WES PRIMARY IMMUNODEFICIENCIES DG 3.7

Gene	Twist X2 covered >10x	Twist X2 covered >20x	WGS covered >10x	WGS covered >20x	Associated Phenotype description and OMIM disease ID
ACD	100.0%	100.0%	100.0%	99.3%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ACP5	100.0%	100.0%	100.0%	99.9%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	100.0%	100.0%	100.0%	99.9%	Baraitser-Winter syndrome 1, 243310 Becker nevus, syndromic or isolated, somatic mosaic, 604919 Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475 Dystonia-deafness syndrome 1, 607371 Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620479
ADA	100.0%	100.0%	100.0%	99.8%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	100.0%	100.0%	100.0%	99.6%	Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	100.0%	100.0%	100.0%	99.3%	?Inflammatory skin and bowel disease, neonatal, 1, 614328

ADAR	100.0%	100.0%	100.0%	99.1%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
AGA	100.0%	100.0%	100.0%	99.6%	Aspartylglucosaminuria, 208400
AICDA	100.0%	100.0%	100.0%	99.2%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	100.0%	100.0%	100.0%	99.9%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	100.0%	100.0%	100.0%	99.3%	Reticular dysgenesis, 267500
ALG13	99.7%	99.0%	97.8%	72.7%	Developmental and epileptic encephalopathy 36, 300884
ALPI	100.0%	100.0%	100.0%	99.8%	
ALPK1	100.0%	100.0%	100.0%	99.6%	ROSAH syndrome, 614979
ANGPT1	100.0%	100.0%	100.0%	99.6%	?Angioedema, hereditary, 5, 619361
AP1S3	90.6%	90.6%	100.0%	98.9%	
AP3B1	100.0%	100.0%	100.0%	99.7%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	100.0%	100.0%	100.0%	99.7%	?Hermansky-Pudlak syndrome 10, 617050
APOL1	100.0%	100.0%	100.0%	99.6%	
ARHGEF1	100.0%	100.0%	100.0%	99.7%	?Immunodeficiency 62, 618459
ARPC1B	100.0%	100.0%	100.0%	99.7%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ATAD3A	100.0%	100.0%	100.0%	99.1%	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATG4A	100.0%	100.0%	98.8%	75.1%	

ATM	100.0%	100.0%	100.0%	99.0%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, T-cell prolymphocytic leukemia, somatic, Lymphoma, mantle cell, somatic,
ATP6AP1	100.0%	99.7%	99.2%	77.2%	Immunodeficiency 47, 300972
B2M	100.0%	100.0%	100.0%	99.5%	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
BACH2	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 60 and autoimmunity, 618394
BCL10	100.0%	100.0%	100.0%	99.6%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245
BCL11B	99.9%	99.6%	100.0%	99.7%	Immunodeficiency 49, severe combined, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BLK	100.0%	100.0%	100.0%	99.6%	Maturity-onset diabetes of the young, type 11, 613375
BLM	100.0%	100.0%	100.0%	99.2%	Bloom syndrome, 210900
BLNK	100.0%	100.0%	100.0%	99.4%	?Agammaglobulinemia 4, 613502
BLOC1S6	100.0%	100.0%	100.0%	99.1%	?Hermansky-Pudlak syndrome 9, 614171
BTK	100.0%	99.9%	98.7%	75.2%	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
C1QA	100.0%	100.0%	100.0%	99.9%	C1q deficiency 1, 613652
C1QB	100.0%	100.0%	100.0%	98.8%	C1q deficiency 2, 620321
C1QC	100.0%	100.0%	100.0%	99.3%	C1q deficiency 3, 620322

C1R	99.9%	98.3%	100.0%	99.9%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	99.9%	99.3%	100.0%	99.6%	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	100.0%	100.0%	100.0%	99.3%	C2 deficiency, 217000
C2orf69	100.0%	100.0%	100.0%	99.3%	Combined oxidative phosphorylation deficiency 53, 619423
C3	100.0%	100.0%	100.0%	99.4%	C3 deficiency, 613779
C5	100.0%	100.0%	100.0%	99.2%	C5 deficiency, 609536
C6	100.0%	99.4%	100.0%	99.1%	C6 deficiency, 612446
C7	99.3%	98.3%	100.0%	99.3%	C7 deficiency, 610102
C8A	100.0%	100.0%	100.0%	99.7%	C8 deficiency, type I, 613790
C8B	100.0%	100.0%	100.0%	99.7%	C8 deficiency, type II, 613789
C8G	100.0%	100.0%	100.0%	99.6%	
C9	100.0%	100.0%	100.0%	99.3%	C9 deficiency, 613825
CA2	100.0%	100.0%	100.0%	99.5%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CARD10	100.0%	100.0%	99.9%	97.9%	?Immunodeficiency 89 and autoimmunity, 619632
CARD11	100.0%	100.0%	100.0%	99.7%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11B with atopic dermatitis, 617638 Immunodeficiency 11A, 615206
CARD14	100.0%	100.0%	100.0%	100.0%	Psoriasis 2, 602723 Pityriasis rubra pilaris, 173200
CARD9	100.0%	100.0%	100.0%	100.0%	Immunodeficiency 103, susceptibility to fungal infection, 212050
CARMIL2	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 58, 618131

CASP10	100.0%	100.0%	100.0%	99.0%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP8	95.1%	95.1%	100.0%	99.5%	?Caspase 8 lymphadenopathy syndrome, 607271 Hepatocellular carcinoma, somatic, 114550
CAVIN1	100.0%	100.0%	100.0%	100.0%	Lipodystrophy, congenital generalized, type 4, 613327
CCBE1	100.0%	100.0%	100.0%	99.9%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD19	100.0%	100.0%	100.0%	99.3%	Immunodeficiency, common variable, 3, 613493
CD247	100.0%	100.0%	100.0%	99.9%	?Immunodeficiency 25, 610163
CD27	100.0%	100.0%	100.0%	99.7%	Lymphoproliferative syndrome 2, 615122
CD28	100.0%	100.0%	100.0%	99.3%	
CD3D	100.0%	100.0%	100.0%	99.9%	Immunodeficiency 19, severe combined, 615617
CD3E	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	100.0%	100.0%	100.0%	98.7%	Immunodeficiency 79, 619238 OKT4 epitope deficiency, 613949
CD40	100.0%	100.0%	100.0%	99.3%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	100.0%	99.6%	98.5%	73.7%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	100.0%	100.0%	100.0%	99.4%	

CD48	100.0%	100.0%	100.0%	99.4%	
CD55	95.8%	92.9%	100.0%	99.7%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	100.0%	100.0%	100.0%	99.6%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	100.0%	100.0%	100.0%	99.2%	Lymphoproliferative syndrome 3, 618261
CD79A	100.0%	99.5%	99.9%	93.2%	Agammaglobulinemia 3, 613501
CD79B	100.0%	100.0%	100.0%	99.8%	Agammaglobulinemia 6, 612692
CD81	100.0%	99.9%	100.0%	100.0%	Immunodeficiency, common variable, 6, 613496
CD8A	100.0%	100.0%	100.0%	99.9%	CD8 deficiency, familial, 608957
CDC42	100.0%	100.0%	100.0%	99.4%	Takenouchi-Kosaki syndrome, 616737
CDCA7	100.0%	100.0%	100.0%	99.6%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDKN2B	100.0%	100.0%	100.0%	99.9%	
CEBPE	100.0%	100.0%	100.0%	99.1%	?Immunodeficiency 108 with autoinflammation, 260570 Specific granule deficiency, 245480
CFB	100.0%	100.0%	100.0%	99.3%	?Complement factor B deficiency, 615561
CFD	100.0%	100.0%	100.0%	99.0%	Complement factor D deficiency, 613912
CFH	100.0%	100.0%	100.0%	99.6%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFI	100.0%	100.0%	100.0%	99.0%	Complement factor I deficiency, 610984

CFP	100.0%	99.8%	98.5%	76.6%	Properdin deficiency, X-linked, 312060
CFTR	100.0%	100.0%	100.0%	99.4%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
CHD7	100.0%	100.0%	100.0%	99.5%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHUK	100.0%	100.0%	100.0%	99.3%	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 ?Cocoon syndrome, 613630
CIB1	100.0%	100.0%	100.0%	99.5%	Epidermodysplasia verruciformis 3, 618267
CIITA	100.0%	100.0%	100.0%	99.6%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CLCN7	100.0%	100.0%	100.0%	99.9%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
CLEC4D	100.0%	100.0%	100.0%	99.7%	
CLEC7A	100.0%	100.0%	100.0%	99.6%	Candidiasis, familial, 4, autosomal recessive, 613108
CLPB	100.0%	100.0%	100.0%	98.9%	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%	100.0%	100.0%	3MC syndrome 2, 265050
COPA	100.0%	100.0%	100.0%	99.4%	

COPG1	100.0%	100.0%	100.0%	99.5%	
CORO1A	100.0%	100.0%	100.0%	98.7%	Immunodeficiency 8, 615401
CR2	100.0%	100.0%	100.0%	99.5%	?Immunodeficiency, common variable, 7, 614699
CRACR2A	100.0%	100.0%	100.0%	99.6%	
CREBBP	100.0%	100.0%	100.0%	99.2%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CSF2RA	97.4%	94.2%	50.0%	48.4%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	100.0%	100.0%	100.0%	99.8%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	100.0%	100.0%	100.0%	99.8%	Neutropenia, severe congenital, 7, autosomal recessive, 617014 ?Neutrophilia, hereditary, 162830
CTC1	100.0%	100.0%	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100.0%	100.0%	100.0%	99.2%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CTNNBL1	100.0%	100.0%	100.0%	99.7%	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846
CTPS1	100.0%	100.0%	100.0%	99.5%	Immunodeficiency 24, 615897
CTSC	100.0%	100.0%	100.0%	99.4%	Periodontitis 1, juvenile, 170650 Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000
CXCR2	100.0%	100.0%	100.0%	99.8%	?WHIM syndrome 2, 619407

CXCR4	100.0%	100.0%	100.0%	99.2%	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670
CYBA	100.0%	100.0%	100.0%	100.0%	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBB	99.8%	98.5%	97.9%	74.3%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400
CYBC1	100.0%	100.0%	100.0%	99.9%	Chronic granulomatous disease 5, autosomal recessive, 618935
DBF4	100.0%	100.0%	100.0%	98.4%	
DBR1	100.0%	100.0%	100.0%	99.2%	
DCLRE1B	100.0%	100.0%	100.0%	99.8%	Dyskeratosis congenita, autosomal recessive 8, 620133
DCLRE1C	100.0%	100.0%	100.0%	99.1%	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DDX41	100.0%	100.0%	100.0%	99.8%	
DDX58	100.0%	100.0%	100.0%	99.5%	Singleton-Merten syndrome 2, 616298
DEF6	100.0%	100.0%	100.0%	99.5%	Immunodeficiency 87 and autoimmunity, 619573
DHFR	100.0%	100.0%	100.0%	99.6%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DIAPH1	100.0%	100.0%	100.0%	97.4%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DKC1	100.0%	100.0%	98.0%	73.8%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 Dyskeratosis congenita, X-linked, 305000

DNASE1	100.0%	100.0%	100.0%	99.8%	
DNASE1L3	100.0%	100.0%	100.0%	99.8%	Systemic lupus erythematosus 16, 614420
DNASE2	100.0%	100.0%	100.0%	99.5%	Autoinflammatory-pancytopenia syndrome, 619858
DNMT3B	100.0%	100.0%	100.0%	99.6%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK2	99.9%	99.5%	100.0%	99.6%	Immunodeficiency 40, 616433
DOCK8	100.0%	100.0%	100.0%	99.4%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	100.0%	100.0%	100.0%	99.9%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	100.0%	99.7%	99.4%	77.2%	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074
EPG5	100.0%	100.0%	100.0%	99.2%	Vici syndrome, 242840
ERBIN	100.0%	100.0%	100.0%	99.3%	
ERCC2	100.0%	100.0%	100.0%	99.7%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100.0%	100.0%	100.0%	99.5%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
EXTL3	100.0%	100.0%	100.0%	99.9%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425

F12	100.0%	100.0%	100.0%	99.9%	Angioedema, hereditary, 3, 610618 Factor XII deficiency, 234000
FAAP24	100.0%	100.0%	100.0%	99.9%	
FADD	100.0%	100.0%	100.0%	99.2%	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759
FAS	100.0%	100.0%	100.0%	98.9%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic,
FASLG	100.0%	100.0%	100.0%	99.4%	Autoimmune lymphoproliferative syndrome, type IB, 601859
FAT4	99.9%	99.8%	100.0%	99.5%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBXW11	100.0%	100.0%	100.0%	99.1%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FCGR3A	100.0%	100.0%	100.0%	99.2%	Immunodeficiency 20, 615707
FCHO1	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 76, 619164
FCN3	100.0%	100.0%	100.0%	99.6%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT1	100.0%	100.0%	100.0%	99.2%	Kindler syndrome, 173650
FERMT3	100.0%	100.0%	100.0%	99.6%	Leukocyte adhesion deficiency, type III, 612840
FNIP1	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOXI3	99.8%	99.0%	100.0%	98.7%	Craniofacial microsomia 2, 620444

FOXN1	100.0%	100.0%	100.0%	99.7%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	100.0%	99.9%	99.6%	80.3%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FPR1	100.0%	100.0%	100.0%	99.3%	
G6PC	100.0%	100.0%	100.0%	99.5%	Glycogen storage disease Ia, 232200
G6PC3	100.0%	100.0%	100.0%	99.8%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	100.0%	99.7%	99.5%	82.4%	Hemolytic anemia, G6PD deficient (favism), 300908
GATA2	100.0%	100.0%	100.0%	99.8%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GFI1	100.0%	100.0%	100.0%	99.7%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GIMAP5	100.0%	100.0%	100.0%	99.4%	Portal hypertension, noncirrhotic, 2, 619463
GINS1	100.0%	100.0%	100.0%	99.8%	Immunodeficiency 55, 617827
GINS4	100.0%	100.0%	100.0%	99.9%	
GJC2	99.8%	98.7%	100.0%	98.6%	Lymphatic malformation 3, 613480 ?Spastic paraplegia 44, autosomal recessive, 613206 Leukodystrophy, hypomyelinating, 2, 608804

GRHL2	100.0%	100.0%	100.0%	99.7%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
GTF2H5	70.4%	70.3%	100.0%	99.5%	Trichothiodystrophy 3, photosensitive, 616395
HAVCR2	100.0%	100.0%	100.0%	99.7%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100.0%	100.0%	100.0%	98.6%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCK	100.0%	100.0%	100.0%	99.7%	Autoinflammation with pulmonary and cutaneous vasculitis, 620296
HELLS	100.0%	100.0%	100.0%	98.8%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HMOX1	100.0%	100.0%	100.0%	99.8%	Heme oxygenase-1 deficiency, 614034
HS3ST6	100.0%	99.6%	100.0%	99.3%	?Angioedema, hereditary, 8, 619367
HYOU1	100.0%	100.0%	100.0%	99.8%	?Immunodeficiency 59 and hypoglycemia, 233600
ICOS	100.0%	100.0%	100.0%	98.0%	Immunodeficiency, common variable, 1, 607594
ICOSLG	100.0%	100.0%	100.0%	100.0%	
IFIH1	100.0%	100.0%	100.0%	99.0%	Immunodeficiency 95, 619773 Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNAR1	97.1%	97.1%	100.0%	98.0%	Immunodeficiency 106, susceptibility to viral infections, 619935
IFNAR2	100.0%	100.0%	100.0%	99.7%	Immunodeficiency 45, 616669
IFNG	100.0%	100.0%	100.0%	99.1%	?Immunodeficiency 69, mycobacteriosis, 618963

IFNGR1	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978
IFNGR2	100.0%	100.0%	100.0%	98.8%	Immunodeficiency 28, mycobacteriosis, 614889
IGHM	100.0%	100.0%	100.0%	99.9%	Agammaglobulinemia 1, 601495
IGLL1	100.0%	100.0%	100.0%	99.8%	Agammaglobulinemia 2, 613500
IKBKB	100.0%	100.0%	100.0%	98.8%	Immunodeficiency 15B, 615592 Immunodeficiency 15A, 618204
IKBKG	99.9%	98.4%	99.1%	80.0%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636 Autoinflammatory disease, systemic, X-linked, 301081
IKZF1	100.0%	100.0%	100.0%	99.7%	Immunodeficiency, common variable, 13, 616873
IKZF2	100.0%	100.0%	100.0%	99.3%	
IKZF3	100.0%	100.0%	100.0%	99.5%	?Immunodeficiency 84, 619437
IL10	100.0%	100.0%	100.0%	99.6%	
IL10RA	100.0%	100.0%	100.0%	99.9%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	100.0%	100.0%	100.0%	99.2%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL12B	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	94.1%	94.1%	100.0%	99.5%	Immunodeficiency 30, 614891

IL17F	100.0%	100.0%	100.0%	99.6%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100.0%	100.0%	100.0%	99.9%	Immunodeficiency 51, 613953
IL17RC	100.0%	100.0%	100.0%	99.8%	Candidiasis, familial, 9, 616445
IL18BP	100.0%	100.0%	100.0%	99.7%	
IL1RN	100.0%	100.0%	100.0%	99.5%	Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852 Interleukin 1 receptor antagonist deficiency, 612852
IL2	100.0%	99.8%	99.9%	98.4%	
IL21	100.0%	100.0%	100.0%	98.3%	?Immunodeficiency, common variable, 11, 615767
IL21R	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 56, 615207
IL2RA	100.0%	100.0%	100.0%	99.7%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL2RB	100.0%	100.0%	100.0%	99.7%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL2RG	100.0%	100.0%	98.6%	73.9%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL36RN	100.0%	100.0%	100.0%	99.0%	Psoriasis 14, pustular, 614204
IL6R	92.5%	92.5%	100.0%	99.6%	Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944

IL6ST	100.0%	100.0%	100.0%	99.3%	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%	100.0%	99.7%	Immunodeficiency 104, severe combined, 608971
INO80	100.0%	100.0%	100.0%	99.5%	
INSR	100.0%	100.0%	100.0%	99.5%	Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Donohue syndrome, 246200 Hyperinsulinemic hypoglycemia, familial, 5, 609968
IPO8	100.0%	100.0%	100.0%	99.3%	VISS syndrome, 619472
IRAK1	100.0%	99.8%	98.6%	77.8%	
IRAK4	100.0%	100.0%	100.0%	98.9%	Immunodeficiency 67, 607676
IRF2BP2	100.0%	100.0%	100.0%	98.6%	?Immunodeficiency, common variable, 14, 617765
IRF3	100.0%	100.0%	100.0%	99.6%	
IRF4	100.0%	100.0%	100.0%	99.6%	
IRF7	100.0%	100.0%	100.0%	99.7%	?Immunodeficiency 39, 616345
IRF8	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990

IRF9	100.0%	100.0%	100.0%	99.8%	Immunodeficiency 65, susceptibility to viral infections, 618648
IRGM	100.0%	100.0%	100.0%	99.5%	
ISG15	100.0%	100.0%	100.0%	100.0%	Immunodeficiency 38, 616126
ITCH	96.0%	96.0%	100.0%	99.2%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGB2	100.0%	100.0%	100.0%	99.9%	Leukocyte adhesion deficiency, 116920
ITK	100.0%	100.0%	100.0%	99.4%	Lymphoproliferative syndrome 1, 613011
ITPKB	100.0%	100.0%	100.0%	99.9%	
ITPR3	100.0%	100.0%	100.0%	99.7%	Charcot-Marie-Tooth disease, demyelinating, type 1J, 620111
IVNS1ABP	100.0%	100.0%	100.0%	99.1%	Immunodeficiency 70, 618969
JAGN1	100.0%	100.0%	100.0%	99.9%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	100.0%	100.0%	100.0%	99.3%	Autoinflammation, immune dysregulation, and eosinophilia, 618999
JAK2	100.0%	100.0%	100.0%	99.2%	Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300
JAK3	100.0%	100.0%	100.0%	99.6%	SCID, autosomal recessive, T-negative/B-positive type, 600802
KDM6A	100.0%	99.9%	98.5%	73.8%	Kabuki syndrome 2, 300867
KMT2A	100.0%	100.0%	100.0%	99.1%	Wiedemann-Steiner syndrome, 605130

KMT2D	100.0%	100.0%	100.0%	99.6%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186 Kabuki syndrome 1, 147920
KNG1	100.0%	100.0%	100.0%	99.4%	Angioedema, hereditary, 6, 619363
KRAS	100.0%	100.0%	100.0%	99.8%	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%	100.0%	99.2%	Juvenile arthritis, 618795
LAMTOR2	100.0%	100.0%	100.0%	99.9%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAT	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 52, 617514
LCK	100.0%	100.0%	100.0%	99.8%	?Immunodeficiency 22, 615758
LCP2	100.0%	100.0%	100.0%	99.4%	?Immunodeficiency 81, 619374
LIG1	100.0%	100.0%	100.0%	99.7%	Immunodeficiency 96, 619774

LIG4	100.0%	100.0%	100.0%	99.4%	LIG4 syndrome, 606593
LPIN2	100.0%	100.0%	100.0%	99.1%	Majeed syndrome, 609628
LRBA	100.0%	99.9%	100.0%	99.3%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC32	100.0%	100.0%	100.0%	99.9%	Cleft palate, proliferative retinopathy, and developmental delay, 619074
LRRC8A	100.0%	100.0%	100.0%	99.9%	?Agammaglobulinemia 5, 613506
LSM11	100.0%	100.0%	100.0%	99.5%	?Aicardi-Goutieres syndrome 8, 619486
LYN	100.0%	100.0%	100.0%	99.5%	Autoinflammatory disease, systemic, with vasculitis, 620376
LYST	100.0%	99.8%	100.0%	99.4%	Chediak-Higashi syndrome, 214500
MAGT1	97.6%	97.6%	98.2%	73.2%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 Congenital disorder of glycosylation, type Icc, 301031
MALT1	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 12, 615468
MAN2B1	100.0%	100.0%	100.0%	99.7%	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	100.0%	100.0%	100.0%	99.9%	
MANBA	100.0%	100.0%	100.0%	99.5%	Mannosidosis, beta, 248510
MAP1LC3B2	100.0%	100.0%	100.0%	100.0%	
MAP3K14	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 112, 620449
MAPK8	100.0%	100.0%	100.0%	99.3%	
MASP2	100.0%	100.0%	100.0%	99.7%	MASP2 deficiency, 613791
MC2R	100.0%	100.0%	100.0%	99.5%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200

MCM10	100.0%	100.0%	100.0%	99.5%	Immunodeficiency 80 with or without cardiomyopathy, 619313
MCM4	95.3%	95.3%	100.0%	99.3%	Immunodeficiency 54, 609981
MEFV	96.1%	96.1%	100.0%	99.7%	Neutrophilic dermatosis, acute febrile, 608068 Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MOGS	100.0%	100.0%	100.0%	99.9%	Congenital disorder of glycosylation, type IIb, 606056
MPEG1	100.0%	100.0%	100.0%	99.8%	Immunodeficiency 77, 619223
MRTFA	100.0%	100.0%	100.0%	99.4%	?Immunodeficiency 66, 618847
MS4A1	100.0%	100.0%	100.0%	98.3%	?Immunodeficiency, common variable, 5, 613495
MSN	100.0%	98.9%	98.4%	73.2%	Immunodeficiency 50, 300988
MTHFD1	100.0%	100.0%	100.0%	99.2%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MVK	90.4%	90.4%	100.0%	100.0%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYD88	100.0%	100.0%	100.0%	99.9%	Macroglobulinemia, Waldenstrom, somatic, 153600 Immunodeficiency 68, 612260
MYOF	100.0%	100.0%	100.0%	99.1%	?Angioedema, hereditary, 7, 619366
MYSM1	100.0%	100.0%	100.0%	99.1%	Bone marrow failure syndrome 4, 618116

NBAS	100.0%	99.9%	100.0%	99.4%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NBN	100.0%	100.0%	100.0%	98.8%	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NCF1	100.0%	99.6%	100.0%	98.9%	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	100.0%	100.0%	100.0%	99.5%	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100.0%	100.0%	100.0%	99.8%	Chronic granulomatous disease 3, autosomal recessive, 613960
NCKAP1L	100.0%	100.0%	100.0%	99.3%	Immunodeficiency 72 with autoinflammation, 618982
NCSTN	100.0%	100.0%	100.0%	99.7%	Acne inversa, familial, 1, 142690
NFAT5	100.0%	100.0%	100.0%	99.2%	
NFATC1	100.0%	100.0%	100.0%	99.5%	
NFE2L2	100.0%	100.0%	100.0%	99.6%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFKB1	100.0%	100.0%	100.0%	99.5%	Immunodeficiency, common variable, 12, 616576
NFKB2	100.0%	100.0%	100.0%	99.5%	Immunodeficiency, common variable, 10, 615577
NFKBIA	100.0%	100.0%	100.0%	99.4%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHEJ1	100.0%	100.0%	100.0%	99.7%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291

NHP2	100.0%	100.0%	100.0%	99.3%	Dyskeratosis congenita, autosomal recessive 2, 613987
NLRC4	100.0%	100.0%	100.0%	99.3%	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050
NLRP1	100.0%	100.0%	100.0%	99.6%	?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225
NLRP12	100.0%	100.0%	100.0%	99.3%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	100.0%	100.0%	100.0%	99.3%	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%	100.0%	99.4%	
NOD2	100.0%	100.0%	100.0%	99.8%	Blau syndrome, 186580
NOP10	100.0%	100.0%	100.0%	99.8%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400 ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425 ?Dyskeratosis congenita, autosomal recessive 1, 224230
NOS2	100.0%	100.0%	100.0%	99.8%	

NRAS	100.0%	100.0%	100.0%	99.8%	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%	100.0%	99.5%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
OAS1	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinemia, 618042
ORAI1	100.0%	100.0%	100.0%	98.8%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
OSTM1	100.0%	100.0%	100.0%	99.7%	Osteopetrosis, autosomal recessive 5, 259720
OTULIN	100.0%	100.0%	100.0%	99.9%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
PARN	97.0%	95.9%	100.0%	99.5%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
PAX1	100.0%	100.0%	100.0%	99.2%	Otofaciocervical syndrome 2, 615560

PAX5	100.0%	100.0%	100.0%	99.7%	
PBX1	100.0%	99.9%	100.0%	99.6%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	100.0%	100.0%	100.0%	99.1%	Propionicacidemia, 606054
PCCB	99.9%	98.0%	100.0%	99.4%	Propionicacidemia, 606054
PDCD1	100.0%	100.0%	100.0%	100.0%	
PEPD	100.0%	100.0%	100.0%	99.9%	Prolidase deficiency, 170100
PEX16	100.0%	100.0%	100.0%	99.3%	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PGM3	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 23, 615816
PIGA	100.0%	100.0%	98.6%	74.4%	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%	100.0%	99.6%	Immunodeficiency 14A, autosomal dominant, 615513 Immunodeficiency 14B, autosomal recessive, 619281 ?Roifman-Chitayat syndrome, digenic, 613328
PIK3CG	100.0%	100.0%	100.0%	99.7%	Immunodeficiency 97 with autoinflammation, 619802
PIK3R1	100.0%	100.0%	100.0%	99.2%	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880

PLCG2	100.0%	100.0%	100.0%	99.6%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEKHM1	100.0%	100.0%	100.0%	99.5%	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLG	100.0%	100.0%	100.0%	99.4%	Dysplasminogenemia, 217090 Angioedema, hereditary, 4, 619360 Plasminogen deficiency, type I, 217090
PMM2	100.0%	100.0%	100.0%	98.7%	Congenital disorder of glycosylation, type Ia, 212065
PNP	100.0%	100.0%	100.0%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POLA1	99.7%	99.4%	98.5%	72.6%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLD1	100.0%	100.0%	100.0%	99.7%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE2	100.0%	100.0%	100.0%	99.1%	
POLR3F	100.0%	100.0%	100.0%	99.7%	?Immunodeficiency 101 (varicella zoster virus-specific), 619872
POMP	100.0%	100.0%	100.0%	98.6%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952

POT1	100.0%	100.0%	100.0%	99.5%	?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368 ?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367
POU2AF1	100.0%	100.0%	100.0%	99.8%	
PRF1	100.0%	100.0%	100.0%	100.0%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRKCD	100.0%	100.0%	100.0%	99.8%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	100.0%	100.0%	99.0%	75.1%	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%	100.0%	98.9%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMA3	100.0%	100.0%	100.0%	99.1%	
PSMB4	100.0%	100.0%	100.0%	98.8%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	100.0%	100.0%	100.0%	99.2%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040

PSMB9	100.0%	100.0%	100.0%	99.5%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMG2	100.0%	100.0%	100.0%	99.7%	?Proteasome-associated autoinflammatory syndrome 4, 619183
PSTPIP1	100.0%	100.0%	100.0%	99.9%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN22	100.0%	100.0%	100.0%	98.9%	
PTPRC	100.0%	99.8%	100.0%	99.3%	Immunodeficiency 105, severe combined, 619924
RAB27A	100.0%	100.0%	100.0%	100.0%	Griscelli syndrome, type 2, 607624
RAC2	100.0%	100.0%	100.0%	99.7%	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%	100.0%	99.7%	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%	100.0%	99.3%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554

RANBP2	100.0%	100.0%	100.0%	98.7%	
RASGRP1	100.0%	100.0%	100.0%	99.8%	Immunodeficiency 64, 618534
RASGRP2	100.0%	100.0%	100.0%	99.7%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	100.0%	100.0%	100.0%	99.6%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RC3H1	100.0%	100.0%	100.0%	99.7%	?Immune dysregulation and systemic hyperinflammation syndrome, 618998
RECQL4	100.0%	100.0%	100.0%	100.0%	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
REL	100.0%	99.5%	100.0%	98.4%	Immunodeficiency 92, 619652
RELA	100.0%	100.0%	100.0%	99.6%	Autoinflammatory disease, familial, Behcet-like-3, 618287
RELB	100.0%	99.9%	100.0%	99.7%	?Immunodeficiency 53, 617585
RFX5	100.0%	100.0%	100.0%	99.4%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	100.0%	100.0%	100.0%	100.0%	Bare lymphocyte syndrome, type II, complementation group B, 209920
RFXAP	100.0%	100.0%	100.0%	99.4%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGS10	100.0%	100.0%	100.0%	99.4%	
RHOG	100.0%	100.0%	100.0%	100.0%	
RHOH	100.0%	100.0%	100.0%	99.2%	

RIPK1	100.0%	100.0%	100.0%	99.3%	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852
RMRP	%	%	%	%	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNASEH2A	100.0%	100.0%	100.0%	99.9%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91.4%	91.4%	100.0%	98.7%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100.0%	100.0%	100.0%	99.7%	Aicardi-Goutieres syndrome 3, 610329
RNF168	100.0%	100.0%	100.0%	99.4%	RIDDLE syndrome, 611943
RNF31	100.0%	100.0%	100.0%	99.7%	
RNU4ATAC	%	%	%	%	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RNU7-1	%	%	%	%	Aicardi-Goutieres syndrome 9, 619487
RORC	100.0%	100.0%	100.0%	99.9%	Immunodeficiency 42, 616622
RPA1	100.0%	100.0%	100.0%	99.7%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767
RPSA	100.0%	100.0%	100.0%	99.5%	Asplenia, isolated congenital, 271400
RSPH9	100.0%	100.0%	100.0%	99.7%	Ciliary dyskinesia, primary, 12, 612650

RTEL1	100.0%	100.0%	100.0%	99.9%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373
SAMD9	100.0%	100.0%	100.0%	99.0%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMD9L	100.0%	100.0%	100.0%	98.9%	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Spinocerebellar ataxia 49, 619806
SAMHD1	100.0%	100.0%	100.0%	99.3%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SASH3	100.0%	99.9%	99.0%	78.9%	Immunodeficiency 102, 301082
SAT1	100.0%	100.0%	99.3%	76.4%	
SBDS	100.0%	100.0%	100.0%	99.1%	Shwachman-Diamond syndrome 1, 260400
SEC61A1	100.0%	100.0%	100.0%	99.4%	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056
SEMA3E	100.0%	100.0%	100.0%	99.7%	
SERAC1	100.0%	100.0%	100.0%	99.1%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	100.0%	100.0%	100.0%	98.9%	Angioedema, hereditary, 1 and 2, 106100 Complement component 4, partial deficiency of, 120790

SH2B3	100.0%	100.0%	100.0%	99.1%	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SH2D1A	100.0%	100.0%	99.8%	79.8%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	99.9%	99.4%	100.0%	99.6%	Cherubism, 118400
SH3KBP1	99.9%	99.5%	98.0%	72.9%	?Immunodeficiency 61, 300310
SKIV2L	100.0%	100.0%	100.0%	99.7%	Trichohepatoenteric syndrome 2, 614602
SLC29A3	100.0%	100.0%	100.0%	99.8%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	100.0%	100.0%	100.0%	99.8%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	100.0%	100.0%	100.0%	100.0%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	100.0%	100.0%	100.0%	99.6%	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC39A4	100.0%	100.0%	100.0%	99.9%	Acrodermatitis enteropathica, 201100
SLC39A7	100.0%	100.0%	100.0%	99.4%	Agammaglobulinemia 9, autosomal recessive, 619693
SLC46A1	100.0%	100.0%	100.0%	99.8%	Folate malabsorption, hereditary, 229050
SLC7A7	100.0%	100.0%	100.0%	99.1%	Lysinuric protein intolerance, 222700
SMARCAL1	100.0%	100.0%	100.0%	99.4%	Schimke immunoosseous dysplasia, 242900
SMARCD2	100.0%	100.0%	100.0%	99.6%	Specific granule deficiency 2, 617475

SNORA31	%	%	%	%	
SNX10	100.0%	100.0%	100.0%	99.6%	Osteopetrosis, autosomal recessive 8, 615085
SOCS1	100.0%	100.0%	100.0%	99.6%	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375
SOCS4	100.0%	100.0%	100.0%	99.8%	
SP110	100.0%	99.7%	100.0%	99.2%	Hepatic venoocclusive disease with immunodeficiency, 235550
SPI1	100.0%	100.0%	100.0%	100.0%	Agammaglobulinemia 10, autosomal dominant, 619707
SPINK5	100.0%	100.0%	100.0%	99.4%	Netherton syndrome, 256500
SPPL2A	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 86, mycobacteriosis, 619549
SRP54	100.0%	100.0%	100.0%	99.6%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	100.0%	100.0%	100.0%	99.0%	Bone marrow failure syndrome 1, 614675
STAT1	96.1%	95.9%	100.0%	99.6%	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%	100.0%	99.7%	Pseudo-TORCH syndrome 3, 618886 Immunodeficiency 44, 616636
STAT3	100.0%	100.0%	100.0%	98.8%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952

STAT4	100.0%	100.0%	100.0%	99.3%	Disabling pansclerotic morphea of childhood, 620443
STAT5B	100.0%	100.0%	100.0%	99.6%	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%	100.0%	99.7%	
STIM1	100.0%	100.0%	100.0%	99.7%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STING1	100.0%	100.0%	100.0%	99.6%	STING-associated vasculopathy, infantile-onset, 615934
STK4	100.0%	100.0%	100.0%	99.4%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	100.0%	100.0%	100.0%	100.0%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	100.0%	99.9%	100.0%	99.9%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
SYK	100.0%	100.0%	100.0%	99.8%	Immunodeficiency 82 with systemic inflammation, 619381
TAP1	100.0%	100.0%	100.0%	99.4%	Bare lymphocyte syndrome, type I, 604571
TAP2	100.0%	100.0%	100.0%	98.7%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	95.9%	95.9%	100.0%	99.3%	Bare lymphocyte syndrome, type I, 604571

TAZ	100.0%	100.0%	99.3%	74.1%	Barth syndrome, 302060
TBX1	97.7%	95.5%	100.0%	97.6%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX21	100.0%	100.0%	100.0%	99.2%	Asthma and nasal polyps, 208550 ?Immunodeficiency 88, 619630
TCF3	100.0%	100.0%	100.0%	99.7%	Agammaglobulinemia 8B, autosomal recessive, 619824 Agammaglobulinemia 8A, autosomal dominant, 616941
TCIRG1	100.0%	100.0%	100.0%	100.0%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100.0%	100.0%	100.0%	99.7%	Transcobalamin II deficiency, 275350
TERC	%	%	%	%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743 Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	100.0%	100.0%	100.0%	100.0%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742
TET2	100.0%	99.4%	100.0%	99.5%	Myelodysplastic syndrome, somatic, 614286 Immunodeficiency 75, 619126
TFRC	100.0%	100.0%	99.9%	98.9%	Immunodeficiency 46, 616740

TGFB1	100.0%	100.0%	100.0%	99.9%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 Camurati-Engelmann disease, 131300
THBD	100.0%	100.0%	100.0%	98.9%	Thrombophilia 12 due to thrombomodulin defect, 614486
TICAM1	100.0%	100.0%	100.0%	99.7%	
TINF2	100.0%	100.0%	100.0%	99.4%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TIRAP	100.0%	100.0%	100.0%	99.7%	
TLR3	100.0%	100.0%	100.0%	99.0%	
TLR4	100.0%	99.9%	100.0%	99.2%	
TLR5	100.0%	100.0%	100.0%	99.3%	
TLR7	100.0%	100.0%	98.6%	69.1%	Immunodeficiency 74, COVID19-related, X-linked, 301051 Systemic lupus erythematosus 17, 301080
TLR8	100.0%	100.0%	98.4%	72.1%	Immunodeficiency 98 with autoinflammation, X-linked, 301078
TMC6	100.0%	100.0%	100.0%	99.8%	Epidermodysplasia verruciformis, 226400
TMC8	100.0%	100.0%	100.0%	99.8%	Epidermodysplasia verruciformis 2, 618231
TNFAIP3	100.0%	100.0%	100.0%	99.7%	Autoinflammatory syndrome, familial, Behcet-like 1, 616744
TNFRSF11A	100.0%	99.6%	100.0%	99.4%	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810
TNFRSF13B	100.0%	100.0%	100.0%	99.9%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529

TNFRSF13C	100.0%	100.0%	100.0%	99.0%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	92.5%	92.5%	100.0%	99.6%	Periodic fever, familial, 142680
TNFRSF4	100.0%	100.0%	100.0%	99.5%	?Immunodeficiency 16, 615593
TNFRSF9	100.0%	100.0%	100.0%	99.3%	Immunodeficiency 109 with lymphoproliferation, 620282
TNFSF11	100.0%	100.0%	100.0%	99.6%	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	100.0%	100.0%	100.0%	99.6%	
TNFSF13	100.0%	100.0%	100.0%	99.7%	
TOM1	100.0%	100.0%	100.0%	99.5%	?Immunodeficiency 85 and autoimmunity, 619510
TOP2B	100.0%	100.0%	100.0%	98.9%	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296
TPP2	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220
TRAC	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	100.0%	100.0%	100.0%	99.0%	
TRAF3IP2	100.0%	100.0%	100.0%	99.2%	?Candidiasis, familial, 8, 615527
TREX1	100.0%	100.0%	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%	100.0%	99.9%	

TRNT1	100.0%	100.0%	100.0%	99.2%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TTC37	100.0%	100.0%	100.0%	99.3%	Trichohepatoenteric syndrome 1, 222470
TTC7A	100.0%	100.0%	100.0%	99.7%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	100.0%	100.0%	100.0%	99.8%	Immunodeficiency 35, 611521
UBA1	100.0%	99.7%	99.0%	77.6%	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
UNC13D	100.0%	100.0%	100.0%	99.7%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	100.0%	99.7%	100.0%	98.7%	
UNG	100.0%	100.0%	100.0%	99.7%	Immunodeficiency with hyper IgM, type 5, 608106
USB1	100.0%	100.0%	100.0%	98.7%	Poikiloderma with neutropenia, 604173
USP18	100.0%	100.0%	100.0%	99.6%	Pseudo-TORCH syndrome 2, 617397
VAV1	98.3%	98.3%	100.0%	99.7%	
VPS13B	99.6%	99.2%	100.0%	99.4%	Cohen syndrome, 216550
VPS45	95.1%	95.1%	100.0%	99.4%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	100.0%	98.8%	98.8%	74.3%	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900

WDR1	100.0%	100.0%	100.0%	99.3%	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550
WIPF1	100.0%	100.0%	100.0%	99.2%	Wiskott-Aldrich syndrome 2, 614493
WRAP53	100.0%	100.0%	100.0%	99.6%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	100.0%	100.0%	98.9%	73.9%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	100.0%	100.0%	100.0%	99.9%	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB24	100.0%	100.0%	100.0%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZNF341	100.0%	100.0%	100.0%	99.6%	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282
ZNFX1	100.0%	100.0%	100.0%	99.6%	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.
TWIST X2 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry.
TWIST X2 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry.
srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS GRCh38 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : March 17th, 2023.
This list is accurate for panel version DG 3.7.0.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors