PRIMARY IMMUNODEFICIENCY GENE PANEL DG 3.3.0 (482 genes)

Releasedate: 13-01-2022

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

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

CARD11	100%	100%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11B with atopic dermatitis, 617638 Immunodeficiency 11A, 615206
CARD14	100%	100%	Psoriasis 2, 602723 Pityriasis rubra pilaris, 173200
CARD9	100%	100%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	100%	100%	Immunodeficiency 58, 618131
CASP10	100%	100%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP8	95%	95%	Hepatocellular carcinoma, somatic, 114550 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271
CAVIN1	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
CCBE1	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD19	100%	100%	Immunodeficiency, common variable, 3, 613493
CD247	100%	100%	?Immunodeficiency 25, 610163
CD27	100%	100%	Lymphoproliferative syndrome 2, 615122
CD28	100%	100%	No OMIM disease ID
CD3D	100%	100%	Immunodeficiency 19, 615617
CD3E	100%	100%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	100%	100%	Immunodeficiency 79, 619238 OKT4 epitope deficiency, 613949
CD40	100%	100%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	100%	100%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	100%	100%	No OMIM disease ID
CD48	100%	100%	No OMIM disease ID
CD55	96%	94%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	64%	64%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	100%	100%	Lymphoproliferative syndrome 3, 618261
CD79A	100%	100%	Agammaglobulinemia 3, 613501
CD79B	100%	100%	Agammaglobulinemia 6, 612692
CD81	100%	100%	Immunodeficiency, common variable, 6, 613496
CD8A	100%	100%	CD8 deficiency, familial, 608957
CDC42	100%	100%	Takenouchi-Kosaki syndrome, 616737
CDCA7	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910

CDKN2B	100%	100%	No OMIM disease ID
CEBPE	100%	100%	Specific granule deficiency, 245480
CFB	100%	100%	?Complement factor B deficiency, 615561
CFD	100%	100%	Complement factor D deficiency, 613912
CFH	100%	100%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	96%	94%	No OMIM disease ID
CFHR2	76%	76%	No OMIM disease ID
CFHR3	96%	95%	No OMIM disease ID
CFHR4	100%	100%	No OMIM disease ID
CFHR5	100%	100%	Nephropathy due to CFHR5 deficiency, 614809
CFI	100%	100%	Complement factor I deficiency, 610984
CFP	100%	100%	Properdin deficiency, X-linked, 312060
CFTR	100%	100%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
CHD7	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CIB1	100%	100%	Epidermodysplasia verruciformis 3, 618267
CIITA	100%	100%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CLCN7	100%	100%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
CLEC4D	100%	100%	No OMIM disease ID
CLEC7A	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108
CLPB	100%	100%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COLEC11	100%	100%	3MC syndrome 2, 265050
COPA	100%	100%	No OMIM disease ID
CORO1A	100%	100%	Immunodeficiency 8, 615401
CR2	100%	100%	Immunodeficiency, common variable, 7, 614699
CREBBP	100%	100%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CSF2RA	95%	91%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	100%	100%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	100%	100%	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100%	100%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100

CTNNBL1	100%	100%	No OMIM disease ID
CTPS1	93%	93%	Immunodeficiency 24, 615897
CTSC	100%	100%	Periodontitis 1, juvenile, 170650 Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000
CXCR4	100%	100%	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670
CYBA	100%	100%	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBB	100%	100%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400
CYBC1	100%	100%	Chronic granulomatous disease 5, autosomal recessive, 618935
DBF4	100%	100%	No OMIM disease ID
DBR1	100%	100%	No OMIM disease ID
DCLRE1C	100%	100%	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DDX41	100%	100%	No OMIM disease ID
DDX58	100%	100%	Singleton-Merten syndrome 2, 616298
DEF6	100%	100%	Immunodeficiency 87 and autoimmunity, 619573
DHFR	100%	100%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DIAPH1	100%	99%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DKC1	100%	100%	Dyskeratosis congenita, X-linked, 305000
DNASE1	100%	100%	No OMIM disease ID
DNASE1L3	100%	100%	Systemic lupus erythematosus 16, 614420
DNASE2	100%	100%	No OMIM disease ID
DNMT3B	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK2	100%	100%	Immunodeficiency 40, 616433
DOCK8	100%	100%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	100%	100%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	100%	100%	No OMIM disease ID
EPG5	100%	100%	Vici syndrome, 242840
ERCC2	100%	100%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756

ERCC3	100%	100%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
EXTL3	100%	100%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
F12	100%	100%	Angioedema, hereditary, 3, 610618 Factor XII deficiency, 234000
FAAP24	100%	100%	No OMIM disease ID
FADD	100%	100%	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759
FAS	100%	100%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic,
FASLG	100%	100%	Autoimmune lymphoproliferative syndrome, type IB, 601859
FAT4	100%	100%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBXW11	100%	100%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FCGR1A	100%	100%	No OMIM disease ID
FCGR2A	100%	100%	No OMIM disease ID
FCGR2B	100%	100%	No OMIM disease ID
FCGR3A	100%	100%	Immunodeficiency 20, 615707
FCGR3B	98%	97%	No OMIM disease ID
FCHO1	100%	100%	Immunodeficiency 76, 619164
FCN3	100%	100%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	100%	100%	Leukocyte adhesion deficiency, type III, 612840
FNIP1	100%	100%	No OMIM disease ID
FOXN1	100%	100%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	100%	100%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FPR1	100%	100%	No OMIM disease ID
G6PC1	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	100%	100%	Hemolytic anemia, G6PD deficient (favism), 300908
GATA2	100%	100%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GFI1	100%	100%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GIMAP5	100%	100%	Portal hypertension, noncirrhotic, 2, 619463
GINS1	100%	100%	Immunodeficiency 55, 617827

GJC2	99%	98%	Lymphatic malformation 3, 613480 ?Spastic paraplegia 44, autosomal recessive, 613206 Leukodystrophy, hypomyelinating, 2, 608804
GRHL2	100%	100%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
GTF2H5	72%	72%	Trichothiodystrophy 3, photosensitive, 616395
HAVCR2	100%	100%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HELLS	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HMOX1	100%	100%	Heme oxygenase-1 deficiency, 614034
HS3ST6	99%	98%	?Angioedema, hereditary, 8, 619367
HYOU1	100%	100%	?Immunodeficiency 59 and hypoglycemia, 233600
ICOS	100%	100%	Immunodeficiency, common variable, 1, 607594
ICOSLG	100%	100%	No OMIM disease ID
IFIH1	100%	100%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNAR1	97%	97%	No OMIM disease ID
IFNAR2	100%	100%	?Immunodeficiency 45, 616669
IFNG	100%	100%	?Immunodeficiency 69, mycobacteriosis, 618963
IFNGR1	100%	100%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978
IFNGR2	100%	100%	Immunodeficiency 28, mycobacteriosis, 614889
IGHM	100%	100%	Agammaglobulinemia 1, 601495
IGLL1	100%	100%	Agammaglobulinemia 2, 613500
IKBKB	100%	100%	Immunodeficiency 15B, 615592 Immunodeficiency 15A, 618204
IKBKG	100%	100%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636
IKZF1	100%	100%	Immunodeficiency, common variable, 13, 616873
IKZF3	100%	100%	?Immunodeficiency 84, 619437
IL10	100%	100%	No OMIM disease ID
IL10RA	100%	100%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	100%	100%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL12B	100%	100%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	94%	94%	Immunodeficiency 30, 614891

IL17F	100%	100%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100%	100%	Immunodeficiency 51, 613953
IL17RC	100%	100%	Candidiasis, familial, 9, 616445
IL18BP	100%	100%	No OMIM disease ID
IL1RN	100%	100%	Interleukin 1 receptor antagonist deficiency, 612852
IL2	100%	100%	No OMIM disease ID
IL21	100%	100%	?Immunodeficiency, common variable, 11, 615767
IL21R	100%	100%	Immunodeficiency 56, 615207
IL2RA	100%	100%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL2RB	100%	100%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL2RG	100%	100%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL36RN	100%	100%	Psoriasis 14, pustular, 614204
IL6R	92%	92%	Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944
IL6ST	100%	100%	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IL7R	100%	100%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INO80	100%	100%	No OMIM disease ID
INSR	100%	100%	Rabson-Mendenhall syndrome, 262190 Leprechaunism, 246200 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
IPO8	100%	100%	VISS syndrome, 619472
IRAK1	100%	100%	No OMIM disease ID
IRAK4	100%	100%	Immunodeficiency 67, 607676
IRF2BP2	100%	100%	?Immunodeficiency, common variable, 14, 617765
IRF3	100%	100%	No OMIM disease ID
IRF4	100%	100%	No OMIM disease ID
IRF7	100%	100%	?Immunodeficiency 39, 616345
IRF8	100%	100%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRF9	100%	100%	Immunodeficiency 65, susceptibility to viral infections, 618648
IRGM	100%	100%	No OMIM disease ID
ISG15	100%	100%	Immunodeficiency 38, 616126
ITCH	95%	93%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGB2	97%	97%	Leukocyte adhesion deficiency, 116920
ITK	100%	100%	Lymphoproliferative syndrome 1, 613011
ITPKB	100%	100%	No OMIM disease ID

ITPR3	100%	100%	No OMIM disease ID
IVNS1ABP	100%	100%	Immunodeficiency 70, 618969
JAGN1	100%	100%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	100%	100%	Autoinflammation, immune dysregulation, and eosinophilia, 618999
JAK2	100%	100%	Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300
JAK3	100%	100%	SCID, autosomal recessive, T-negative/B-positive type, 600802
KDM6A	100%	100%	Kabuki syndrome 2, 300867
KMT2D	100%	100%	Kabuki syndrome 1, 147920
KNG1	100%	100%	Angioedema, hereditary, 6, 619363
KRAS	100%	100%	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%	100%	Juvenile arthritis, 618795
LAMTOR2	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAT	100%	100%	Immunodeficiency 52, 617514
LCK	100%	100%	?Immunodeficiency 22, 615758
LCP2	100%	100%	?Immunodeficiency 81, 619374
LIG1	100%	100%	No OMIM disease ID
LIG4	100%	100%	LIG4 syndrome, 606593
LPIN2	100%	100%	Majeed syndrome, 609628
LRBA	100%	100%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC32	100%	100%	Cleft palate, proliferative retinopathy, and developmental delay, 619074
LRRC8A	100%	100%	?Agammaglobulinemia 5, 613506
LSM11	100%	100%	?Aicardi-Goutieres syndrome 8, 619486

LYST	100%	100%	Chediak-Higashi syndrome, 214500
MAGT1	98%	98%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 Congenital disorder of glycosylation, type Icc, 301031
MAL2	100%	100%	No OMIM disease ID
MALT1	100%	100%	Immunodeficiency 12, 615468
MAN2B1	100%	100%	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	100%	100%	No OMIM disease ID
MANBA	100%	100%	Mannosidosis, beta, 248510
MAP1LC3B2	100%	100%	No OMIM disease ID
MAP3K14	100%	100%	No OMIM disease ID
MAPK8	100%	100%	No OMIM disease ID
MASP2	100%	100%	MASP2 deficiency, 613791
MBL2	100%	100%	No OMIM disease ID
MC2R	100%	100%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM10	100%	100%	Immunodeficiency 80 with or without cardiomyopathy, 619313
MCM4	95%	95%	Immunodeficiency 54, 609981
MEFV	96%	96%	Neutrophilic dermatosis, acute febrile, 608068 Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MOGS	100%	100%	Congenital disorder of glycosylation, type IIb, 606056
MPEG1	100%	100%	Immunodeficiency 77, 619223
MRE11	100%	100%	Ataxia-telangiectasia-like disorder 1, 604391
MRTFA	92%	92%	?Immunodeficiency 66, 618847
MS4A1	100%	100%	?Immunodeficiency, common variable, 5, 613495
MSN	100%	100%	Immunodeficiency 50, 300988
MTHFD1	100%	100%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MVK	90%	90%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYD88	100%	100%	Macroglobulinemia, Waldenstrom, somatic, 153600 Immunodeficiency 68, 612260
MYOF	100%	100%	?Angioedema, hereditary, 7, 619366
MYSM1	96%	96%	Bone marrow failure syndrome 4, 618116
NBAS	100%	100%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483

NBN	100%	100%	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NCF1	100%	99%	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	100%	100%	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100%	100%	Chronic granulomatous disease 3, autosomal recessive, 613960
NCKAP1L	100%	100%	Immunodeficiency 72 with autoinflammation, 618982
NCSTN	100%	100%	Acne inversa, familial, 1, 142690
NFAT5	100%	100%	No OMIM disease ID
NFATC1	100%	100%	No OMIM disease ID
NFE2L2	100%	100%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFKB1	100%	100%	Immunodeficiency, common variable, 12, 616576
NFKB2	100%	100%	Immunodeficiency, common variable, 10, 615577
NFKBIA	100%	100%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHEJ1	100%	100%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	100%	100%	Dyskeratosis congenita, autosomal recessive 2, 613987
NLRC4	100%	100%	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050
NLRP1	100%	100%	?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225
NLRP12	100%	100%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	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	100%	100%	No OMIM disease ID
NLRP7	100%	100%	Hydatidiform mole, recurrent, 1, 231090
NOD2	100%	100%	Blau syndrome, 186580
NOP10	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOS2	100%	100%	No OMIM disease ID
NRAS	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%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
OAS1	100%	100%	No OMIM disease ID
ORAI1	100%	99%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
OSTM1	100%	100%	Osteopetrosis, autosomal recessive 5, 259720
OTULIN	100%	99%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
PARN	90%	87%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX1	100%	100%	Otofaciocervical syndrome 2, 615560
PAX5	100%	100%	No OMIM disease ID
PBX1	100%	100%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	100%	100%	Propionicacidemia, 606054
PCCB	99%	98%	Propionicacidemia, 606054
PDCD1	100%	100%	No OMIM disease ID
PEPD	100%	100%	Prolidase deficiency, 170100
PEX16	100%	100%	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PGM3	91%	91%	Immunodeficiency 23, 615816
PIGA	100%	100%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIK3CD	100%	100%	Immunodeficiency 14A, autosomal dominant, 615513 Immunodeficiency 14B, autosomal recessive, 619281 ?Roifman-Chitayat syndrome, digenic, 613328
PIK3CG	100%	100%	No OMIM disease ID
PIK3R1	100%	100%	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PLCG2	100%	100%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEKHM1	100%	100%	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107

PLG	100%	100%	Dysplasminogenemia, 217090 Angioedema, hereditary, 4, 619360 Plasminogen deficiency, type I, 217090
PMM2	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNP	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POLA1	100%	100%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLD1	100%	100%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE2	100%	100%	No OMIM disease ID
POMP	100%	100%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POT1	100%	100%	No OMIM disease ID
POU2AF1	100%	100%	No OMIM disease ID
PRF1	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRKCD	100%	100%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	100%	100%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	100%	100%	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%	100%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMA3	100%	100%	No OMIM disease ID
PSMB4	100%	100%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	100%	100%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	100%	100%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMG2	100%	100%	?Proteasome-associated autoinflammatory syndrome 4, 619183
PSTPIP1	100%	100%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN22	100%	100%	No OMIM disease ID
PTPRC	100%	100%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
RAB27A	100%	100%	Griscelli syndrome, type 2, 607624
RAC2	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%	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%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RANBP2	100%	100%	No OMIM disease ID
RASGRP1	100%	100%	Immunodeficiency 64, 618534
RASGRP2	100%	100%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	100%	100%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RC3H1	100%	100%	?Immune dysregulation and systemic hyperinflammation syndrome, 618998
RECQL4	100%	100%	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
REL	99%	98%	No OMIM disease ID
RELA	100%	100%	?Mucocutaneous ulceration, chronic, 618287
RELB	100%	100%	?Immunodeficiency 53, 617585
RFX5	100%	100%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	100%	100%	MHC class II deficiency, complementation group B, 209920
RFXAP	100%	100%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGS10	100%	100%	No OMIM disease ID
RHOG	100%	100%	No OMIM disease ID
RHOH	100%	100%	No OMIM disease ID
RIPK1	100%	100%	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%	100%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91%	91%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNF168	100%	100%	RIDDLE syndrome, 611943
RNF31	100%	100%	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%	100%	Immunodeficiency 42, 616622
RPA1	100%	100%	No OMIM disease ID
RPSA	100%	100%	Asplenia, isolated congenital, 271400
RSPH9	100%	100%	Ciliary dyskinesia, primary, 12, 612650
RTEL1	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	100%	100%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMD9L	100%	100%	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SAMHD1	100%	100%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SASH3	100%	100%	No OMIM disease ID
SBDS	100%	100%	Shwachman-Diamond syndrome, 260400
SEC61A1	100%	100%	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056
SEMA3E	100%	100%	?CHARGE syndrome, 214800
SERAC1	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	100%	100%	Angioedema, hereditary, 1 and 2, 106100 Complement component 4, partial deficiency of, 120790
SH2B3	100%	100%	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SH2D1A	100%	100%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	99%	98%	Cherubism, 118400
SH3KBP1	100%	100%	?Immunodeficiency 61, 300310
SKIV2L	100%	100%	Trichohepatoenteric syndrome 2, 614602
SLC29A3	100%	100%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	100%	100%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	100%	100%	Congenital disorder of glycosylation, type IIc, 266265

SLC37A4	100%	100%	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC39A4	100%	100%	Acrodermatitis enteropathica, 201100
SLC39A7	100%	100%	No OMIM disease ID
SLC46A1	100%	100%	Folate malabsorption, hereditary, 229050
SLC7A7	100%	100%	Lysinuric protein intolerance, 222700
SMARCAL1	100%	100%	Schimke immunoosseous dysplasia, 242900
SMARCD2	100%	100%	Specific granule deficiency 2, 617475
SNORA31	NC	NC	No OMIM disease ID
SNX10	100%	99%	Osteopetrosis, autosomal recessive 8, 615085
SOCS1	100%	100%	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375
SOCS4	100%	100%	No OMIM disease ID
SP110	100%	100%	Hepatic venoocclusive disease with immunodeficiency, 235550
SPINK5	100%	100%	Netherton syndrome, 256500
SPPL2A	100%	100%	Immunodeficiency 86, mycobacteriosis, 619549
SRP54	100%	100%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	100%	100%	Bone marrow failure syndrome 1, 614675
STAT1	95%	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%	100%	Pseudo-TORCH syndrome 3, 618886 Immunodeficiency 44, 616636
STAT3	100%	100%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT4	100%	100%	No OMIM disease ID
STAT5B	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%	100%	No OMIM disease ID
STIM1	100%	100%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STING1	100%	100%	STING-associated vasculopathy, infantile-onset, 615934
STK4	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	99%	99%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101

SYK	100%	100%	Immunodeficiency 82 with systemic inflammation, 619381
TAFAZZIN	100%	100%	Barth syndrome, 302060
TAP1	100%	100%	Bare lymphocyte syndrome, type I, 604571
TAP2	100%	100%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96%	96%	Bare lymphocyte syndrome, type I, 604571
TBX1	97%	94%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX21	100%	100%	Asthma and nasal polyps, 208550 ?Immunodeficiency 88, 619630
TCF3	100%	100%	Agammaglobulinemia 8, autosomal dominant, 616941
TCIRG1	100%	100%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100%	100%	Transcobalamin II deficiency, 275350
TERC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	100%	100%	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%	100%	Myelodysplastic syndrome, somatic, 614286 Immunodeficiency 75, 619126
TFRC	100%	100%	Immunodeficiency 46, 616740
TGFB1	100%	100%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 Camurati-Engelmann disease, 131300
THBD	100%	100%	Thrombophilia due to thrombomodulin defect, 614486
TICAM1	100%	100%	No OMIM disease ID
TINF2	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TIRAP	100%	100%	No OMIM disease ID
TLR3	100%	100%	No OMIM disease ID
TLR4	100%	100%	No OMIM disease ID
TLR5	100%	100%	No OMIM disease ID
TLR7	100%	100%	Immunodeficiency 74, COVID19-related, X-linked, 301051
TLR8	100%	100%	No OMIM disease ID
TMC6	100%	100%	Epidermodysplasia verruciformis, 226400
TMC8	100%	100%	Epidermodysplasia verruciformis 2, 618231
TNFAIP3	100%	100%	Autoinflammatory syndrome, familial, Behcet-like, 616744

TNFRSF11A	99%	99%	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810
TNFRSF13B	100%	100%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	100%	100%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	92%	92%	Periodic fever, familial, 142680
TNFRSF4	100%	100%	?Immunodeficiency 16, 615593
TNFRSF9	100%	100%	No OMIM disease ID
TNFSF11	100%	100%	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	100%	100%	No OMIM disease ID
TNFSF13	100%	100%	No OMIM disease ID
TOM1	100%	100%	?Immunodeficiency 85 and autoimmunity, 619510
TOP2B	100%	100%	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296
TPP2	100%	100%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220
TRAC	100%	100%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	100%	100%	No OMIM disease ID
TRAF3IP2	100%	100%	?Candidiasis, familial, 8, 615527
TREX1	100%	100%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM22	100%	100%	No OMIM disease ID
TRNT1	100%	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TTC37	100%	100%	Trichohepatoenteric syndrome 1, 222470
TTC7A	100%	100%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	100%	100%	Immunodeficiency 35, 611521
UBA1	100%	99%	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
UNC13D	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	100%	100%	No OMIM disease ID
UNG	100%	100%	Immunodeficiency with hyper IgM, type 5, 608106
USB1	100%	100%	Poikiloderma with neutropenia, 604173
USP18	100%	100%	Pseudo-TORCH syndrome 2, 617397
VAV1	97%	97%	No OMIM disease ID
VPS13B	99%	99%	Cohen syndrome, 216550
VPS45	95%	95%	Neutropenia, severe congenital, 5, autosomal recessive, 615285

WAS	100%	100%	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WDR1	100%	100%	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550
WIPF1	100%	100%	Wiskott-Aldrich syndrome 2, 614493
WRAP53	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	100%	100%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	100%	100%	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB24	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZNF341	100%	100%	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282
ZNFX1	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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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 : January 13th , 2022.
This list is accurate for panel version DG 3.3.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors