100	Bloom syndrome, 210900
BLNK	97,1	95,5	100	100	?Agammaglobulinemia 4, 613502
BLOC1S6	99,9	97,8	100	100	?Hermansky-pudlak syndrome 9, 614171
BTK	100	99,9	100	99,9	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
C1QA	100	100	100	100	C1q deficiency, 613652
C1QB	100	100	100	100	C1q deficiency, 613652
C1QC	100	99,2	100	100	C1q deficiency, 613652
C1R	100	100	99	96,9	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	99,9	99	99,5	97,7	Ehlers-Danlos syndrome, periodontal type, 2, 617174 C1s deficiency, 613783

C2	100	100	100	100	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C3	99,9	99,2	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 C3 deficiency, 613779 {Macular degeneration, age-related, 9}, 611378
C5	99,9	98,5	100	100	[Eculizumab, poor response to], 615749 C5 deficiency, 609536
C6	100	99,7	100	100	C6 deficiency, 612446 Combined C6/C7 deficiency, 0
C7	100	98,9	100	100	C7 deficiency, 610102
C8A	100	99,6	100	100	C8 deficiency, type I, 613790
C8B	100	99,2	100	100	C8 deficiency, type II, 613789
C8G	100	100	100	100	No OMIM disease ID
C9	99,9	99,5	100	100	{Macular degeneration, age-related, 15, susceptibility to}, 615591 C9 deficiency, 613825
CA2	100	100	100	100	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CARD11	100	99,9	100	100	Immunodeficiency 11B with atopic dermatitis, 617638 B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206
CARD14	100	98,9	100	100	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	99,9	98,4	100	100	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	96,3	94,5	99,7	98,2	Immunodeficiency 58, 618131
CASP10	99,5	97,3	100	100	Lymphoma, non-Hodgkin, somatic, 605027 Gastric cancer, somatic, 613659 Autoimmune lymphoproliferative syndrome, type II, 603909
CASP8	95,6	95,4	95,6	95,6	{Lung cancer, protection against}, 211980 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480
CAVIN1	100	100	100	100	Lipodystrophy, congenital generalized, type 4, 613327
CCBE1	99,8	98,8	100	100	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD19	100	100	100	100	Immunodeficiency, common variable, 3, 613493

CD247	100	100	100	100	?Immunodeficiency 25, 610163
CD27	99,9	96,9	100	100	Lymphoproliferative syndrome 2, 615122
CD3D	100	100	100	100	Immunodeficiency 19, 615617
CD3E	100	99,5	100	100	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	100	100	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	100	100	100	100	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	97,3	88,1	100	100	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	99,9	99,4	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD55	92,9	85,4	94,1	92	[Blood group Cromer], 613793 Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	80	71,6	64,5	64,5	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	99,8	97,7	100	100	Lymphoproliferative syndrome 3, 618261
CD79A	100	100	100	100	Agammaglobulinemia 3, 613501
CD79B	100	100	100	100	Agammaglobulinemia 6, 612692
CD81	100	99,9	100	100	Immunodeficiency, common variable, 6, 613496
CD8A	100	99,8	100	100	CD8 deficiency, familial, 608957
CDCA7	100	99,6	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDKN2B	100	99,9	100	100	No OMIM disease ID
CEBPE	100	100	100	100	Specific granule deficiency, 245480
CFB	100	100	100	100	?Complement factor B deficiency, 615561 {Macular degeneration, age-related, 14, reduced risk of}, 615489 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924
CFD	89,3	83,7	100	100	Complement factor D deficiency, 613912
CFH	99,9	99	100	99,9	Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 Basal laminar drusen, 126700 {Macular degeneration, age-related, 4}, 610698

CFHR1	96,4	94,9	95,4	93,8	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR2	76	74,3	76,5	76,1	No OMIM disease ID
CFHR3	94	92,2	96	95,2	{Macular degeneration, age-related, reduced risk of}, 603075 {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR4	100	99,9	100	100	No OMIM disease ID
CFHR5	99,6	98,4	100	100	Nephropathy due to CFHR5 deficiency, 614809
CFI	99,2	96,8	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 Complement factor I deficiency, 610984 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFP	100	99	100	100	Properdin deficiency, X-linked, 312060
CFTR	99,6	97,9	100	100	{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 {Pancreatitis, hereditary}, 167800 Sweat chloride elevation without CF, 0 {Hypertrypsinemia, neonatal}, 0
CHD7	100	99,5	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CIB1	97,3	93,6	100	100	Epidermodysplasia verruciformis 3, 618267
CIITA	100	99,5	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CLCN7	99,7	98,4	100	100	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600 Hypopigmentation, organomegaly, and delayed myelination and development, 618541
CLEC4D	100	99,8	100	100	No OMIM disease ID
CLEC7A	100	100	100	100	{Aspergillosis, susceptibility to}, 614079 Candidiasis, familial, 4, autosomal recessive, 613108
CLPB	94,9	94,9	100	100	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COLEC11	100	100	100	100	3MC syndrome 2, 265050
COPA	100	99,2	100	100	{Autoimmune interstitial lung, joint, and kidney disease}, 616414
CORO1A	100	98,6	100	100	Immunodeficiency 8, 615401

CR2	100	99,8	100	100	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 Immunodeficiency, common variable, 7, 614699
CREBBP	99,7	98,5	100	100	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CSF2RA	89,9	87,5	95,6	92,1	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	100	99	100	100	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	99,6	98,2	100	100	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	100	99,6	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Celiac disease, susceptibility to, 3}, 609755 Autoimmune lymphoproliferative syndrome, type V, 616100 {Hashimoto thyroiditis}, 140300
CTPS1	93	93	93	93	Immunodeficiency 24, 615897
CTSC	100	100	100	100	Periodontitis 1, juvenile, 170650 Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010
CXCR4	100	100	100	100	WHIM syndrome, 193670 Myelokathexis, isolated, 0
CYBA	95	82,4	100	100	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBB	99,9	99,3	100	100	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400
CYBC1	99,6	97	100	100	Chronic granulomatous disease 5, autosomal recessive, 618935
DBF4	96,6	89,6	100	100	No OMIM disease ID
DBR1	100	99,3	100	100	No OMIM disease ID
DCLRE1C	100	99,4	100	100	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDX58	99,9	99	100	100	Singleton-Merten syndrome 2, 616298
DEF6	96,7	93,8	100	99,9	No OMIM disease ID
DHFR	92,1	78,9	100	100	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	99,8	98,7	100	99,7	Dyskeratosis congenita, X-linked, 305000

DNASE1	100	99,9	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700
DNASE1L3	100	100	100	100	Systemic lupus erythematosus 16, 614420
DNASE2	99,7	97,1	100	100	No OMIM disease ID
DNMT3B	100	100	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	100	99,6	100	100	Immunodeficiency 40, 616433
DOCK8	100	99,6	100	100	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	99,7	97,4	100	100	Neutropenia, severe congenital 1, autosomal dominant, 202700 Neutropenia, cyclic, 162800
ELF4	100	99,7	100	100	No OMIM disease ID
EPG5	99,5	98,5	100	100	Vici syndrome, 242840
ERCC2	100	99,7	100	100	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	96,9	96,3	100	100	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
EXTL3	100	100	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
F12	99,9	98,8	100	100	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
FAAP24	99,3	96,7	100	100	No OMIM disease ID
FADD	100	100	100	100	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAS	100	99,6	100	100	Autoimmune lymphoproliferative syndrome, type IA, 601859 {Autoimmune lymphoproliferative syndrome}, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0
FASLG	100	99,6	100	100	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FAT4	100	100	100	100	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FCGR1A	46,8	44,1	100	100	[IgG receptor I, phagocytic, familial deficiency of], 0
FCGR2A	100	100	100	100	{Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700 {Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162

FCGR2B	99,5	95,4	100	100	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700
FCGR3A	99	97,1	100	100	Immunodeficiency 20, 615707
FCGR3B	99,3	97,9	98,1	98	No OMIM disease ID
FCHO1	98,9	97,7	100	100	No OMIM disease ID
FCN3	100	100	100	100	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	100	100	100	100	Leukocyte adhesion deficiency, type III, 612840
FOXN1	100	99,6	100	100	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	99,2	95,5	100	100	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FPR1	100	100	100	100	No OMIM disease ID
G6PC	100	100	100	100	Glycogen storage disease Ia, 232200
G6PC3	100	99,9	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	99,3	98	100	100	Hemolytic anemia, G6PD deficient (favism), 300908 {Resistance to malaria due to G6PD deficiency}, 611162
GATA2	100	98,3	100	100	Emberger syndrome, 614038 {Myelodysplastic syndrome, susceptibility to}, 614286 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626
GFI1	100	99,2	100	100	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GINS1	99,3	94,9	100	100	Immunodeficiency 55, 617827
GJC2	78,2	58,7	96,9	91,4	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GRHL2	100	100	100	100	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GTF2H5	72,5	72,2	72,5	72,5	Trichothiodystrophy 3, photosensitive, 616395
HAVCR2	100	100	100	100	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100	100	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738

HELLS	97,8	92,1	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HMOX1	98,4	89,9	100	100	{Pulmonary disease, chronic obstructive, susceptibility to}, 606963 Heme oxygenase-1 deficiency, 614034
HYOU1	100	99,5	100	100	?Immunodeficiency 59 and hypoglycemia, 233600
ICOS	99,9	99,8	100	100	Immunodeficiency, common variable, 1, 607594
ICOSLG	99,5	98,8	100	100	No OMIM disease ID
IFIH1	99,7	98,4	100	100	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNAR1	97,7	97,1	97,8	97,8	No OMIM disease ID
IFNAR2	100	99,7	100	100	?Immunodeficiency 45, 616669 {Hepatitis B virus, susceptibility to}, 610424
IFNG	100	100	100	100	?Immunodeficiency 69, mycobacteriosis, 618963 {AIDS, rapid progression to}, 609423 {TSC2 angiomyolipomas, renal, modifier of}, 613254 {Hepatitis C virus, response to therapy of}, 609532 {Aplastic anemia}, 609135 {Tuberculosis, protection against}, 607948
IFNGR1	98,2	97,5	100	100	Immunodeficiency 27A, mycobacteriosis, AR, 209950 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424
IFNGR2	93,3	93,2	100	99,8	Immunodeficiency 28, mycobacteriosis, 614889
IGHM	100	100	100	100	Agammaglobulinemia 1, 601495
IGLL1	99,9	96,9	100	100	Agammaglobulinemia 2, 613500
IKBKB	99,8	97,4	100	100	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	84,1	77,2	100	100	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291
IKZF1	99,3	99,3	100	100	Immunodeficiency, common variable, 13, 616873

IL10	99,8	98,2	100	100	{Graft-versus-host disease, protection against}, 614395 {HIV-1, susceptibility to}, 609423 {Rheumatoid arthritis, progression of}, 180300
IL10RA	100	100	100	100	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	99,8	98	100	100	{Hepatitis B virus, susceptibility to}, 610424 Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL12B	100	99,3	100	100	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	98,9	96,3	94,1	94,1	Immunodeficiency 30, 614891
IL17F	99,9	97,2	100	100	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100	99,4	100	100	Immunodeficiency 51, 613953
IL17RC	100	99,9	100	100	Candidiasis, familial, 9, 616445
IL18BP	100	100	100	100	{?Hepatitis, fulminant viral, susceptibility to}, 618549
IL1RN	100	100	100	100	{Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628 Interleukin 1 receptor antagonist deficiency, 612852
IL2	94,5	88	100	100	No OMIM disease ID
IL21	99,4	95,7	100	100	?Immunodeficiency, common variable, 11, 615767
IL21R	100	100	100	100	[IgE, elevated level of], 147050 Immunodeficiency 56, 615207
IL2RA	100	99,7	100	100	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RB	100	99,7	100	100	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL2RG	99,8	97,1	100	100	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL36RN	100	100	100	100	Psoriasis 14, pustular, 614204
IL6R	98,4	94,2	92,8	92,7	[Interleukin-6 receptor, soluble, serum level of, QTL], 614689 [Interleukin 6, serum level of, QTL], 614752 Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944
IL6ST	96,4	90,3	100	100	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IL7R	100	99,8	100	100	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INO80	100	99,1	100	100	No OMIM disease ID

INSR	97,8	94,7	99,9	99,2	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200
IRAK1	99,3	94,9	99,9	99,4	No OMIM disease ID
IRAK4	99,8	97,7	100	100	Immunodeficiency 67, 607676
IRF2BP2	93,9	77,7	100	99,9	?Immunodeficiency, common variable, 14, 617765
IRF3	100	99,8	100	100	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532
IRF4	100	100	100	100	[Skin/hair/eye pigmentation, variation in, 8], 611724
IRF7	100	99,9	100	100	?Immunodeficiency 39, 616345
IRF8	99	95,7	100	100	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893
IRF9	100	100	100	100	Immunodeficiency 65, susceptibility to viral infections, 618648
ISG15	100	100	100	100	Immunodeficiency 38, 616126
ITCH	91,6	91,3	95,9	95	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGB2	97,2	97,2	97,2	97,2	Leukocyte adhesion deficiency, 116920
ITK	100	98,9	100	100	Lymphoproliferative syndrome 1, 613011
ITPR3	100	99,7	100	100	{Diabetes, type 1, susceptibility to}, 222100
IVNS1ABP	99,9	98,4	100	100	Immunodeficiency 70, 618969
JAGN1	100	100	99,7	98	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	100	99,8	100	99,7	Autoinflammation, immune dysregulation, and eosinophilia, 618999
JAK2	98,1	95,8	100	100	Myelofibrosis, somatic, 254450 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300 {Budd-Chiari syndrome, somatic}, 600880 Leukemia, acute myeloid, somatic, 601626 Erythrocytosis, somatic, 133100
JAK3	99,9	98,7	100	100	SCID, autosomal recessive, T-negative/B-positive type, 600802
KDM6A	96,1	88,7	100	99,9	Kabuki syndrome 2, 300867

KMT2D	100	99,4	100	100	Kabuki syndrome 1, 147920
LACC1	100	99,4	100	100	Juvenile arthritis, 618795
LAMTOR2	100	99,7	100	100	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAT	100	99,2	100	100	Immunodeficiency 52, 617514
LCK	98,9	96,6	100	100	?Immunodeficiency 22, 615758
LIG1	100	99,7	100	100	No OMIM disease ID
LIG4	100	99,9	100	100	{Multiple myeloma, resistance to}, 254500 LIG4 syndrome, 606593
LPIN2	100	100	100	100	Majeed syndrome, 609628
LRBA	99,9	99,6	100	100	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	100	99,8	100	100	?Agammaglobulinemia 5, 613506
LYST	99,6	98,3	100	100	Chediak-Higashi syndrome, 214500
MAGT1	98,5	96,5	98,7	98,7	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 Congenital disorder of glycosylation, type Icc, 301031
MAL2	99,9	98	100	99,9	No OMIM disease ID
MALT1	91,2	89,4	100	100	Immunodeficiency 12, 615468
MAN2B1	99,8	97,9	100	100	Mannosidosis, alpha-, types I and II, 248500
MANBA	87,8	86,5	100	100	Mannosidosis, beta, 248510
MAP3K14	100	99,9	100	100	No OMIM disease ID
MASP2	100	99,6	100	100	MASP2 deficiency, 613791
MBL2	100	99,8	100	100	{Chronic infections, due to MBL deficiency}, 614372
MC2R	99,9	98,3	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM4	95,5	95	95,5	95,5	Immunodeficiency 54, 609981
MEFV	99,9	98,6	96,4	96,4	Neutrophilic dermatosis, acute febrile, 608068 Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MOGS	100	99,9	100	100	Congenital disorder of glycosylation, type IIb, 606056

MRE11	98,9	93,3	100	100	Ataxia-telangiectasia-like disorder 1, 604391
MRTFA	91,4	90,2	92,8	92,8	?Immunodeficiency 66, 618847
MS4A1	99,8	98,8	100	100	?Immunodeficiency, common variable, 5, 613495
MSN	99	95,7	100	100	Immunodeficiency 50, 300988
MTHFD1	100	99,5	100	100	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MVK	90,9	90,5	90,5	90,5	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYD88	100	100	100	100	Immunodeficiency 68, 612260 Macroglobulinemia, Waldenstrom, somatic, 153600
MYSM1	96,4	95,5	96,4	96,4	Bone marrow failure syndrome 4, 618116
NBAS	100	99,6	100	100	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBN	99,9	98,6	100	100	Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135
NCF1	26	25,8	100	99,8	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	99,9	98,3	100	100	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100	100	100	100	Chronic granulomatous disease 3, autosomal recessive, 613960
NCKAP1L	100	99,9	100	100	Immunodeficiency 72 with autoinflammation, 618982
NCSTN	100	99,8	100	100	Acne inversa, familial, 1, 142690
NFAT5	99,8	99,1	100	100	No OMIM disease ID
NFE2L2	100	99,9	100	100	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFKB1	100	99,4	100	100	Immunodeficiency, common variable, 12, 616576
NFKB2	98,8	95,6	100	100	Immunodeficiency, common variable, 10, 615577
NFKBIA	95,2	88	100	100	Ectodermal dysplasia and immunodeficiency 2, 612132
NHEJ1	100	96,2	100	100	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987

NLRC4	100	100	100	100	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
NLRP1	99,6	98	100	100	Palmoplantar carcinoma, multiple self-healing, 615225 Autoinflammation with arthritis and dyskeratosis, 617388 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803
NLRP12	100	99,9	100	100	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	100	99,9	100	100	Familial cold inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Keratoendothelitis fugax hereditaria, 148200
NLRP6	98,7	95,9	100	100	No OMIM disease ID
NLRP7	100	99,6	100	100	Hydatidiform mole, recurrent, 1, 231090
NOD2	100	99,9	100	100	{Yao syndrome}, 617321 Blau syndrome, 186580 {Inflammatory bowel disease 1, Crohn disease}, 266600
NOP10	100	99,8	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	100	100	100	100	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
NSMCE3	100	100	100	100	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
OAS1	100	100	100	100	No OMIM disease ID
ORAI1	99,1	96,4	99,6	97,1	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782
OSTM1	98,6	94	100	100	Osteopetrosis, autosomal recessive 5, 259720
OTULIN	92,6	86,5	99,2	95	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
PARN	81,2	81,1	88,1	87,6	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353

PAX5	98,7	96,1	100	100	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PBX1	100	99,4	100	100	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	99,5	96,7	100	100	Propionicacidemia, 606054
PCCB	97,9	96	98,7	96,2	Propionicacidemia, 606054
PEPD	100	98,8	100	100	Prolidase deficiency, 170100
PEX16	97,9	94,2	100	100	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PGM3	100	99,8	91,7	91,7	Immunodeficiency 23, 615816
PIGA	93,8	86,7	100	100	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIK3CD	98,8	96,9	100	100	Immunodeficiency 14, 615513
PIK3CG	100	100	100	100	No OMIM disease ID
PIK3R1	99,8	99	100	100	?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880 Immunodeficiency 36, 616005
PLCG2	100	99,8	100	100	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLEKHM1	100	99,8	100	100	Osteopetrosis, autosomal dominant 3, 618107 ?Osteopetrosis, autosomal recessive 6, 611497
PLG	87,8	87,5	100	100	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PMM2	100	100	100	100	Congenital disorder of glycosylation, type Ia, 212065
PNP	99,8	98,9	100	100	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POLA1	99,3	95,4	100	99,9	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLE2	97,3	89,8	100	100	No OMIM disease ID
POMP	100	99,1	100	100	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POT1	99,9	99	100	100	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
POU2AF1	100	99,3	100	100	No OMIM disease ID

PRF1	91,2	90,8	100	100	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRKCD	100	100	100	100	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	99,7	98	100	100	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	86,4	86,4	100	100	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500 Arts syndrome, 301835 Gout, PRPS-related, 300661
PSENEN	100	100	100	100	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMA3	99,8	97,2	100	100	No OMIM disease ID
PSMB4	100	100	100	100	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	99,9	98,5	100	100	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	99,9	97,7	100	100	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMG2	100	98,9	100	100	No OMIM disease ID
PSTPIP1	100	99,1	100	99,9	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN22	99,5	97,1	100	100	{Diabetes, type 1, susceptibility to}, 222100 {Systemic lupus erythematosus susceptibility to}, 152700 {Rheumatoid arthritis, susceptibility to}, 180300
PTPRC	99	95,1	100	100	{Hepatitis C virus, susceptibility to}, 609532 Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
RAB27A	100	100	100	100	Griscelli syndrome, type 2, 607624
RAC2	99,9	98,3	100	100	?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986 Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203
RAG1	100	100	100	100	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 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	100	100	100	100	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RANBP2	49,7	49,3	100	100	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033
RASGRP1	100	99,6	100	100	Immunodeficiency 64, 618534
RASGRP2	99,7	97,3	100	100	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	99,9	98,2	100	100	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RC3H1	100	99,4	100	100	?Immune dysregulation and systemic hyperinflammation syndrome, 618998
RECQL4	99,8	98,1	100	99,9	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
RELB	98,8	88,7	100	100	?Immunodeficiency 53, 617585
RFX5	99,7	98,1	100	100	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	100	99,5	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	99,3	97	100	99,9	Bare lymphocyte syndrome, type II, complementation group D, 209920
RHOG	100	100	100	100	No OMIM disease ID
RHOH	100	100	100	100	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307
RIPK1	100	99	100	100	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852
RMRP					Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	100	100	100	100	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	80,6	78,1	91	90,9	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100	99,5	100	100	Aicardi-Goutieres syndrome 3, 610329
RNF168	100	99,8	100	100	RIDDLE syndrome, 611943
RNF31	99,9	99	100	100	No OMIM disease ID

RNU4ATAC					Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
RORC	100	100	100	100	Immunodeficiency 42, 616622
RPSA	100	99,8	100	100	Asplenia, isolated congenital, 271400
RSPH9	99,9	97,9	100	100	Ciliary dyskinesia, primary, 12, 612650
RTEL1	99,5	96,8	100	100	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
SAMD9	100	99,8	100	100	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041
SAMD9L	100	100	100	100	Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Ataxia-pancytopenia syndrome, 159550
SAMHD1	98,7	98,4	100	100	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SASH3	99,9	97,6	100	100	No OMIM disease ID
SBDS	100	100	100	100	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400
SEC61A1	100	100	100	100	Hyperuricemic nephropathy, familial juvenile, 4, 617056
SEMA3E	99,2	98,9	100	100	?CHARGE syndrome, 214800
SERAC1	99,9	99,5	100	100	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	99,7	97,5	100	100	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SH2B3	99,4	95,1	100	99,9	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SH2D1A	97,2	94	100	100	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	91,4	91,2	97	95,3	Cherubism, 118400
SH3KBP1	99,7	95,9	100	100	?Immunodeficiency 61, 300310
SKIV2L	100	99,8	100	100	Trichohepatoenteric syndrome 2, 614602
SLC29A3	100	99,6	100	100	Histiocytosis-lymphadenopathy plus syndrome, 602782

SLC35A1	100	99,7	100	100	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	99,9	98,7	100	100	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	100	99,2	100	100	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC39A4	99,5	95,5	100	100	Acrodermatitis enteropathica, 201100
SLC39A7	100	100	100	100	No OMIM disease ID
SLC46A1	99,9	98,5	100	100	Folate malabsorption, hereditary, 229050
SLC7A7	100	99,9	100	100	Lysinuric protein intolerance, 222700
SMARCAL1	100	99,9	100	100	Schimke immunoosseous dysplasia, 242900
SMARCD2	87	85,9	99,6	97	Specific granule deficiency 2, 617475
SNX10	96,2	95,7	100	99,6	Osteopetrosis, autosomal recessive 8, 615085
SOCS1	100	100	100	100	No OMIM disease ID
SOCS4	99,9	99,2	100	100	No OMIM disease ID
SP110	100	100	100	100	{Mycobacterium tuberculosis, susceptibility to}, 607948 Hepatic venoocclusive disease with immunodeficiency, 235550
SPINK5	99,9	99,5	100	100	Netherton syndrome, 256500
SPPL2A	85,9	74,6	100	100	No OMIM disease ID
SRP54	99,5	96,5	100	100	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	97,6	89,7	100	100	Bone marrow failure syndrome 1, 614675
STAT1	93,7	91,7	95,7	94,8	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796
STAT2	100	99,9	100	100	Immunodeficiency 44, 616636 Pseudo-TORCH syndrome 3, 618886
STAT3	100	99,8	100	100	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT4	99,9	99,6	100	100	{Systemic lupus erythematosus, susceptibility to, 11}, 612253
STAT5B	100	98,5	100	100	Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 Leukemia, acute promyelocytic, somatic, 102578

STAT6	100	99,9	100	100	No OMIM disease ID
STIM1	99,8	98	100	100	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STING1	99,7	95,3	100	100	STING-associated vasculopathy, infantile-onset, 615934
STK4	100	99,8	100	100	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	100	100	100	100	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	82,1	79,7	99,3	97,1	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TAP1	100	99,2	100	100	Bare lymphocyte syndrome, type I, 604571
TAP2	99,9	99,3	100	100	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,5	95,5	96,6	96,6	Bare lymphocyte syndrome, type I, 604571
TAZ	99,1	95,5	100	100	Barth syndrome, 302060
TBX1	87	77,5	94	89,9	Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500
TBX21	95,4	86,6	100	100	{Asthma, aspirin-induced, susceptibility to}, 208550 Asthma and nasal polyps, 208550
TCF3	97,1	94	100	100	Agammaglobulinemia 8, autosomal dominant, 616941
TCIRG1	97,6	90,1	100	100	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100	100	100	100	Transcobalamin II deficiency, 275350
TERC					{Aplastic anemia}, 614743 Dyskeratosis congenita, autosomal dominant 1, 127550 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERT	96,2	94,5	100	100	{Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 {Leukemia, acute myeloid}, 601626 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Dyskeratosis congenita, autosomal dominant 2}, 613989
TFRC	100	99,8	100	100	Immunodeficiency 46, 616740

TGFB1	100	99,9	100	100	{Cystic fibrosis lung disease, modifier of}, 219700 Camurati-Engelmann disease, 131300 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213
THBD	100	99,7	100	100	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TICAM1	100	100	100	100	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850
TINF2	100	100	100	100	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TIRAP	100	100	100	100	{Bacteremia, protection against}, 614382 {Malaria, protection against}, 611162 {Tuberculosis, protection against}, 607948
TLR3	100	99,6	100	100	{HIV1 infection, resistance to}, 609423 {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002
TLR4	100	99,8	100	100	No OMIM disease ID
TLR7	100	99,9	100	100	Immunodeficiency 74, COVID19-related, X-linked, 301051
TLR8	100	99,8	100	100	No OMIM disease ID
TMC6	100	99,3	100	100	Epidermodysplasia verruciformis, 226400
TMC8	100	98,7	100	100	Epidermodysplasia verruciformis 2, 618231
TNFAIP3	100	100	100	100	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF11A	94,6	93,3	99,2	98	Osteolysis, familial expansile, 174810 {Paget disease of bone 2, early-onset}, 602080 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF13B	100	100	100	100	Immunoglobulin A deficiency 2, 609529 Immunodeficiency, common variable, 2, 240500
TNFRSF13C	80,1	75,4	100	99,9	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	90,6	87,6	92,8	92,8	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFRSF4	99,4	95,4	100	100	?Immunodeficiency 16, 615593
TNFRSF9	100	100	100	100	No OMIM disease ID
TNFSF11	100	99,9	100	100	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	98	93,6	100	100	No OMIM disease ID

TOP2B	99,4	96,3	100	100	No OMIM disease ID
TPP2	99,2	96,8	100	100	No OMIM disease ID
TRAC	100	100	100	100	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	100	99,9	100	100	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849
TRAF3IP2	100	99,3	100	100	?Candidiasis, familial, 8, 615527 {Psoriasis susceptibility 13}, 614070
TREX1	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM22	100	100	100	100	No OMIM disease ID
TRNT1	99,5	96,5	100	100	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TTC37	100	99,3	100	100	Trichohepatoenteric syndrome 1, 222470
TTC7A	99,3	95,4	100	100	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	99,9	99	100	100	Immunodeficiency 35, 611521
UBA1	99,4	98,2	99,8	99	VEXAS syndrome, somatic, 301054 Spinal muscular atrophy, X-linked 2, infantile, 301830
UNC13D	99,7	98,1	100	100	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	60,6	58,8	100	100	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551
UNG	100	98,8	99,9	99,3	Immunodeficiency with hyper IgM, type 5, 608106
USB1	100	99,4	100	100	Poikiloderma with neutropenia, 604173
USP18	95,9	95,9	100	100	Pseudo-TORCH syndrome 2, 617397
VAV1	98,5	97,1	97,1	97,1	No OMIM disease ID
VPS13B	99,5	98,2	99,5	99,4	Cohen syndrome, 216550
VPS45	99,2	95,7	95,3	95,3	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	95,9	85,3	100	99,8	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299

WDR1	100	99,6	100	100	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550
WIPF1	100	99,9	100	100	Wiskott-Aldrich syndrome 2, 614493
WRAP53	100	100	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	93	88,8	100	100	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	100	99,3	100	100	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840
ZBTB24	100	100	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZNF341	97,2	95	100	100	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282

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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
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 coverage denoting NC are non-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors