PRIMARY IMMUNODEFICIENCY GENE PANEL DG 3.4.0 (474 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ACD	100,0%	100,0%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ACP5	100,0%	100,0%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	100,0%	100,0%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ADA	100,0%	100,0%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	100,0%	100,0%	Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	100,0%	100,0%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAR	100,0%	100,0%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
AGA	100,0%	100,0%	Aspartylglucosaminuria, 208400
AICDA	100,0%	100,0%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	100,0%	100,0%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	100,0%	100,0%	Reticular dysgenesis, 267500
ALG13	100,0%	99,9%	Developmental and epileptic encephalopathy 36, 300884
ALPI	100,0%	100,0%	No OMIM Disease ID
ANGPT1	100,0%	100,0%	?Angioedema, hereditary, 5, 619361
AP1S3	90,5%	90,5%	No OMIM Disease ID
AP3B1	100,0%	100,0%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	100,0%	100,0%	?Hermansky-Pudlak syndrome 10, 617050
APOL1	100,0%	100,0%	No OMIM Disease ID
ARHGEF1	100,0%	100,0%	?Immunodeficiency 62, 618459
ARPC1B	100,0%	100,0%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ATG4A	100,0%	99,9%	No OMIM Disease ID
ATM	100,0%	100,0%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic,

			T-cell prolymphocytic leukemia, somatic, Lymphoma, mantle cell, somatic,
ATP6AP1	100,0%	100,0%	Immunodeficiency 47, 300972
B2M	100,0%	100,0%	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
BACH2	100,0%	100,0%	Immunodeficiency 60 and autoimmunity, 618394
BCL10	100,0%	100,0%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245
BCL11B	100,0%	99,9%	Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 Immunodeficiency 49, 617237
BLK	100,0%	100,0%	Maturity-onset diabetes of the young, type 11, 613375
BLM	100,0%	100,0%	Bloom syndrome, 210900
BLNK	100,0%	100,0%	?Agammaglobulinemia 4, 613502
BLOC1S6	100,0%	100,0%	?Hermansky-Pudlak syndrome 9, 614171
BTK	100,0%	100,0%	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
C1QA	100,0%	100,0%	C1q deficiency, 613652
C1QB	100,0%	100,0%	C1q deficiency, 613652
C1QC	100,0%	100,0%	C1q deficiency, 613652
C1R	100,0%	99,1%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	99,9%	99,2%	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	100,0%	100,0%	C2 deficiency, 217000
C2orf69	100,0%	100,0%	Combined oxidative phosphorylation deficiency 53, 619423
C3	100,0%	100,0%	C3 deficiency, 613779
C5	100,0%	100,0%	C5 deficiency, 609536
C6	100,0%	100,0%	C6 deficiency, 612446 Combined C6/C7 deficiency,
C7	100,0%	100,0%	C7 deficiency, 610102
C8A	100,0%	100,0%	C8 deficiency, type I, 613790
C8B	100,0%	100,0%	C8 deficiency, type II, 613789
C8G	100,0%	100,0%	No OMIM Disease ID
C9	100,0%	100,0%	C9 deficiency, 613825
CA2	100,0%	100,0%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CARD10	100,0%	100,0%	?Immunodeficiency 89 and autoimmunity, 619632

CARD11	100,0%	100,0%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11B with atopic dermatitis, 617638 Immunodeficiency 11A, 615206
CARD14	100,0%	100,0%	Psoriasis 2, 602723 Pityriasis rubra pilaris, 173200
CARD9	100,0%	100,0%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	100,0%	100,0%	Immunodeficiency 58, 618131
CASP10	100,0%	100,0%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP8	95,6%	95,6%	Hepatocellular carcinoma, somatic, 114550 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271
CAVIN1	100,0%	100,0%	Lipodystrophy, congenital generalized, type 4, 613327
CCBE1	100,0%	100,0%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD19	100,0%	100,0%	Immunodeficiency, common variable, 3, 613493
CD247	100,0%	100,0%	?Immunodeficiency 25, 610163
CD27	100,0%	100,0%	Lymphoproliferative syndrome 2, 615122
CD28	100,0%	100,0%	No OMIM Disease ID
CD3D	100,0%	100,0%	Immunodeficiency 19, 615617
CD3E	100,0%	100,0%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	100,0%	100,0%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	100,0%	100,0%	Immunodeficiency 79, 619238 OKT4 epitope deficiency, 613949
CD40	100,0%	100,0%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	100,0%	100,0%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	100,0%	100,0%	No OMIM Disease ID
CD48	100,0%	100,0%	No OMIM Disease ID
CD55	95,9%	93,9%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	64,5%	64,5%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	100,0%	100,0%	Lymphoproliferative syndrome 3, 618261
CD79A	100,0%	100,0%	Agammaglobulinemia 3, 613501
CD79B	100,0%	100,0%	Agammaglobulinemia 6, 612692
CD81	100,0%	100,0%	Immunodeficiency, common variable, 6, 613496
CD8A	100,0%	100,0%	CD8 deficiency, familial, 608957
CDC42	100,0%	100,0%	Takenouchi-Kosaki syndrome, 616737
CDCA7	100,0%	100,0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910

CDKN2B	100,0%	100,0%	No OMIM Disease ID
CEBPE	100,0%	100,0%	Specific granule deficiency, 245480
CFB	100,0%	100,0%	?Complement factor B deficiency, 615561
CFD	100,0%	100,0%	Complement factor D deficiency, 613912
CFH	100,0%	100,0%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFI	100,0%	100,0%	Complement factor I deficiency, 610984
CFP	100,0%	100,0%	Properdin deficiency, X-linked, 312060
CFTR	100,0%	100,0%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
CHD7	100,0%	100,0%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CIB1	100,0%	100,0%	Epidermodysplasia verruciformis 3, 618267
CIITA	100,0%	100,0%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CLCN7	100,0%	100,0%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
CLEC4D	100,0%	100,0%	No OMIM Disease ID
CLEC7A	100,0%	100,0%	Candidiasis, familial, 4, autosomal recessive, 613108
CLPB	100,0%	100,0%	Neutropenia, severe congenital, 9, autosomal dominant, 619813 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
COLEC11	100,0%	100,0%	3MC syndrome 2, 265050
COPA	100,0%	100,0%	No OMIM Disease ID
CORO1A	100,0%	100,0%	Immunodeficiency 8, 615401
CR2	100,0%	100,0%	Immunodeficiency, common variable, 7, 614699
CRACR2A	100,0%	100,0%	No OMIM Disease ID
CREBBP	100,0%	100,0%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CSF2RA	96,0%	92,4%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	100,0%	100,0%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	100,0%	100,0%	Neutropenia, severe congenital, 7, autosomal recessive, 617014 ?Neutrophilia, hereditary, 162830
CTC1	100,0%	100,0%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100,0%	100,0%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CTNNBL1	100,0%	100,0%	No OMIM Disease ID

CTPS1	93,0%	93,0%	Immunodeficiency 24, 615897
CTSC	100,0%	100,0%	Periodontitis 1, juvenile, 170650 Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000
CXCR4	100,0%	100,0%	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670
CYBA	100,0%	100,0%	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBB	100,0%	100,0%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400
CYBC1	100,0%	100,0%	Chronic granulomatous disease 5, autosomal recessive, 618935
DBF4	100,0%	100,0%	No OMIM Disease ID
DBR1	100,0%	100,0%	No OMIM Disease ID
DCLRE1C	100,0%	100,0%	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DDX41	100,0%	100,0%	No OMIM Disease ID
DDX58	100,0%	100,0%	Singleton-Merten syndrome 2, 616298
DEF6	100,0%	100,0%	Immunodeficiency 87 and autoimmunity, 619573
DHFR	100,0%	100,0%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DIAPH1	100,0%	100,0%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DKC1	100,0%	100,0%	Dyskeratosis congenita, X-linked, 305000
DNASE1	100,0%	100,0%	No OMIM Disease ID
DNASE1L3	100,0%	100,0%	Systemic lupus erythematosus 16, 614420
DNASE2	100,0%	100,0%	No OMIM Disease ID
DNMT3B	100,0%	100,0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK2	100,0%	100,0%	Immunodeficiency 40, 616433
DOCK8	100,0%	100,0%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	100,0%	100,0%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	100,0%	100,0%	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074
EPG5	100,0%	100,0%	Vici syndrome, 242840
ERCC2	100,0%	100,0%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100,0%	100,0%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651

EXTL3	100,0%	100,0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
F12	100,0%	100,0%	Angioedema, hereditary, 3, 610618 Factor XII deficiency, 234000
FAAP24	100,0%	100,0%	No OMIM Disease ID
FADD	100,0%	100,0%	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759
FAS	100,0%	100,0%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic,
FASLG	100,0%	100,0%	Autoimmune lymphoproliferative syndrome, type IB, 601859
FAT4	100,0%	100,0%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBXW11	100,0%	100,0%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FCGR3A	100,0%	100,0%	Immunodeficiency 20, 615707
FCHO1	100,0%	100,0%	Immunodeficiency 76, 619164
FCN3	100,0%	100,0%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT1	100,0%	100,0%	Kindler syndrome, 173650
FERMT3	100,0%	100,0%	Leukocyte adhesion deficiency, type III, 612840
FNIP1	100,0%	100,0%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOXN1	100,0%	100,0%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	100,0%	100,0%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FPR1	100,0%	100,0%	No OMIM Disease ID
G6PC	100,0%	100,0%	Glycogen storage disease Ia, 232200
G6PC3	100,0%	100,0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	100,0%	100,0%	Hemolytic anemia, G6PD deficient (favism), 300908
GATA2	100,0%	100,0%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GFI1	100,0%	100,0%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GIMAP5	100,0%	100,0%	Portal hypertension, noncirrhotic, 2, 619463
GINS1	100,0%	100,0%	Immunodeficiency 55, 617827
GJC2	99,9%	99,5%	Lymphatic malformation 3, 613480 ?Spastic paraplegia 44, autosomal recessive, 613206 Leukodystrophy, hypomyelinating, 2, 608804
GRHL2	100,0%	100,0%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031

GTF2H5	72,5%	72,5%	Trichothiodystrophy 3, photosensitive, 616395
HAVCR2	100,0%	100,0%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100,0%	100,0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HELLS	100,0%	100,0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HMOX1	100,0%	100,0%	Heme oxygenase-1 deficiency, 614034
HS3ST6	100,0%	99,6%	?Angioedema, hereditary, 8, 619367
HYOU1	100,0%	100,0%	?Immunodeficiency 59 and hypoglycemia, 233600
ICOS	100,0%	100,0%	Immunodeficiency, common variable, 1, 607594
ICOSLG	100,0%	100,0%	No OMIM Disease ID
IFIH1	100,0%	100,0%	Immunodeficiency 95, 619773 Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNAR1	97,8%	97,8%	No OMIM Disease ID
IFNAR2	100,0%	100,0%	?Immunodeficiency 45, 616669
IFNG	100,0%	100,0%	?Immunodeficiency 69, mycobacteriosis, 618963
IFNGR1	100,0%	100,0%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978
IFNGR2	100,0%	100,0%	Immunodeficiency 28, mycobacteriosis, 614889
IGHM	100,0%	100,0%	Agammaglobulinemia 1, 601495
IGLL1	100,0%	100,0%	Agammaglobulinemia 2, 613500
IKBKB	100,0%	100,0%	Immunodeficiency 15B, 615592 Immunodeficiency 15A, 618204
IKBKG	100,0%	100,0%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636
IKZF1	100,0%	100,0%	Immunodeficiency, common variable, 13, 616873
IKZF3	100,0%	100,0%	?Immunodeficiency 84, 619437
IL10	100,0%	100,0%	No OMIM Disease ID
IL10RA	100,0%	100,0%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	100,0%	100,0%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL12B	100,0%	100,0%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	94,1%	94,1%	Immunodeficiency 30, 614891
IL17F	100,0%	100,0%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100,0%	100,0%	Immunodeficiency 51, 613953
IL17RC	100,0%	100,0%	Candidiasis, familial, 9, 616445
IL18BP	100,0%	100,0%	No OMIM Disease ID
IL1RN	100,0%	100,0%	Interleukin 1 receptor antagonist deficiency, 612852

IL2	100,0%	100,0%	No OMIM Disease ID
IL21	100,0%	100,0%	?Immunodeficiency, common variable, 11, 615767
IL21R	100,0%	100,0%	Immunodeficiency 56, 615207
IL2RA	100,0%	100,0%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL2RB	100,0%	100,0%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL2RG	100,0%	100,0%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL36RN	100,0%	100,0%	Psoriasis 14, pustular, 614204
IL6R	92,7%	92,7%	Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944
IL6ST	100,0%	100,0%	Stuve-Wiedemann syndrome 2, 619751 Hyper-IgE recurrent infection syndrome 4A, autosomal dominant, 619752 ?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750 Hyper-IgE recurrent infection syndrome 4B, autosomal recessive, 618523
IL7R	100,0%	100,0%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INO80	100,0%	100,0%	No OMIM Disease ID
INSR	100,0%	100,0%	Rabson-Mendenhall syndrome, 262190 Leprechaunism, 246200 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
IPO8	100,0%	100,0%	VISS syndrome, 619472
IRAK1	100,0%	100,0%	No OMIM Disease ID
IRAK4	100,0%	100,0%	Immunodeficiency 67, 607676
IRF2BP2	100,0%	100,0%	?Immunodeficiency, common variable, 14, 617765
IRF3	100,0%	100,0%	No OMIM Disease ID
IRF4	100,0%	100,0%	No OMIM Disease ID
IRF7	100,0%	100,0%	?Immunodeficiency 39, 616345
IRF8	100,0%	100,0%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRF9	100,0%	100,0%	Immunodeficiency 65, susceptibility to viral infections, 618648
IRGM	100,0%	100,0%	No OMIM Disease ID
ISG15	100,0%	100,0%	Immunodeficiency 38, 616126
ITCH	95,6%	93,9%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGB2	97,2%	97,2%	Leukocyte adhesion deficiency, 116920
ITK	100,0%	100,0%	Lymphoproliferative syndrome 1, 613011
ITPKB	100,0%	100,0%	No OMIM Disease ID
ITPR3	100,0%	100,0%	No OMIM Disease ID
IVNS1ABP	100,0%	100,0%	Immunodeficiency 70, 618969

JAGN1	100,0%	99,8%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	100,0%	100,0%	Autoinflammation, immune dysregulation, and eosinophilia, 618999
JAK2	100,0%	100,0%	Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300
JAK3	100,0%	100,0%	SCID, autosomal recessive, T-negative/B-positive type, 600802
KDM6A	100,0%	100,0%	Kabuki syndrome 2, 300867
KMT2D	100,0%	100,0%	Kabuki syndrome 1, 147920
KNG1	100,0%	100,0%	Angioedema, hereditary, 6, 619363
KRAS	100,0%	100,0%	Gastric cancer, somatic, 613659 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	100,0%	100,0%	Juvenile arthritis, 618795
LAMTOR2	100,0%	100,0%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAT	100,0%	100,0%	Immunodeficiency 52, 617514
LCK	100,0%	100,0%	?Immunodeficiency 22, 615758
LCP2	100,0%	100,0%	?Immunodeficiency 81, 619374
LIG1	100,0%	100,0%	Immunodeficiency 96, 619774
LIG4	100,0%	100,0%	LIG4 syndrome, 606593
LPIN2	100,0%	100,0%	Majeed syndrome, 609628
LRBA	100,0%	100,0%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC32	100,0%	100,0%	Cleft palate, proliferative retinopathy, and developmental delay, 619074
LRRC8A	100,0%	100,0%	?Agammaglobulinemia 5, 613506
LSM11	100,0%	100,0%	?Aicardi-Goutieres syndrome 8, 619486
LYST	100,0%	100,0%	Chediak-Higashi syndrome, 214500

MAGT1	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	100,0%	100,0%	No OMIM Disease ID
MALT1	100,0%	100,0%	Immunodeficiency 12, 615468
MAN2B1	100,0%	100,0%	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	100,0%	100,0%	No OMIM Disease ID
MANBA	100,0%	100,0%	Mannosidosis, beta, 248510
MAP1LC3B2	100,0%	100,0%	No OMIM Disease ID
MAP3K14	100,0%	100,0%	No OMIM Disease ID
MAPK8	100,0%	100,0%	No OMIM Disease ID
MASP2	100,0%	100,0%	MASP2 deficiency, 613791
MBL2	100,0%	100,0%	No OMIM Disease ID
MC2R	100,0%	100,0%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM10	100,0%	100,0%	Immunodeficiency 80 with or without cardiomyopathy, 619313
MCM4	95,5%	95,5%	Immunodeficiency 54, 609981
MEFV	96,4%	96,4%	Neutrophilic dermatosis, acute febrile, 608068 Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MOGS	100,0%	100,0%	Congenital disorder of glycosylation, type IIb, 606056
MPEG1	100,0%	100,0%	Immunodeficiency 77, 619223
MRTFA	92,8%	92,8%	?Immunodeficiency 66, 618847
MS4A1	100,0%	100,0%	?Immunodeficiency, common variable, 5, 613495
MSN	100,0%	100,0%	Immunodeficiency 50, 300988
MTHFD1	100,0%	100,0%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MVK	90,5%	90,5%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYD88	100,0%	100,0%	Macroglobulinemia, Waldenstrom, somatic, 153600 Immunodeficiency 68, 612260
MYOF	100,0%	100,0%	?Angioedema, hereditary, 7, 619366
MYSM1	96,4%	96,4%	Bone marrow failure syndrome 4, 618116
NBAS	100,0%	100,0%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NBN	100,0%	100,0%	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NCF1	100,0%	100,0%	Chronic granulomatous disease 1, autosomal recessive, 233700

NCF2	100,0%	100,0%	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100,0%	100,0%	Chronic granulomatous disease 3, autosomal recessive, 613960
NCKAP1L	100,0%	100,0%	Immunodeficiency 72 with autoinflammation, 618982
NCSTN	100,0%	100,0%	Acne inversa, familial, 1, 142690
NFAT5	100,0%	100,0%	No OMIM Disease ID
NFATC1	100,0%	100,0%	No OMIM Disease ID
NFE2L2	100,0%	100,0%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFKB1	100,0%	100,0%	Immunodeficiency, common variable, 12, 616576
NFKB2	100,0%	100,0%	Immunodeficiency, common variable, 10, 615577
NFKBIA	100,0%	100,0%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHEJ1	100,0%	100,0%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 2, 613987
NLRC4	100,0%	100,0%	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050
NLRP1	100,0%	100,0%	?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225
NLRP12	100,0%	100,0%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	100,0%	100,0%	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	100,0%	100,0%	No OMIM Disease ID
NLRP7	100,0%	100,0%	Hydatidiform mole, recurrent, 1, 231090
NOD2	100,0%	100,0%	Blau syndrome, 186580
NOP10	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOS2	100,0%	100,0%	No OMIM Disease ID
NRAS	100,0%	100,0%	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	100,0%	100,0%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241

OAS1	100,0%	100,0%	No OMIM Disease ID
ORAI1	100,0%	100,0%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
OSTM1	100,0%	100,0%	Osteopetrosis, autosomal recessive 5, 259720
OTULIN	100,0%	100,0%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
PARN	89,5%	87,8%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX1	100,0%	100,0%	Otofaciocervical syndrome 2, 615560
PAX5	100,0%	100,0%	No OMIM Disease ID
PBX1	100,0%	100,0%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	100,0%	100,0%	Propionicacidemia, 606054
PCCB	99,9%	98,1%	Propionicacidemia, 606054
PDCD1	100,0%	100,0%	No OMIM Disease ID
PEPD	100,0%	100,0%	Prolidase deficiency, 170100
PEX16	100,0%	100,0%	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PGM3	91,7%	91,7%	Immunodeficiency 23, 615816
PIGA	100,0%	100,0%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIK3CD	100,0%	100,0%	Immunodeficiency 14A, autosomal dominant, 615513 Immunodeficiency 14B, autosomal recessive, 619281 ?Roifman-Chitayat syndrome, digenic, 613328
PIK3CG	100,0%	100,0%	Immunodeficiency 97 with autoinflammation, 619802
PIK3R1	100,0%	100,0%	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PLCG2	100,0%	100,0%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEKHM1	100,0%	100,0%	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLG	100,0%	100,0%	Dysplasminogenemia, 217090 Angioedema, hereditary, 4, 619360 Plasminogen deficiency, type I, 217090
PMM2	100,0%	100,0%	Congenital disorder of glycosylation, type Ia, 212065
PNP	100,0%	100,0%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179

POLA1	100,0%	100,0%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLD1	100,0%	100,0%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE2	100,0%	100,0%	No OMIM Disease ID
POMP	100,0%	100,0%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POT1	100,0%	100,0%	No OMIM Disease ID
POU2AF1	100,0%	100,0%	No OMIM Disease ID
PRF1	100,0%	100,0%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRKCD	100,0%	100,0%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	100,0%	100,0%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	100,0%	100,0%	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	100,0%	100,0%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMA3	100,0%	100,0%	No OMIM Disease ID
PSMB4	100,0%	100,0%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	100,0%	100,0%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	100,0%	100,0%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMG2	100,0%	100,0%	?Proteasome-associated autoinflammatory syndrome 4, 619183
PSTPIP1	100,0%	100,0%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN22	100,0%	100,0%	No OMIM Disease ID
PTPRC	100,0%	100,0%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
RAB27A	100,0%	100,0%	Griscelli syndrome, type 2, 607624
RAC2	100,0%	100,0%	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	100,0%	100,0%	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	100,0%	100,0%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RANBP2	100,0%	100,0%	No OMIM Disease ID
RASGRP1	100,0%	100,0%	Immunodeficiency 64, 618534
RASGRP2	100,0%	100,0%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	100,0%	100,0%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RC3H1	100,0%	100,0%	?Immune dysregulation and systemic hyperinflammation syndrome, 618998
RECQL4	100,0%	100,0%	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
REL	99,9%	98,3%	Immunodeficiency 92, 619652
RELA	100,0%	100,0%	?Mucocutaneous ulceration, chronic, 618287
RELB	100,0%	100,0%	?Immunodeficiency 53, 617585
RFX5	100,0%	100,0%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	100,0%	100,0%	MHC class II deficiency, complementation group B, 209920
RFXAP	100,0%	100,0%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGS10	100,0%	100,0%	No OMIM Disease ID
RHOG	100,0%	100,0%	No OMIM Disease ID
RHOH	100,0%	100,0%	No OMIM Disease ID
RIPK1	100,0%	100,0%	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852
RMRP	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNASEH2A	100,0%	100,0%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91,0%	91,0%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100,0%	100,0%	Aicardi-Goutieres syndrome 3, 610329
RNF168	100,0%	100,0%	RIDDLE syndrome, 611943
RNF31	100,0%	100,0%	No OMIM Disease ID
RNU4ATAC	NC	NC	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RNU7-1	NC	NC	Aicardi-Goutieres syndrome 9, 619487
RORC	100,0%	100,0%	Immunodeficiency 42, 616622
RPA1	100,0%	100,0%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 6, 619767

RPSA	100,0%	100,0%	Asplenia, isolated congenital, 271400
RSPH9	100,0%	100,0%	Ciliary dyskinesia, primary, 12, 612650
RTEL1	100,0%	100,0%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
SAMD9	100,0%	100,0%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMD9L	100,0%	100,0%	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Spinocerebellar ataxia 49, 619806
SAMHD1	100,0%	100,0%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SASH3	100,0%	100,0%	No OMIM Disease ID
SBDS	100,0%	100,0%	Shwachman-Diamond syndrome, 260400
SEC61A1	100,0%	100,0%	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056
SEMA3E	100,0%	100,0%	?CHARGE syndrome, 214800
SERAC1	100,0%	100,0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	100,0%	100,0%	Angioedema, hereditary, 1 and 2, 106100 Complement component 4, partial deficiency of, 120790
SH2B3	100,0%	100,0%	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SH2D1A	100,0%	100,0%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	99,9%	99,4%	Cherubism, 118400
SH3KBP1	100,0%	100,0%	?Immunodeficiency 61, 300310
SKIV2L	100,0%	100,0%	Trichohepatoenteric syndrome 2, 614602
SLC29A3	100,0%	100,0%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	100,0%	100,0%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	100,0%	100,0%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	100,0%	100,0%	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC39A4	100,0%	100,0%	Acrodermatitis enteropathica, 201100
SLC39A7	100,0%	100,0%	Agammaglobulinemia 9, autosomal recessive, 619693
SLC46A1	100,0%	100,0%	Folate malabsorption, hereditary, 229050
SLC7A7	100,0%	100,0%	Lysinuric protein intolerance, 222700

SMARCAL1	100,0%	100,0%	Schimke immunoosseous dysplasia, 242900
SMARCD2	100,0%	100,0%	Specific granule deficiency 2, 617475
SNORA31	NC	NC	No OMIM Disease ID
SNX10	100,0%	99,9%	Osteopetrosis, autosomal recessive 8, 615085
SOCS1	100,0%	100,0%	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375
SOCS4	100,0%	100,0%	No OMIM Disease ID
SP110	100,0%	100,0%	Hepatic venoocclusive disease with immunodeficiency, 235550
SPINK5	100,0%	100,0%	Netherton syndrome, 256500
SPPL2A	100,0%	100,0%	Immunodeficiency 86, mycobacteriosis, 619549
SRP54	100,0%	100,0%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	100,0%	100,0%	Bone marrow failure syndrome 1, 614675
STAT1	95,6%	95,5%	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796
STAT2	100,0%	100,0%	Pseudo-TORCH syndrome 3, 618886 Immunodeficiency 44, 616636
STAT3	100,0%	100,0%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT4	100,0%	100,0%	No OMIM Disease ID
STAT5B	100,0%	100,0%	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	100,0%	100,0%	No OMIM Disease ID
STIM1	100,0%	100,0%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STING1	100,0%	100,0%	STING-associated vasculopathy, infantile-onset, 615934
STK4	100,0%	100,0%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	100,0%	100,0%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	99,8%	98,7%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
SYK	100,0%	100,0%	Immunodeficiency 82 with systemic inflammation, 619381
TAZ	100,0%	100,0%	Barth syndrome, 302060
TAP1	100,0%	100,0%	Bare lymphocyte syndrome, type I, 604571
TAP2	100,0%	100,0%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,6%	96,6%	Bare lymphocyte syndrome, type I, 604571
TBX1	98,1%	95,9%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400

			Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX21	100,0%	100,0%	Asthma and nasal polyps, 208550 ?Immunodeficiency 88, 619630
TCF3	100,0%	100,0%	Agammaglobulinemia 8B, autosomal recessive, 619824 Agammaglobulinemia 8A, autosomal dominant, 616941
TCIRG1	100,0%	100,0%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100,0%	100,0%	Transcobalamin II deficiency, 275350
TERC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742
TET2	100,0%	100,0%	Myelodysplastic syndrome, somatic, 614286 Immunodeficiency 75, 619126
TFRC	100,0%	100,0%	Immunodeficiency 46, 616740
TGFB1	100,0%	100,0%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 Camurati-Engelmann disease, 131300
THBD	100,0%	100,0%	Thrombophilia 12 due to thrombomodulin defect, 614486
TICAM1	100,0%	100,0%	No OMIM Disease ID
TINF2	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TIRAP	100,0%	100,0%	No OMIM Disease ID
TLR3	100,0%	100,0%	No OMIM Disease ID
TLR4	100,0%	100,0%	No OMIM Disease ID
TLR5	100,0%	100,0%	No OMIM Disease ID
TLR7	100,0%	100,0%	Immunodeficiency 74, COVID19-related, X-linked, 301051
TLR8	100,0%	100,0%	No OMIM Disease ID
TMC6	100,0%	100,0%	Epidermodysplasia verruciformis, 226400
TMC8	100,0%	100,0%	Epidermodysplasia verruciformis 2, 618231
TNFAIP3	100,0%	100,0%	Autoinflammatory syndrome, familial, Behcet-like 1, 616744
TNFRSF11A	100,0%	99,7%	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810
TNFRSF13B	100,0%	100,0%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	100,0%	100,0%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	92,8%	92,8%	Periodic fever, familial, 142680
TNFRSF4	100,0%	100,0%	?Immunodeficiency 16, 615593

TNFRSF9	100,0%	100,0%	No OMIM Disease ID
TNFSF11	100,0%	100,0%	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	100,0%	100,0%	No OMIM Disease ID
TNFSF13	100,0%	100,0%	No OMIM Disease ID
TOM1	100,0%	100,0%	?Immunodeficiency 85 and autoimmunity, 619510
TOP2B	100,0%	100,0%	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296
TPP2	100,0%	100,0%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220
TRAC	100,0%	100,0%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	100,0%	100,0%	No OMIM Disease ID
TRAF3IP2	100,0%	100,0%	?Candidiasis, familial, 8, 615527
TREX1	100,0%	100,0%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM22	100,0%	100,0%	No OMIM Disease ID
TRNT1	100,0%	100,0%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TTC37	100,0%	100,0%	Trichohepatoenteric syndrome 1, 222470
TTC7A	100,0%	100,0%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	100,0%	100,0%	Immunodeficiency 35, 611521
UBA1	100,0%	99,8%	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
UNC13D	100,0%	100,0%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	100,0%	100,0%	No OMIM Disease ID
UNG	100,0%	100,0%	Immunodeficiency with hyper IgM, type 5, 608106
USB1	100,0%	100,0%	Poikiloderma with neutropenia, 604173
USP18	100,0%	100,0%	Pseudo-TORCH syndrome 2, 617397
VAV1	97,1%	97,1%	No OMIM Disease ID
VPS13B	99,5%	99,4%	Cohen syndrome, 216550
VPS45	95,3%	95,3%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	100,0%	100,0%	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WDR1	100,0%	100,0%	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550
WIPF1	100,0%	100,0%	Wiskott-Aldrich syndrome 2, 614493
WRAP53	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	100,0%	100,0%	Lymphoproliferative syndrome, X-linked, 2, 300635

ZAP70	100,0%	100,0%	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB24	100,0%	100,0%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZNF341	100,0%	100,0%	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282
ZNFX1	100,0%	100,0%	Immunodeficiency 91 and hyperinflammation, 619644

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.
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.
TWIST is the chemistry used for 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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors