PRIMARY IMMUNODEFICIENCY GENE PANEL DG 3.2.0 (471 genes)

Releasedate: 16-09-2021

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 recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ACP5	99,9	98,9	100	100	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	99,9	97,2	100	100	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ADA	99,7	96,1	100	100	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	99,9	97,3	100	100	Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	99,6	98	100	99,9	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAR	100	99,4	100	100	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
AGA	100	99,9	100	100	Aspartylglucosaminuria, 208400
AICDA	100	99,9	100	100	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	100	99,9	100	100	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	98,7	95,2	100	99,7	Reticular dysgenesis, 267500
ALG13	97,4	90	99,9	99,4	?Congenital disorder of glycosylation, type Is, 300884 Developmental and epileptic encephalopathy 36, 300884
ALPI	100	99,7	100	100	No OMIM disease ID
ANGPT1	99,7	98,7	100	100	?Angioedema, hereditary, 5, 619361
AP1S3	90,4	90,1	90,5	90,5	No OMIM disease ID
AP3B1	99,2	96,4	100	99,9	Hermansky-Pudlak syndrome 2, 608233
AP3D1	99,6	98,4	100	100	?Hermansky-Pudlak syndrome 10, 617050
APOL1	100	100	100	100	No OMIM disease ID
ARHGEF1	99,9	98,4	100	100	?Immunodeficiency 62, 618459

ARPC1B	100	100	100	100	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ATG4A	99	94,3	99,9	97,9	No OMIM disease ID
ATM	99,4	97,1	100	100	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, T-cell prolymphocytic leukemia, somatic, Lymphoma, mantle cell, somatic,
ATP6AP1	98,2	93	100	100	Immunodeficiency 47, 300972
B2M	100	100	100	100	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
BACH2	100	99,9	100	100	Immunodeficiency 60, 618394
BCL10	100	100	100	100	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245
BCL11B	99,6	96,5	99,4	97,9	Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 Immunodeficiency 49, 617237
BLK	100	100	100	100	Maturity-onset diabetes of the young, type 11, 613375
BLM	99,3	97,7	100	100	Bloom syndrome, 210900
BLNK	96,9	92,6	100	100	?Agammaglobulinemia 4, 613502
BLOC1S6	99,3	97,6	100	100	?Hermansky-Pudlak syndrome 9, 614171
BTK	100	99,7	100	99,8	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	99,7	100	100	C1q deficiency, 613652
C1R	100	100	99,7	98	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	99,9	98,8	99,7	97,7	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	100	100	100	100	C2 deficiency, 217000
C2orf69	96,7	85,6	100	100	Combined oxidative phosphorylation deficiency 53, 619423
C3	99,9	98,5	100	100	C3 deficiency, 613779
C5	99,6	98,2	100	99,8	C5 deficiency, 609536
C6	100	99,8	100	100	C6 deficiency, 612446 Combined C6/C7 deficiency,
C7	99,8	97,4	100	100	C7 deficiency, 610102

C8A	100	99,4	100	100	C8 deficiency, type I, 613790
C8B	99,9	98,6	100	100	C8 deficiency, type II, 613789
C8G	100	100	100	100	No OMIM disease ID
C9	99,7	99,2	100	100	C9 deficiency, 613825
CA2	100	100	100	100	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CARD11	100	99,9	100	100	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11B with atopic dermatitis, 617638 Immunodeficiency 11A, 615206
CARD14	100	99,5	100	100	Psoriasis 2, 602723 Pityriasis rubra pilaris, 173200
CARD9	100	99	100	100	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	96,4	95,2	99,8	98,8	Immunodeficiency 58, 618131
CASP10	99,3	96,8	100	100	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP8	95,6	95,4	95,6	95,6	Hepatocellular carcinoma, somatic, 114550 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271
CAVIN1	100	100	100	100	Lipodystrophy, congenital generalized, type 4, 613327
CCBE1	99,9	98,8	100	100	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD19	100	100	100	100	Immunodeficiency, common variable, 3, 613493
CD247	100	99,4	100	100	?Immunodeficiency 25, 610163
CD27	99,9	98,3	100	100	Lymphoproliferative syndrome 2, 615122
CD28	100	99,9	100	100	No OMIM disease ID
CD3D	100	99,9	100	100	Immunodeficiency 19, 615617
CD3E	100	98,8	100	100	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	100	100	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	100	99,7	100	100	Immunodeficiency 79, 619238 OKT4 epitope deficiency, 613949
CD40	100	100	100	100	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	92,4	81	100	100	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	99,7	98,9	100	99,9	No OMIM disease ID

CD55	91,5	82,6	95	92,6	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	75,5	67	64,5	64,5	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	100	96,8	100	100	Lymphoproliferative syndrome 3, 618261
CD79A	100	100	100	100	Agammaglobulinemia 3, 613501
CD79B	100	100	100	100	Agammaglobulinemia 6, 612692
CD81	100	99,6	100	99,8	Immunodeficiency, common variable, 6, 613496
CD8A	100	100	100	100	CD8 deficiency, familial, 608957
CDC42	96,3	87,9	100	100	Takenouchi-Kosaki syndrome, 616737
CDCA7	100	99,6	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDKN2B	100	99,7	100	100	No OMIM disease ID
CEBPE	100	100	100	100	Specific granule deficiency, 245480
CFB	100	99,6	100	100	?Complement factor B deficiency, 615561
CFD	90,9	84,6	100	100	Complement factor D deficiency, 613912
CFH	99,8	98,5	100	99,9	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	91,7	89,6	96,3	94,1	No OMIM disease ID
CFHR2	75,7	74,4	76,1	76,1	No OMIM disease ID
CFHR3	89	87,8	97,7	96	No OMIM disease ID
CFHR4	99,8	99,5	100	99,7	No OMIM disease ID
CFHR5	99,8	97,6	100	100	Nephropathy due to CFHR5 deficiency, 614809
CFI	99,3	96	100	99,9	Complement factor I deficiency, 610984
CFP	100	99	100	100	Properdin deficiency, X-linked, 312060
CFTR	99,5	97,9	100	100	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
CHD7	100	99,2	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CIB1	98,1	94,5	100	100	Epidermodysplasia verruciformis 3, 618267
CIITA	100	99,4	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920

CLCN7	99,4	97,8	100	100	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
CLEC4D	100	99,9	100	100	No OMIM disease ID
CLEC7A	100	99,7	100	100	Candidiasis, familial, 4, autosomal recessive, 613108
CLPB	94,9	94	100	100	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COLEC11	100	100	100	100	3MC syndrome 2, 265050
COPA	99,9	99,2	100	100	No OMIM disease ID
CORO1A	99,9	98,9	100	99,9	Immunodeficiency 8, 615401
CR2	100	99,9	100	100	Immunodeficiency, common variable, 7, 614699
CREBBP	99,6	97,8	100	100	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CSF2RA	89,6	84,4	94,6	90,9	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	99,9	99	100	100	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	99,8	98,5	100	100	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	100	99,1	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100	100	100	100	Autoimmune lymphoproliferative syndrome, type V, 616100
CTNNBL1	99,8	98,7	100	100	No OMIM disease ID
CTPS1	93	93	93	93	Immunodeficiency 24, 615897
CTSC	100	100	100	100	Periodontitis 1, juvenile, 170650 Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000
CXCR4	100	100	100	100	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670
CYBA	96	82,5	100	100	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBB	99,8	98,8	100	100	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400
CYBC1	100	99,3	100	100	Chronic granulomatous disease 5, autosomal recessive, 618935
DBF4	96	88,9	100	99,9	No OMIM disease ID
DBR1	99,8	99	100	100	No OMIM disease ID
DCLRE1C	99,8	98,2	100	99,9	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554

DDX41	100	100	100	100	No OMIM disease ID
DDX58	99,6	98,3	100	99,9	Singleton-Merten syndrome 2, 616298
DEF6	96,4	92,9	100	100	No OMIM disease ID
DHFR	88,9	76,3	100	100	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DIAPH1	99,8	98,4	99,9	99	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DKC1	99,7	97,2	100	99,6	Dyskeratosis congenita, X-linked, 305000
DNASE1	100	100	100	100	No OMIM disease ID
DNASE1L3	100	100	100	100	Systemic lupus erythematosus 16, 614420
DNASE2	98,4	95,1	100	100	No OMIM disease ID
DNMT3B	100	99,9	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK2	99,8	98,8	100	100	Immunodeficiency 40, 616433
DOCK8	100	99	100	100	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	99,9	98,8	100	100	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	99,9	98,7	100	100	No OMIM disease ID
EPG5	99,2	97,8	100	100	Vici syndrome, 242840
ERCC2	100	99,4	100	100	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	96,8	95,6	100	100	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
EXTL3	100	100	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
F12	100	98,6	100	100	Angioedema, hereditary, 3, 610618 Factor XII deficiency, 234000
FAAP24	98,3	94,8	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 Squamous cell carcinoma, burn scar-related, somatic,
FASLG	100	99,1	100	100	Autoimmune lymphoproliferative syndrome, type IB, 601859
FAT4	100	100	100	100	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006

FBXW11	99,5	95,9	100	100	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FCGR1A	46,5	41,8	100	99,9	No OMIM disease ID
FCGR2A	100	100	100	100	No OMIM disease ID
FCGR2B	99,5	96,1	100	100	No OMIM disease ID
FCGR3A	98,6	96,8	100	99,9	Immunodeficiency 20, 615707
FCGR3B	99,1	97,3	99,5	97,9	No OMIM disease ID
FCHO1	99,3	97,5	100	100	Immunodeficiency 76, 619164
FCN3	100	99,3	100	100	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	100	100	100	100	Leukocyte adhesion deficiency, type III, 612840
FNIP1	99,9	99,9	100	100	No OMIM disease ID
FOXN1	100	99,1	100	100	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	98,5	94,8	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	99,1	97,4	100	100	Hemolytic anemia, G6PD deficient (favism), 300908
GATA2	99,8	97	100	100	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GFI1	100	99,9	100	100	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GIMAP5	100	100	100	100	Portal hypertension, noncirrhotic, 2, 619463
GINS1	98,4	93,4	100	100	Immunodeficiency 55, 617827
GJC2	82,3	64,5	97,8	93,2	Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206 Leukodystrophy, hypomyelinating, 2, 608804
GRHL2	100	99,9	100	100	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
GTF2H5	72,2	71,7	72,5	72,5	Trichothiodystrophy 3, photosensitive, 616395
HAVCR2	100	99,8	100	100	T-cell lymphoma, subcutaneous panniculitis-like, 618398

HAX1	100	100	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HELLS	98,2	91,9	100	99,9	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HMOX1	97,7	90,1	100	100	Heme oxygenase-1 deficiency, 614034
HYOU1	99,9	99	100	100	?Immunodeficiency 59 and hypoglycemia, 233600
ICOS	99,9	99,8	100	99,9	Immunodeficiency, common variable, 1, 607594
ICOSLG	99,6	99,1	100	100	No OMIM disease ID
IFIH1	99,5	97,3	100	100	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNAR1	97,4	96,6	97,8	97,7	No OMIM disease ID
IFNAR2	99,8	98,8	100	100	?Immunodeficiency 45, 616669
IFNG	100	99,9	100	100	?Immunodeficiency 69, mycobacteriosis, 618963
IFNGR1	98	97,3	100	100	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978
IFNGR2	93,7	93,2	100	99,5	Immunodeficiency 28, mycobacteriosis, 614889
IGHM	100	100	100	100	Agammaglobulinemia 1, 601495
IGLL1	100	99,7	100	100	Agammaglobulinemia 2, 613500
IKBKB	99	96,1	100	100	Immunodeficiency 15B, 615592 Immunodeficiency 15A, 618204
IKBKG	84,6	75,2	100	100	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636
IKZF1	99,3	99,3	100	100	Immunodeficiency, common variable, 13, 616873
IKZF3	100	100	100	100	?Immunodeficiency 84, 619437
IL10	100	98,1	100	100	No OMIM disease ID
IL10RA	100	99,9	100	100	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	99,9	97,8	100	100	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL12B	100	99,1	100	100	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	98,7	96,1	94,1	94,1	Immunodeficiency 30, 614891
IL17F	99,9	97,6	100	100	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100	99,8	100	100	Immunodeficiency 51, 613953
IL17RC	100	99,9	100	100	Candidiasis, familial, 9, 616445

IL18BP	100	100	100	100	No OMIM disease ID
IL1RN	100	99,9	100	100	Interleukin 1 receptor antagonist deficiency, 612852
IL2	96,2	88,8	100	100	No OMIM disease ID
IL21	99,2	93,5	100	100	?Immunodeficiency, common variable, 11, 615767
IL21R	100	100	100	100	Immunodeficiency 56, 615207
IL2RA	100	99,1	100	100	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL2RB	100	99,8	100	100	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL2RG	99,8	92,8	100	100	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL36RN	100	99,9	100	100	Psoriasis 14, pustular, 614204
IL6R	99,1	94,2	92,7	92,7	Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944
IL6ST	94,9	89,4	100	100	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IL7R	99,9	99,3	100	100	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INO80	99,9	98	100	100	No OMIM disease ID
INSR	97,3	93	100	99,6	Rabson-Mendenhall syndrome, 262190 Leprechaunism, 246200 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
IPO8	99,5	97	100	99,9	VISS syndrome, 619472
IRAK1	99,4	95,9	99,7	98,9	No OMIM disease ID
IRAK4	99,5	95,7	100	99,8	Immunodeficiency 67, 607676
IRF2BP2	97,2	83,5	100	100	?Immunodeficiency, common variable, 14, 617765
IRF3	100	99,9	100	100	No OMIM disease ID
IRF4	100	100	100	100	No OMIM disease ID
IRF7	100	99,8	100	100	?Immunodeficiency 39, 616345
IRF8	98,7	96	100	100	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRF9	100	99,9	100	100	Immunodeficiency 65, susceptibility to viral infections, 618648
IRGM	100	100	100	100	No OMIM disease ID
ISG15	100	100	100	100	Immunodeficiency 38, 616126
ITCH	91,5	90,8	95,3	93,1	Autoimmune disease, multisystem, with facial dysmorphism, 613385

ITGB2	97,2	97,2	97,2	97,2	Leukocyte adhesion deficiency, 116920
ITK	99,8	98,6	100	100	Lymphoproliferative syndrome 1, 613011
ITPR3	100	99,6	100	100	No OMIM disease ID
IVNS1ABP	99,1	96,4	100	100	Immunodeficiency 70, 618969
JAGN1	100	100	100	99,2	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	99,9	99,4	100	100	Autoinflammation, immune dysregulation, and eosinophilia, 618999
JAK2	97,6	95,2	100	99,9	Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300
JAK3	99,5	97,6	100	100	SCID, autosomal recessive, T-negative/B-positive type, 600802
KDM6A	94,2	85,9	100	99,9	Kabuki syndrome 2, 300867
KMT2D	99,9	99	100	100	Kabuki syndrome 1, 147920
KRAS	99	97,8	100	100	Gastric cancer, somatic, 137215 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 Noonan syndrome 3, 609942 RAS-associated autoimmune leukoproliferative disorder, 614470 Arteriovenous malformation of the brain, somatic, 108010 Lung cancer, somatic, 211980 Pancreatic carcinoma, somatic, 260350 Leukemia, acute myeloid, somatic, 601626 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Cardiofaciocutaneous syndrome 2, 615278 Bladder cancer, somatic, 109800
LACC1	99,7	98,4	100	100	Juvenile arthritis, 618795
LAMTOR2	100	100	100	100	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAT	100	99,4	100	100	Immunodeficiency 52, 617514
LCK	98,2	96,1	100	100	?Immunodeficiency 22, 615758
LCP2	99,6	95,4	100	100	?Immunodeficiency 81, 619374
LIG1	100	99,7	100	100	No OMIM disease ID
LIG4	99,8	99,3	100	100	LIG4 syndrome, 606593

LPIN2	99,9	99,7	100	100	Majeed syndrome, 609628
LRBA	99,9	99,7	100	100	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC32	100	100	100	100	Cleft palate, proliferative retinopathy, and developmental delay, 619074
LRRC8A	100	99,7	100	100	?Agammaglobulinemia 5, 613506
LSM11	99,9	97,6	100	98,9	?Aicardi-Goutieres syndrome 8, 619486
LYST	99,4	97,8	100	100	Chediak-Higashi syndrome, 214500
MAGT1	98	94,5	98,7	98,4	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 Congenital disorder of glycosylation, type Icc, 301031
MAL2	100	99	100	99,7	No OMIM disease ID
MALT1	90,9	87,7	100	99,9	Immunodeficiency 12, 615468
MAN2B1	99,6	97,4	100	100	Mannosidosis, alpha-, types I and II, 248500
MANBA	87,1	84,9	100	99,9	Mannosidosis, beta, 248510
MAP1LC3B2	100	100	100	100	No OMIM disease ID
MAP3K14	100	99,8	100	100	No OMIM disease ID
MAPK8	99,9	99,7	100	100	No OMIM disease ID
MASP2	100	99,3	100	100	MASP2 deficiency, 613791
MBL2	99,9	99,4	100	100	No OMIM disease ID
MC2R	99,7	97,4	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM10	100	99,5	100	100	Immunodeficiency 80 with or without cardiomyopathy, 619313
MCM4	95,3	95	95,5	95,5	Immunodeficiency 54, 609981
MEFV	99,6	97,6	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
MPEG1	100	100	100	100	Immunodeficiency 77, 619223
MRE11	98,2	88,6	100	100	Ataxia-telangiectasia-like disorder 1, 604391
MRTFA	91	89	92,8	92,8	?Immunodeficiency 66, 618847
MS4A1	99,5	96,5	100	99,9	?Immunodeficiency, common variable, 5, 613495
MSN	98,2	92	100	100	Immunodeficiency 50, 300988
MTHFD1	99,9	98,4	100	100	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780

MVK	91,4	90,5	90,5	90,5	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYD88	100	99,5	100	100	Macroglobulinemia, Waldenstrom, somatic, 153600 Immunodeficiency 68, 612260
MYSM1	96,1	95,4	96,4	96,3	Bone marrow failure syndrome 4, 618116
NBAS	99,9	99,3	100	100	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NBN	99,2	97,8	100	99,9	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NCF1	26	25,7	100	99,9	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	99,8	97	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	99,9	Immunodeficiency 72 with autoinflammation, 618982
NCSTN	100	99,7	100	100	Acne inversa, familial, 1, 142690
NFAT5	99,9	98,7	100	100	No OMIM disease ID
NFATC1	100	100	100	99,9	No OMIM disease ID
NFE2L2	100	99,9	100	100	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFKB1	99,5	97,7	100	100	Immunodeficiency, common variable, 12, 616576
NFKB2	97,7	94,9	100	100	Immunodeficiency, common variable, 10, 615577
NFKBIA	92,4	83	100	100	Ectodermal dysplasia and immunodeficiency 2, 612132
NHEJ1	99,8	97,2	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	99,9	100	100	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050
NLRP1	99,3	97,2	100	100	?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225
NLRP12	100	100	100	100	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	100	99,9	100	100	CINCA syndrome, 607115 Familial cold inflammatory syndrome 1, 120100

					Keratoendothelitis fugax hereditaria, 148200 Deafness, autosomal dominant 34, with or without inflammation, 617772 Muckle-Wells syndrome, 191900
NLRP6	99,2	97,6	100	100	No OMIM disease ID
NLRP7	99,9	99	100	100	Hydatidiform mole, recurrent, 1, 231090
NOD2	100	99,9	100	99,9	Blau syndrome, 186580
NOP10	100	99,2	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NOS2	96,6	92,9	100	100	No OMIM disease ID
NRAS	100	100	100	100	Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Melanocytic nevus syndrome, congenital, somatic, 137550 Epidermal nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Colorectal cancer, somatic, 114500
NSMCE3	100	100	100	100	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
OAS1	100	100	100	100	No OMIM disease ID
ORAI1	99,3	97,1	99,4	96,7	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
OSTM1	98,7	92,9	100	100	Osteopetrosis, autosomal recessive 5, 259720
OTULIN	92,9	87	98,8	94,1	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
PARN	81,1	80,4	88,3	87,6	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX1	92,6	87,5	100	99,7	Otofaciocervical syndrome 2, 615560
PAX5	98,8	95,4	100	100	No OMIM disease ID
PBX1	100	99,1	100	100	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	98,9	93,4	100	100	Propionicacidemia, 606054
PCCB	96,7	95,4	99	96,2	Propionicacidemia, 606054
PDCD1	100	100	100	100	No OMIM disease ID
PEPD	100	99,4	100	100	Prolidase deficiency, 170100

PEX16	97,1	93,9	100	100	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PGM3	99,9	99,7	91,7	91,7	Immunodeficiency 23, 615816
PIGA	91,6	82,5	100	99,8	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIK3CD	99,3	97,6	100	100	Immunodeficiency 14A, autosomal dominant, 615513 Immunodeficiency 14B, autosomal recessive, 619281 ?Roifman-Chitayat syndrome, digenic, 613328
PIK3CG	100	100	100	100	No OMIM disease ID
PIK3R1	99,7	98,4	100	100	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PLCG2	100	99,3	100	100	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEKHM1	100	99,9	100	100	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLG	87,8	87,6	100	100	Dysplasminogenemia, 217090 Angioedema, hereditary, 4, 619360 Plasminogen deficiency, type I, 217090
PMM2	99,8	99,8	100	100	Congenital disorder of glycosylation, type Ia, 212065
PNP	99,8	98,7	100	100	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POLA1	99	93,6	100	99,7	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLE2	96,8	86,4	100	99,8	No OMIM disease ID
POMP	99,7	98,8	100	100	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POT1	99,5	98,5	100	100	No OMIM disease ID
POU2AF1	99,9	97,4	100	100	No OMIM disease ID
PRF1	91,2	90,1	100	100	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRKCD	100	99,9	100	100	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	99,2	96,9	100	100	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	86,4	86,3	100	99,7	Arts syndrome, 301835 Phosphoribosylpyrophosphate synthetase superactivity, 300661

					Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661
PSENEN	100	99,9	100	100	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMA3	98,7	92,4	100	100	No OMIM disease ID
PSMB4	100	100	100	100	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	99,8	97,5	100	100	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	99,5	95,4	100	100	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMG2	99,8	98,2	100	100	?Proteasome-associated autoinflammatory syndrome 4, 619183
PSTPIP1	100	99,2	100	100	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN22	99,6	95,2	100	100	No OMIM disease ID
PTPRC	98,8	93,9	100	99,9	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
RAB27A	99,5	99,5	100	99,9	Griscelli syndrome, type 2, 607624
RAC2	99,8	95,4	100	100	Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203 ?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986
RAG1	100	100	100	100	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889
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	49,5	49,1	100	100	No OMIM disease ID
RASGRP1	100	99,6	100	100	Immunodeficiency 64, 618534
RASGRP2	100	98,3	100	100	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	99,9	98,3	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	99,9	98,6	100	100	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
RELB	99	91,5	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	99,7	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	99,9	98,6	100	100	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGS10	95,9	91,7	100	100	No OMIM disease ID
RHOG	100	100	100	100	No OMIM disease ID
RHOH	100	100	100	100	No OMIM disease ID
RIPK1	99,8	98,5	100	100	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNASEH2A	100	99,7	100	100	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	81	78,2	91	90,9	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100	100	100	100	Aicardi-Goutieres syndrome 3, 610329
RNF168	99,9	99,6	100	100	RIDDLE syndrome, 611943
RNF31	100	98,6	100	100	No OMIM disease ID
RNU4ATAC	NC	NC	NC	NC	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RNU7-1	NC	NC	NC	NC	Aicardi-Goutieres syndrome 9, 619487
RORC	100	100	100	100	Immunodeficiency 42, 616622
RPSA	100	99,9	100	100	Asplenia, isolated congenital, 271400
RSPH9	99,7	96,3	100	100	Ciliary dyskinesia, primary, 12, 612650
RTEL1	99,7	97,2	100	100	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
SAMD9	99,9	99,8	100	100	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMD9L	100	99,9	100	100	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270

SAMHD1	98,5	97,9	100	100	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SASH3	99,8	94,9	100	100	No OMIM disease ID
SBDS	100	99,9	100	100	Shwachman-Diamond syndrome, 260400
SCIMP	80,4	79,9	97,8	89,1	No OMIM disease ID
SEC61A1	100	100	100	100	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056
SEMA3E	99,1	98,9	100	100	?CHARGE syndrome, 214800
SERAC1	99,6	99,5	100	99,9	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	99,6	96,4	100	100	Angioedema, hereditary, 1 and 2, 106100 Complement component 4, partial deficiency of, 120790
SH2B3	99	94,7	100	100	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SH2D1A	97,8	92,9	100	100	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	91,4	91,3	97,2	95,9	Cherubism, 118400
SH3KBP1	98,9	93,5	100	99,9	?Immunodeficiency 61, 300310
SKIV2L	100	99,5	100	100	Trichohepatoenteric syndrome 2, 614602
SLC29A3	100	99,5	100	100	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	99,7	99,3	100	100	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	100	99,4	100	100	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	99,8	97,6	100	100	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC39A4	99,3	96,3	100	100	Acrodermatitis enteropathica, 201100
SLC39A7	100	99,8	100	100	No OMIM disease ID
SLC46A1	100	98,5	100	100	Folate malabsorption, hereditary, 229050
SLC7A7	100	99,9	100	100	Lysinuric protein intolerance, 222700
SMARCAL1	100	99,8	100	100	Schimke immunoosseous dysplasia, 242900
SMARCD2	87	85,8	99,9	98,6	Specific granule deficiency 2, 617475
SNORA31	NC	NC	NC	NC	No OMIM disease ID
SNX10	96,2	95,9	99,9	99,3	Osteopetrosis, autosomal recessive 8, 615085

SOCS1	100	100	100	100	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375
SOCS4	99,9	99,3	100	99,9	No OMIM disease ID
SP110	100	100	100	100	Hepatic venoocclusive disease with immunodeficiency, 235550
SPINK5	99,8	99,6	100	99,9	Netherton syndrome, 256500
SPPL2A	84,3	70,5	100	99,7	No OMIM disease ID
SRP54	98	93,4	100	100	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	98	90,1	100	100	Bone marrow failure syndrome 1, 614675
STAT1	93,1	90,2	95,7	95	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	99,4	100	100	Pseudo-TORCH syndrome 3, 618886 Immunodeficiency 44, 616636
STAT3	99,9	99	100	100	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT4	99,8	99,4	100	100	No OMIM disease ID
STAT5B	99,9	98,1	100	100	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 Leukemia, acute promyelocytic, somatic, 102578
STAT6	100	99,3	100	100	No OMIM disease ID
STIM1	99,9	97,5	100	100	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STING1	99,7	96,3	100	100	STING-associated vasculopathy, infantile-onset, 615934
STK4	99,9	99,7	100	100	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	100	100	100	100	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	82,4	79,9	99,7	98	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
SYK	100	100	100	100	Immunodeficiency 82 with systemic inflammation, 619381
TAZ	99,3	93,7	100	100	Barth syndrome, 302060
TAP1	100	97,6	100	100	Bare lymphocyte syndrome, type I, 604571
TAP2	99,9	98,6	100	100	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,5	95,1	96,6	96,6	Bare lymphocyte syndrome, type I, 604571

TBX1	87,4	77,6	93,7	90,2	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX21	97,8	88	100	100	Asthma and nasal polyps, 208550
TCF3	98	93,8	100	100	Agammaglobulinemia 8, autosomal dominant, 616941
TCIRG1	98,5	93,4	100	100	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100	100	100	100	Transcobalamin II deficiency, 275350
TERC	NC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	97	94,8	100	100	No OMIM disease ID
TET2	100	100	100	100	Myelodysplastic syndrome, somatic, 614286 Immunodeficiency 75, 619126
TFRC	99,9	99,6	100	100	Immunodeficiency 46, 616740
TGFB1	100	98,8	100	100	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 Camurati-Engelmann disease, 131300
THBD	100	99,9	100	100	Thrombophilia due to thrombomodulin defect, 614486
TICAM1	100	98,8	100	100	No OMIM disease ID
TINF2	100	100	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TIRAP	100	100	100	100	No OMIM disease ID
TLR3	99,8	99	100	100	No OMIM disease ID
TLR4	100	99	100	100	No OMIM disease ID
TLR5	100	100	100	100	No OMIM disease ID
TLR7	100	99,7	100	100	Immunodeficiency 74, COVID19-related, X-linked, 301051
TLR8	99,9	99,8	100	100	No OMIM disease ID
TMC6	100	99,6	100	100	Epidermodysplasia verruciformis, 226400
TMC8	99,9	98,9	100	100	Epidermodysplasia verruciformis 2, 618231
TNFAIP3	100	99,9	100	100	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF11A	94,9	93,8	99,1	97,7	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810
TNFRSF13B	100	99,9	100	100	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529

TNFRSF13C	85	75,6	100	100	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	89,3	86,5	92,8	92,8	Periodic fever, familial, 142680
TNFRSF4	97,7	89	100	100	?Immunodeficiency 16, 615593
TNFRSF9	100	100	100	100	No OMIM disease ID
TNFSF11	100	100	100	100	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	99,5	95,3	100	100	No OMIM disease ID
TNFSF13	99	94,2	100	100	No OMIM disease ID
TOP2B	98,9	95,7	100	99,9	No OMIM disease ID
TPP2	99,2	95,1	100	100	Immunodeficiency 78 with autoimmunity and developmental delay, 619220
TRAC	100	100	100	100	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	100	99,2	100	100	No OMIM disease ID
TRAF3IP2	99,7	97,8	100	100	?Candidiasis, familial, 8, 615527
TREX1	100	100	100	100	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM22	100	99,9	100	100	No OMIM disease ID
TRNT1	99,7	97,4	100	99,9	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TTC37	99,7	98,8	100	100	Trichohepatoenteric syndrome 1, 222470
TTC7A	99,6	97,1	100	100	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	100	99,3	100	100	Immunodeficiency 35, 611521
UBA1	99,2	97,3	99,9	99,3	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
UNC13D	99,3	97,4	100	100	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	60,6	60	100	100	No OMIM disease ID
UNG	99,9	97,9	100	100	Immunodeficiency with hyper IgM, type 5, 608106
USB1	100	98,8	100	100	Poikiloderma with neutropenia, 604173
USP18	95,9	95,9	100	100	Pseudo-TORCH syndrome 2, 617397
VAV1	98,2	95,1	97,1	97,1	No OMIM disease ID
VPS13B	99,4	97,8	99,4	99,3	Cohen syndrome, 216550

VPS45	97,8	95,1	95,3	95,3	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	94,1	83,7	100	100	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WDR1	99,9	98,7	100	100	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550
WIPF1	99,9	98,5	100	100	Wiskott-Aldrich syndrome 2, 614493
WRAP53	100	100	100	99,9	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	93,1	88,3	99,9	99,6	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	100	99,7	100	100	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB24	100	100	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZNF341	97,3	95,9	100	100	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282
ZNFX1	100	99,7	100	100	No OMIM disease ID

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.
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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-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors