PRIMARY IMMUNODEFICIENCIES PANEL DG-4.1.0 (509 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
ACD	100%	100%	100%	98.9%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553
ACP5	100%	100%	100%	98.7%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	100%	100%	100%	99.3%	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, 620470

ADA	87.2%	84.6%	100%	99.5%	Adenosine deaminase deficiency, partial, 102700;Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	95.2%	93.2%	100%	99.3%	Sneddon syndrome, 182410;Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	99.2%	99.2%	100%	99.4%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAR	100%	100%	100%	99.2%	Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010
AGA	100%	100%	100%	99.4%	Aspartylglucosaminuria, 208400
AICDA	92.1%	92%	100%	98.8%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	100%	100%	100%	99%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	100%	100%	100%	99.7%	Reticular dysgenesis, 267500
ALG13	100%	99.8%	99.1%	72.6%	Developmental and epileptic encephalopathy 36, 300884

ALPI	100%	100%	100%	98.4%	
ALPK1	100%	100%	100%	99.6%	ROSAH syndrome, 614979
ANGPT1	100%	100%	100%	99.7%	?Angioedema, hereditary, 5, 619361
AP1S3	90.6%	90.6%	100%	98.9%	{Psoriasis 15, pustular, susceptibility to}, 616106
AP3B1	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	100%	100%	100%	99%	?Hermansky-Pudlak syndrome 10, 617050
APOL1	100%	100%	100%	99.4%	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
ARHGEF1	100%	99.5%	100%	98.3%	?Immunodeficiency 62, 618459
ARPC1B	100%	100%	100%	99.2%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ARPC5	100%	100%	100%	99.1%	Immunodeficiency 133 with autoimmunity and autoinflammation, 620565
ASXL1	100%	100%	100%	99.5%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039

ATAD3A	100%	100%	99.9%	97.4%	Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATG4A	100%	99.9%	99.1%	75.6%	
ATM	100%	100%	100%	99.5%	Lymphoma, B-cell non-Hodgkin, somatic;Ataxia-telangiectasia, 208900;{Breast cancer, susceptibility to}, 114480;T-cell prolymphocytic leukemia, somatic;Lymphoma, mantle cell, somatic
ATP6AP1	100%	99.2%	98.2%	70.9%	Immunodeficiency 47, 300972
B2M	100%	100%	100%	99.8%	Amyloidosis, hereditary systemic 6, 620659;Immunodeficiency 43, 241600
BACH2	100%	100%	100%	98.8%	Immunodeficiency 60 and autoimmunity, 618394

BCL10	100%	100%	100%	99.5%	{Lymphoma, follicular, somatic}, 605027;?Immunodeficiency 37, 616098;{Sezary syndrome, somatic};{Male germ cell tumor, somatic}, 273300;Lymphoma, MALT, somatic, 137245;{Mesothelioma, somatic}, 156240
BCL11B	99.9%	99.3%	100%	94.8%	Immunodeficiency 49, severe combined, 617237;Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BLK	100%	100%	100%	99.2%	Maturity-onset diabetes of the young, type 11, 613375
BLM	96.8%	96.6%	100%	99.7%	Bloom syndrome, 210900
BLNK	94.4%	94.4%	100%	99.5%	?Agammaglobulinemia 4, 613502
BLOC1S6	100%	100%	100%	99.6%	Hermansky-Pudlak syndrome 9, 614171
BTK	100%	99.7%	99%	71.9%	Agammaglobulinemia, X-linked 1, 300755;Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
C1QA	77%	74%	100%	99.6%	C1q deficiency 1, 613652
C1QB	77.3%	76.6%	100%	98.2%	C1q deficiency 2, 620321

C1QC	99.7%	97.8%	100%	99.3%	C1q deficiency 3, 620322
C1R	100%	100%	100%	99.3%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	100%	100%	100%	99.5%	C1s deficiency, 613783;Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	100%	100%	100%	99.6%	C2 deficiency, 217000;{Macular degeneration, age-related, 14, reduced risk of}, 615489
C2orf69	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 53, 619423
C3	97.5%	97.5%	100%	98.1%	C3 deficiency, 613779;{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925;{Macular degeneration, age-related, 9}, 611378
C5	100%	100%	100%	99.8%	C5 deficiency, 609536;[Eculizumab, poor response to], 615749
C6	100%	100%	100%	99.8%	C6 deficiency, 612446
C7	98.2%	97.2%	100%	99.6%	C7 deficiency, 610102
C8A	100%	100%	100%	99.7%	C8 deficiency, type I, 613790
C8B	100%	100%	100%	99.9%	C8 deficiency, type II, 613789

C8G	100%	100%	100%	98.3%	
C9	99.3%	99.3%	100%	99.8%	C9 deficiency, 613825;{Macular degeneration, age-related, 15, susceptibility to}, 615591
CA2	100%	100%	100%	99.8%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CARD10	100%	100%	100%	98.7%	?Immunodeficiency 89 and autoimmunity, 619632
CARD11	100%	100%	100%	99.1%	B-cell expansion with NFKB and T-cell anergy, 616452;Immunodeficiency 11B with atopic dermatitis, 617638;Immunodeficiency 11A, 615206
CARD14	100%	100%	100%	99.4%	Psoriasis 2, 602723;Pityriasis rubra pilaris, 173200
CARD9	100%	100%	100%	99.2%	Immunodeficiency 103, susceptibility to fungal infection, 212050
CARMIL2	100%	99.9%	100%	98.3%	Immunodeficiency 58, 618131
CASP10	100%	99.5%	100%	99.6%	Autoimmune lymphoproliferative syndrome, type II, 603909;Gastric cancer, somatic, 613659;Lymphoma, non-Hodgkin, somatic, 605027

CASP8	97%	97%	100%	99.5%	{Breast cancer, protection against}, 114480;?Caspase 8 lymphadenopathy syndrome, 607271;Hepatocellular carcinoma, somatic, 114550;{Lung cancer, protection against}, 211980
CAVIN1	100%	100%	100%	97.2%	Lipodystrophy, congenital generalized, type 4, 613327
CBLB	100%	100%	100%	99.5%	Autoimmune disease, multisystem, infantile-onset, 3, 620430
CCBE1	100%	100%	100%	99.5%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCR2	100%	100%	100%	99.7%	{HIV infection, susceptibility/resistance to}, 609423;Polycystic lung disease, 219600
CD19	100%	100%	100%	98.4%	Immunodeficiency, common variable, 3, 613493
CD247	77.5%	71.7%	100%	99.5%	?Immunodeficiency 25, 610163
CD27	100%	100%	100%	99%	Lymphoproliferative syndrome 2, 615122
CD28	100%	100%	100%	99.6%	?Immunodeficiency 123 with HPV-related verrucosis, 620901

CD3D	100%	100%	100%	99.2%	Immunodeficiency 19, severe combined, 615617
CD3E	100%	100%	100%	99.2%	Immunodeficiency 18, 615615;Immunodeficiency 18, SCID variant, 615615
CD3G	100%	100%	100%	98.7%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	100%	100%	100%	97.6%	Immunodeficiency 79, 619238;OKT4 epitope deficiency, 613949
CD40	100%	100%	100%	99.1%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	100%	99.9%	99.5%	76%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	100%	100%	100%	99.9%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD48	100%	100%	100%	99.3%	
CD55	96.4%	91.2%	100%	99.3%	[Blood group Cromer], 613793;Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	100%	100%	100%	99.9%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	100%	100%	99.9%	97.5%	Lymphoproliferative syndrome 3, 618261

CD79A	100%	99.4%	100%	98.4%	Agammaglobulinemia 3, 613501
CD79B	100%	100%	100%	99.5%	Agammaglobulinemia 6, 612692
CD81	100%	99.1%	100%	98%	Immunodeficiency, common variable, 6, 613496
CD8A	100%	100%	100%	98.4%	Immunodeficiency 116, 608957
CDC42	100%	100%	100%	99.8%	Takenouchi-Kosaki syndrome, 616737
CDCA7	100%	100%	100%	99.5%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CEBPE	100%	100%	100%	98%	?Immunodeficiency 108 with autoinflammation, 260570;Specific granule deficiency, 245480
CFB	100%	100%	100%	99%	?Complement factor B deficiency, 615561;{Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924;{Macular degeneration, age-related, 14, reduced risk of}, 615489
CFD	100%	100%	100%	95.8%	Complement factor D deficiency, 613912

CFH	97.5%	97.5%	100%	99.8%	{Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CFI	100%	100%	100%	99.9%	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923;{Macular degeneration, age-related, 13, susceptibility to}, 615439;Complement factor I deficiency, 610984
CFP	100%	98.8%	98.9%	67.2%	Properdin deficiency, X-linked, 312060
CFTR	100%	100%	100%	99.5%	Cystic fibrosis, 219700;Sweat chloride elevation without CF;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;{Hypertrypsinemia, neonatal}

CHD7	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CHUK	100%	100%	100%	99.7%	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339;?Cocoon syndrome, 613630
CIB1	100%	100%	100%	99.4%	{Epidermodysplasia verruciformis, susceptibility to, 3}, 618267
CIITA	100%	100%	100%	98.5%	{Rheumatoid arthritis, susceptibility to}, 180300;MHC class II deficiency 1, 209920
CLCN7	100%	100%	100%	99%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600
CLEC4D	100%	100%	100%	99.8%	
CLEC7A	100%	100%	100%	99.8%	Candidiasis, familial, 4, autosomal recessive, 613108;{Aspergillosis, susceptibility to}, 614079

CLPB	100%	100%	100%	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%	100%	100%	99%	3MC syndrome 2, 265050
COPA	100%	100%	100%	99.4%	{Autoinflammation and autoimmunity, systemic, with immune dysregulation}, 616414
COPG1	100%	100%	100%	99.3%	?Immunodeficiency 128, 620983
CORO1A	100%	100%	100%	99.2%	Immunodeficiency 8, 615401
CR2	100%	100%	100%	99.7%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927;?Immunodeficiency, common variable, 7, 614699
CRACR2A	100%	100%	100%	99.6%	
CREBBP	100%	100%	100%	99%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CSF2RA	48.8%	48.2%	50%	49.5%	Surfactant metabolism dysfunction, pulmonary, 4, 300770

CSF2RB	100%	100%	100%	98.7%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	100%	100%	100%	99.3%	Neutropenia, severe congenital, 7, autosomal recessive, 617014;?Neutrophilia, hereditary, 162830
CTC1	100%	100%	100%	99.3%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	93.2%	93.2%	100%	99.4%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100;{Diabetes mellitus, insulin-dependent, 12}, 601388;{Celiac disease, susceptibility to, 3}, 609755;{Hashimoto thyroiditis}, 140300;{Systemic lupus erythematosus, susceptibility to}, 152700
CTNNBL1	100%	100%	100%	99.7%	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846
CTPS1	100%	100%	100%	99.8%	Immunodeficiency 24, 615897

CTSC	94.8%	94.3%	100%	99.5%	Periodontitis 1, juvenile, 170650;Haim-Munk syndrome, 245010;Papillon-Lefevre syndrome, 245000
CXCR2	100%	100%	100%	99.6%	?WHIM syndrome 2, 619407
CXCR4	99%	99%	100%	99.2%	WHIM syndrome 1, 193670;Myelokathexis, isolated, 193670
CYBA	71.5%	69.6%	100%	99.3%	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBB	100%	99.9%	98.9%	74.5%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645;Chronic granulomatous disease, X-linked, 306400
CYBC1	100%	100%	100%	99.1%	Chronic granulomatous disease 5, autosomal recessive, 618935
DBF4	100%	100%	100%	99.5%	
DBR1	100%	100%	100%	99.4%	Xerosis and growth failure with immune and pulmonary dysfunction syndrome, 620510;{Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441
DCLRE1B	100%	100%	100%	99%	Dyskeratosis congenita, autosomal recessive 8, 620133

DCLRE1C	97.1%	97.1%	100%	99.6%	Severe combined immunodeficiency, Athabascan type, 602450;Omenn syndrome, 603554
DDX41	100%	100%	100%	98.7%	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871
DEF6	100%	100%	100%	97.5%	Immunodeficiency 87 and autoimmunity, 619573
DHFR	100%	100%	100%	99.5%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DIAPH1	100%	100%	100%	99.2%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632
DKC1	100%	99.4%	99%	70.2%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DNASE1	100%	100%	100%	99.4%	{Systemic lupus erythematosus, susceptibility to}, 152700
DNASE1L3	100%	100%	100%	99.7%	Systemic lupus erythematosus 16, 614420

DNASE2	100%	100%	100%	99%	Autoinflammatory-pancytopenia syndrome, 619858
DNMT3B	100%	100%	100%	99.1%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860;Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK11	100%	99.8%	99.2%	74.8%	Autoinflammatory disease, multisystem, with immune dysregulation, X-linked, 301109
DOCK2	100%	100%	100%	99.7%	Immunodeficiency 40, 616433
DOCK8	98.6%	98.6%	100%	99.6%	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700
DPP9	100%	100%	100%	98.6%	Hatipoglu immunodeficiency syndrome, 620331
ELANE	100%	100%	100%	98.6%	Neutropenia, cyclic, 162800;Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF4	100%	99.3%	98.7%	66.8%	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074
EPG5	100%	100%	100%	99.6%	Vici syndrome, 242840
ERBIN	100%	100%	100%	99.8%	

EXTL3	100%	99.5%	100%	99.7%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
F12	100%	100%	100%	99.4%	Angioedema, hereditary, 3, 610618;Factor XII deficiency, 234000
FAAP24	100%	100%	100%	99.9%	
FADD	100%	99.9%	100%	97%	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759
FAS	100%	99.8%	100%	99.3%	Squamous cell carcinoma, burn scar-related, somatic;Autoimmune lymphoproliferative syndrome, type IA, 601859;{Autoimmune lymphoproliferative syndrome}, 601859
FASLG	100%	100%	100%	99.3%	Autoimmune lymphoproliferative syndrome, type IB, 601859;{Lung cancer, susceptibility to}, 211980
FAT4	100%	100%	100%	99.6%	Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBXW11	100%	100%	100%	99.5%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914

FCGR3A	99.7%	96.8%	100%	99.3%	Immunodeficiency 20, 615707
FCHO1	98%	96.1%	100%	99.1%	Immunodeficiency 76, 619164
FCN3	100%	100%	100%	99%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT1	100%	100%	100%	99.4%	Kindler syndrome, 173650
FERMT3	100%	100%	100%	99.3%	Leukocyte adhesion deficiency, type III, 612840
FLT3LG	100%	100%	100%	99.1%	?Immunodeficiency 125, 620926
FNIP1	100%	100%	100%	99.7%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOXI3	99.2%	95.9%	100%	94%	Craniofacial microsomia 2, 620444
FOXN1	100%	100%	100%	99.3%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806;T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	100%	99.6%	98.5%	67.1%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FPR1	100%	100%	100%	99.7%	
G6PC1	100%	100%	100%	99.6%	Glycogen storage disease Ia, 232200

G6PC3	96.7%	96.7%	100%	99.7%	Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	86.3%	85.6%	98.6%	69%	Anemia, congenital, nonspherocytic hemolytic, 1, G6PD deficient, 300908;{Resistance to malaria due to G6PD deficiency}, 611162
GATA1	100%	99.7%	98%	66.6%	Anemia, congenital, nonspherocytic hemolytic, 9, 301083;Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 159595;Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367;Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835;Thrombocytopenia with beta-thalassemia, X-linked, 314050

GATA2	85.7%	85.7%	100%	99.3%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286
GFI1	100%	100%	100%	99.2%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847;Neutropenia, severe congenital 2, autosomal dominant, 613107
GIMAP5	100%	100%	100%	99.5%	Portal hypertension, noncirrhotic, 2, 619463
GINS1	81%	81%	100%	99.9%	Immunodeficiency 55, 617827
GINS4	100%	100%	100%	99.8%	
GJC2	99.9%	97.5%	100%	97.2%	Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804
GNAI2	100%	100%	100%	98.3%	Ventricular tachycardia, idiopathic, 192605;Pituitary adenoma, ACTH-secreting, somatic

GRHL2	100%	100%	100%	99.5%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
GTF2H5	59.2%	59.2%	100%	99.6%	Trichothiodystrophy 3, photosensitive, 616395
HAVCR2	100%	100%	100%	99.6%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100%	100%	100%	99.4%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCK	100%	100%	100%	99.1%	Autoinflammation with pulmonary and cutaneous vasculitis, 620296
HELLS	100%	100%	100%	99.9%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HMOX1	100%	100%	100%	99.4%	Heme oxygenase-1 deficiency, 614034;{Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HS3ST6	100%	98.6%	99.9%	92.9%	?Angioedema, hereditary, 8, 619367
HYOU1	100%	100%	100%	99.4%	?Immunodeficiency 59 and hypoglycemia, 233600

ICOS	100%	100%	100%	99.6%	Immunodeficiency, common variable, 1, 607594
ICOSLG	100%	100%	100%	99.1%	?Immunodeficiency 119, 620825
IFIH1	100%	100%	100%	99.8%	Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250
IFNAR1	94.5%	94.5%	100%	99.6%	Immunodeficiency 106, susceptibility to viral infections, 619935
IFNAR2	100%	100%	100%	99.7%	{Hepatitis B virus, susceptibility to}, 610424;Immunodeficiency 45, 616669
IFNG	100%	100%	100%	99.9%	{Hepatitis C virus, response to therapy of}, 609532;{TSC2 angiomyolipomas, renal, modifier of}, 613254;{Aplastic anemia}, 609135;?Immunodeficiency 69, mycobacteriosis, 618963;{Tuberculosis, protection against}, 607948;{AIDS, rapid progression to}, 609423

IFNGR1	100%	100%	100%	99.6%	{H. pylori infection, susceptibility to}, 600263;Immunodeficiency 27A, mycobacteriosis, AR, 209950;Immunodeficiency 27B, mycobacteriosis, AD, 615978;{Tuberculosis infection, protection against}, 607948;{Tuberculosis, susceptibility to}, 607948;{Hepatitis B virus infection, susceptibility to}, 610424
IFNGR2	100%	100%	100%	99.3%	Immunodeficiency 28, mycobacteriosis, 614889
IGHM	100%	100%	100%	99%	Agammaglobulinemia 1, 601495
IGLL1	100%	100%	100%	99.1%	Agammaglobulinemia 2, 613500
IKBKB	100%	100%	100%	99.3%	Immunodeficiency 15B, 615592;Immunodeficiency 15A, 618204
IKBKG	96.4%	94.1%	99.3%	75%	Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081

IKZF1	100%	100%	100%	98.6%	Immunodeficiency, common variable, 13, 616873
IKZF2	100%	100%	100%	99.6%	
IKZF3	100%	100%	100%	99.4%	?Immunodeficiency 84, 619437
IL10	100%	100%	100%	100%	{Rheumatoid arthritis, progression of}, 180300;{Graft-versus-host disease, protection against}, 614395;{HIV-1, susceptibility to}, 609423
IL10RA	100%	100%	100%	99.3%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	100%	100%	100%	99.4%	{Hepatitis B virus, susceptibility to}, 610424;Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL12B	100%	100%	100%	99.9%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	94.1%	94.1%	100%	98.9%	Immunodeficiency 30, 614891
IL17F	100%	100%	100%	99.3%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100%	100%	100%	99.4%	Immunodeficiency 51, 613953
IL17RC	100%	100%	100%	98.9%	Candidiasis, familial, 9, 616445

IL18BP	100%	100%	100%	99.4%	{?Hepatitis, fulminant viral, susceptibility to}, 618549
IL1R1	97.7%	97.7%	100%	99.6%	?Chronic recurrent multifocal osteomyelitis 3, 259680
IL1RN	100%	100%	100%	99.8%	Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852;{Gastric cancer risk after H. pylori infection}, 613659;{Microvascular complications of diabetes 4}, 612628;Interleukin 1 receptor antagonist deficiency, 612852
IL2	100%	100%	100%	99.9%	
IL21	100%	100%	100%	100%	?Immunodeficiency, common variable, 11, 615767
IL21R	100%	100%	100%	99.3%	Immunodeficiency 56, 615207
IL2RA	100%	100%	100%	98.5%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367;{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RB	96.1%	96.1%	100%	98.5%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495

IL2RG	100%	99.8%	98.2%	68.6%	Combined immunodeficiency, X-linked, moderate, 312863;Severe combined immunodeficiency, X-linked, 300400
IL36RN	100%	100%	100%	99.5%	Psoriasis 14, pustular, 614204
IL6R	92.5%	92.5%	100%	99.2%	[Interleukin 6, serum level of, QTL], 614752;Hyper-IgE syndrome 5, autosomal recessive, with recurrent infections, 618944;[Interleukin-6 receptor, soluble, serum level of, QTL], 614689
IL6ST	100%	100%	100%	99.6%	Hyper-IgE syndrome 4A, autosomal dominant, with recurrent infections, 619752;Stuve-Wiedemann syndrome 2, 619751;Hyper-IgE syndrome 4B, autosomal recessive, with recurrent infections, 618523;?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750
IL7	100%	100%	100%	99.8%	{?Epidermodysplasia verruciformis, susceptibility to, 5}, 618309
IL7R	100%	100%	100%	99.6%	Immunodeficiency 104, severe combined, 608971

INO80	100%	100%	100%	99.4%	
INSR	100%	100%	100%	98.9%	Rabson-Mendenhall syndrome, 262190;Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549;Donohue syndrome, 246200;Hyperinsulinemic hypoglycemia, familial, 5, 609968
IPO8	100%	100%	100%	99.8%	VISS syndrome, 619472
IRAK1	100%	99.5%	98.2%	64.9%	
IRAK4	100%	100%	100%	99.9%	Immunodeficiency 67, 607676
IRF1	100%	100%	100%	99.3%	Nonsmall cell lung cancer, somatic, 211980;Gastric cancer, somatic, 613659;Immunodeficiency 117, mycobacteriosis, autosomal recessive, 620668
IRF2BP2	100%	100%	100%	94.9%	?Immunodeficiency, common variable, 14, 617765
IRF3	100%	100%	100%	99.1%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532
IRF4	100%	100%	100%	98.9%	[Skin/hair/eye pigmentation, variation in, 8], 611724

IRF7	100%	100%	100%	98.9%	?Immunodeficiency 39, 616345
IRF8	100%	100%	100%	99.1%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893;Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRF9	100%	100%	100%	99%	Immunodeficiency 65, susceptibility to viral infections, 618648
IRGM	100%	100%	100%	99.8%	{Mycobacterium tuberculosis, protection against}, 607948;{Inflammatory bowel disease (Crohn disease) 19}, 612278
ISG15	100%	100%	100%	99.5%	Immunodeficiency 38, 616126
ITCH	92.5%	92.4%	100%	99.8%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGB2	100%	100%	100%	99.2%	Leukocyte adhesion deficiency, 116920
ITK	100%	100%	100%	99.5%	Lymphoproliferative syndrome 1, 613011
ITPKB	100%	100%	100%	98.6%	

ITPR3	100%	100%	100%	98.8%	Charcot-Marie-Tooth disease, demyelinating, type 1J, 620111;{Diabetes, type 1, susceptibility to}, 222100
IVNS1ABP	100%	100%	100%	99.8%	Immunodeficiency 70, 618969
JAGN1	100%	100%	100%	99.7%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	100%	100%	100%	99.5%	Autoinflammation, immune dysregulation, and eosinophilia, 618999
JAK2	100%	100%	100%	99.5%	{Budd-Chiari syndrome, somatic}, 600880;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100;Leukemia, acute myeloid, somatic, 601626;Thrombocythemia 3, 614521;Polycythemia vera, somatic, 263300
JAK3	100%	100%	100%	97.9%	Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type, 600802
KDM6A	100%	99.8%	99.2%	74.3%	Kabuki syndrome 2, 300867
KMT2A	99.2%	99.2%	100%	99.2%	Wiedemann-Steiner syndrome, 605130

KMT2D	100%	100%	100%	98.6%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
KNG1	100%	100%	100%	99.6%	[Kininogen deficiency], 228960;Angioedema, hereditary, 6, 619363;[High molecular weight kininogen deficiency], 228960
KRAS	100%	100%	100%	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%	100%	100%	99.7%	Juvenile arthritis, 618795
LAMTOR2	100%	100%	100%	99.2%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAT	100%	100%	100%	99.3%	Immunodeficiency 52, 617514
LCK	100%	100%	100%	99.2%	Immunodeficiency 22, 615758
LCP1	100%	100%	100%	99.8%	
LCP2	100%	100%	100%	99.3%	Immunodeficiency 81, 619374
LIG1	100%	100%	100%	98.4%	Immunodeficiency 96, 619774
LIG4	100%	100%	100%	99.8%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
LPIN2	99.5%	99.2%	100%	99.5%	Majeed syndrome, 609628
LRBA	99.8%	99.8%	100%	99.7%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC32	100%	100%	100%	99.1%	Cleft palate, proliferative retinopathy, and developmental delay, 619074
LRRC8A	100%	100%	100%	98.6%	?Agammaglobulinemia 5, 613506
LSM11	100%	100%	100%	96.7%	?Aicardi-Goutieres syndrome 8, 619486

LYN	100%	100%	100%	99.6%	Autoinflammatory disease, systemic, with vasculitis, 620376
LYST	99.5%	99.4%	100%	99.8%	Chediak-Higashi syndrome, 214500
MAGT1	93.8%	93.5%	98.8%	72.1%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853;Congenital disorder of glycosylation, type Icc, 301031
MALT1	100%	100%	100%	99.6%	Immunodeficiency 12, 615468
MAN2B1	100%	100%	100%	98.3%	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	100%	100%	100%	99%	
MANBA	100%	100%	100%	99.7%	Mannosidosis, beta, 248510
MAP1LC3B2	100%	100%	100%	99.2%	
MAP3K14	100%	100%	100%	99%	Immunodeficiency 112, 620449
MAPK8	100%	100%	100%	99.8%	
MASP2	100%	100%	100%	99.5%	MASP2 deficiency, 613791
MC2R	100%	100%	100%	99.8%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM10	100%	100%	100%	99.5%	Immunodeficiency 80 with or without cardiomyopathy, 619313

MCM4	95.3%	95.3%	100%	99.6%	Immunodeficiency 54, 609981
MCTS1	100%	99.9%	99.2%	76.4%	Immunodeficiency 118, mycobacteriosis, 301115
MEFV	96.1%	96.1%	100%	99.3%	Neutrophilic dermatosis, acute febrile, 608068;Familial Mediterranean fever, AR, 249100;Familial Mediterranean fever, AD, 134610
MOGS	100%	100%	100%	99.3%	Congenital disorder of glycosylation, type IIb, 606056
MPEG1	100%	100%	100%	99%	Immunodeficiency 77, 619223
MRTFA	100%	100%	100%	98.7%	?Immunodeficiency 66, 618847
MS4A1	100%	100%	100%	99.7%	?Immunodeficiency, common variable, 5, 613495
MSN	100%	98.3%	99.1%	74.4%	Immunodeficiency 50, 300988
MTHFD1	100%	100%	100%	99.4%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780

MVK	100%	100%	100%	99.2%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
MYD88	100%	100%	100%	99.7%	Macroglobulinemia, Waldenstrom, somatic, 153600;Immunodeficiency 68, 612260
MYOF	100%	100%	100%	99.4%	?Angioedema, hereditary, 7, 619366
MYSM1	100%	100%	100%	99.8%	Bone marrow failure syndrome 4, 618116
NBAS	100%	99.9%	100%	99.7%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483
NBN	97.5%	97.5%	100%	99.9%	Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260
NCF1	100%	99.7%	100%	97.3%	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	100%	100%	100%	99.5%	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100%	100%	100%	99%	Chronic granulomatous disease 3, autosomal recessive, 613960

NCKAP1L	100%	100%	100%	99.5%	Immunodeficiency 72 with autoinflammation, 618982
NCSTN	100%	100%	100%	98.8%	Acne inversa, familial, 1, 142690
NFAT5	100%	100%	100%	99.8%	
NFATC1	100%	100%	100%	96.8%	
NFE2L2	81.2%	81.2%	100%	99.4%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFKB1	100%	100%	100%	99.5%	Immunodeficiency, common variable, 12, 616576
NFKB2	100%	100%	100%	98.7%	Immunodeficiency, common variable, 10, 615577
NFKBIA	100%	100%	100%	99.4%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHEJ1	100%	100%	100%	99.4%	Microphthalmia/coloboma 13, 620968;Immunodeficiency 124, severe combined, 611291
NHP2	100%	100%	100%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987
NLRC4	100%	100%	100%	99.8%	?Familial cold autoinflammatory syndrome 4, 616115;Autoinflammation with infantile enterocolitis, 616050

NLRP1	98.1%	98.1%	100%	99.4%	{Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579;?Respiratory papillomatosis, juvenile recurrent, congenital, 618803;Autoinflammation with arthritis and dyskeratosis, 617388;Palmoplantar carcinoma, multiple self-healing, 615225
NLRP12	100%	100%	100%	99.1%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	100%	100%	100%	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%	100%	100%	99.6%	
NOD2	100%	100%	100%	99.6%	Blau syndrome, 186580;{Yao syndrome}, 617321;{Inflammatory bowel disease 1, Crohn disease}, 266600

NOP10	92.5%	92.5%	100%	98.8%	?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%	100%	100%	99.2%	{Malaria, resistance to}, 611162
NRAS	100%	100%	100%	99.6%	Noonan syndrome 6, 613224;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500
NSMCE3	100%	100%	100%	98.4%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241

NUDCD3	100%	100%	100%	99.7%	
OAS1	100%	100%	100%	99%	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinemia, 618042
ORAI1	100%	100%	100%	97.8%	Immunodeficiency 9, 612782;Myopathy, tubular aggregate, 2, 615883
OSTM1	99.8%	98.5%	100%	99.5%	Osteopetrosis, autosomal recessive 5, 259720
OTULIN	100%	100%	100%	99.2%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099;{Immunodeficiency 107, susceptibility to invasive staphylococcus aureus infection}, 619986
PARN	97.3%	95.3%	100%	99.7%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
PAX1	100%	100%	100%	97.3%	Otofaciocervical syndrome 2 with T-cell deficiency, 615560
PAX5	100%	100%	100%	99.1%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545

PBX1	100%	100%	100%	99%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	100%	100%	100%	99.8%	Propionicacidemia, 606054
PCCB	99.2%	96.1%	100%	99.7%	Propionicacidemia, 606054
PDCD1	100%	100%	100%	98.6%	?Autoimmune disease with susceptibility to mycobacterium tuberculosis, 621004
PEPD	93.9%	93.9%	100%	99.4%	Prolidase deficiency, 170100
PEX16	100%	100%	100%	99.1%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PGM3	100%	100%	100%	99.6%	Immunodeficiency 23, 615816
PI4KA	100%	99.7%	100%	99%	Spastic paraplegia 84, autosomal recessive, 619621;Gastrointestinal defects and immunodeficiency syndrome 2, 619708;Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531

PIGA	100%	99.9%	98.4%	74%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIK3CD	100%	100%	100%	98.8%	Immunodeficiency 14A, autosomal dominant, 615513;Immunodeficiency 14B, autosomal recessive, 619281;?Roifman-Chitayat syndrome, digenic, 613328
PIK3CG	100%	100%	100%	99.3%	Immunodeficiency 97 with autoinflammation, 619802
PIK3R1	100%	99.9%	100%	99.9%	Immunodeficiency 36, 616005;?Agammaglobulinemia 7, autosomal recessive, 615214;SHORT syndrome, 269880
PLCG2	100%	100%	100%	99.3%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878;Familial cold autoinflammatory syndrome 3, 614468
PLEKHM1	100%	100%	100%	98.6%	?Osteopetrosis, autosomal recessive 6, 611497;Osteopetrosis, autosomal dominant 3, 618107

PLG	100%	100%	100%	99.6%	Dysplasminogenemia, 217090;Angioedema, hereditary, 4, 619360;Plasminogen deficiency, type I, 217090
PMM2	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMVK	100%	100%	100%	99.2%	Porokeratosis 1, multiple types, 175800
PNP	100%	100%	100%	99.2%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POLA1	100%	99.8%	99.2%	74.4%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220;Van Esch-O'Driscoll syndrome, 301030
POLD1	100%	100%	100%	98.3%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591
POLD3	100%	100%	100%	99.8%	Immunodeficiency 122, 620869
POLE2	100%	100%	100%	99.9%	

POLR3F	91.6%	90.5%	100%	99.9%	?Immunodeficiency 101 (varicella zoster virus-specific), 619872
POMP	83.2%	83.2%	100%	99.9%	Proteasome-associated autoinflammatory syndrome 2, 618048;Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POT1	100%	100%	100%	99.8%	Tumor predisposition syndrome 3, 615848;?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368;?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367
POU2AF1	100%	100%	100%	99.1%	
PRF1	100%	100%	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PRIM1	91.8%	91.8%	100%	99.4%	Primordial dwarfism-immunodeficiency-lipodystrophy syndrome, 620005
PRKCD	100%	100%	100%	99.2%	Autoimmune lymphoproliferative syndrome, type III, 615559

PRKDC	100%	100%	100%	99.6%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	100%	99.9%	98.5%	70.4%	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%	100%	96.6%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMA3	100%	100%	100%	99.6%	
PSMB10	100%	100%	100%	99.5%	Immunodeficiency 121 with autoinflammation, 620807;Proteasome-associated autoinflammatory syndrome 5, 619175
PSMB4	100%	100%	100%	99.8%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	100%	100%	100%	99.7%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040

PSMB9	100%	100%	100%	99.4%	Proteasome-associated autoinflammatory syndrome 6, 620796
PSMG2	88.4%	88.4%	100%	99.9%	?Proteasome-associated autoinflammatory syndrome 4, 619183
PSTPIP1	100%	100%	100%	99.4%	Autoinflammatory syndrome with cytopenia, hyperzincemia, and hypercalprotectinemia, 601979;Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCRA	100%	100%	100%	99.1%	Immunodeficiency 126, 620931
PTEN	94.5%	94.5%	99.9%	96.5%	{Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309
PTPN22	100%	100%	100%	99.8%	{Rheumatoid arthritis, susceptibility to}, 180300;{Systemic lupus erythematosus susceptibility to}, 152700;{Diabetes, type 1, susceptibility to}, 222100
PTPRC	100%	100%	100%	99.7%	Immunodeficiency 105, severe combined, 619924

RAB27A	100%	100%	100%	99.5%	Griscelli syndrome, type 2, 607624
RAC2	100%	100%	100%	97.9%	Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203;?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987;Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986
RAG1	100%	100%	100%	99.4%	Omenn syndrome, 603554;Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889

RAG2	100%	100%	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Omenn syndrome, 603554
RANBP2	100%	100%	100%	99.8%	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033
RASGRP1	95%	95%	100%	99.5%	Immunodeficiency 64, 618534
RASGRP2	100%	100%	100%	98.6%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	100%	100%	100%	97.5%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RC3H1	100%	100%	100%	99.4%	?Immune dysregulation and systemic hyperinflammation syndrome, 618998
RECQL4	100%	100%	100%	98.9%	Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280
REL	100%	99.4%	100%	99.8%	Immunodeficiency 92, 619652
RELA	100%	100%	100%	98.8%	Autoinflammatory disease, familial, Behcet-like-3, 618287

RELB	100%	99.8%	100%	96.4%	?Immunodeficiency 53, 617585
RFX5	100%	100%	100%	99.3%	?MHC class II deficiency 5, 620818;MHC class II deficiency 3, 620816
RFXANK	100%	100%	100%	98.2%	MHC class II deficiency 2, 620815
RFXAP	100%	100%	100%	96.5%	MHC class II deficiency 4, 620817
RGS10	100%	100%	100%	99.4%	
RHBDF2	100%	99.9%	99.9%	97.5%	Tylosis with esophageal cancer, 148500
RHOG	100%	100%	100%	99.4%	
RHOH	100%	100%	100%	99.5%	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307
RIGI	98.6%	98.6%	100%	99.6%	Singleton-Merten syndrome 2, 616298
RIPK1	100%	100%	100%	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%	100%	100%	99.2%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91.4%	91.4%	100%	99.2%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	100%	100%	98.1%	Aicardi-Goutieres syndrome 3, 610329
RNF168	100%	100%	100%	99.4%	RIDDLE syndrome, 611943
RNF31	100%	100%	100%	98.8%	Immunodeficiency 115 with autoinflammation, 620632
RNU4ATAC					Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RNU7-1					Aicardi-Goutieres syndrome 9, 619487
RORC	100%	100%	100%	99.1%	Immunodeficiency 42, 616622
RPA1	100%	100%	100%	99.3%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767
RPSA	100%	100%	100%	100%	Asplenia, isolated congenital, 271400
RSPH9	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 12, 612650

RTEL1	100%	100%	100%	99.1%	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%	100%	100%	99.6%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SAMD9L	100%	100%	100%	99.8%	Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SAMHD1	100%	100%	100%	99.8%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SASH3	100%	99.7%	98%	70.2%	Immunodeficiency 102, 301082
SAT1	100%	100%	99.6%	74%	

SBDS	100%	100%	100%	99.7%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400
SEC61A1	100%	100%	100%	99.6%	Immunodeficiency, common variable, 15, 620670;?Neutropenia, severe congenital, 11, autosomal dominant, 620674;Tubulointerstitial kidney disease, autosomal dominant, 5, 617056
SEMA3E	100%	100%	100%	99.7%	
SERAC1	100%	100%	100%	99.8%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	100%	100%	100%	99.5%	Angioedema, hereditary, 1 and 2, 106100;Complement component 4, partial deficiency of, 120790
SH2B3	100%	100%	100%	98.7%	Thrombocythemia, somatic, 187950;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100
SH2D1A	100%	100%	99.1%	73.8%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	99.9%	99%	100%	98.8%	Cherubism, 118400

SH3KBP1	98.8%	98.4%	99%	72.4%	?Immunodeficiency 61, 300310
SHARPIN	100%	100%	100%	99.5%	Autoinflammation with episodic fever and immune dysregulation, 620795
SKIC2	100%	100%	100%	99.2%	Trichohepatoenteric syndrome 2, 614602
SKIC3	98.9%	98.9%	100%	99.6%	Trichohepatoenteric syndrome 1, 222470
SLC19A1	100%	100%	100%	97.9%	Immunodeficiency 114, folate-responsive, 620603;?Megaloblastic anemia, folate-responsive, 601775
SLC29A3	100%	100%	100%	99.3%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	100%	100%	100%	99.2%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	100%	100%	100%	99.5%	Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240
SLC39A4	100%	100%	100%	99.5%	Acrodermatitis enteropathica, 201100

SLC39A7	100%	100%	100%	98.6%	Agammaglobulinemia 9, autosomal recessive, 619693
SLC46A1	100%	100%	100%	98.7%	Folate malabsorption, hereditary, 229050
SLC7A7	100%	100%	100%	97.5%	Lysinuric protein intolerance, 222700
SMARCAL1	100%	100%	100%	99.4%	Schimke immunoosseous dysplasia, 242900
SMARCD2	100%	100%	100%	98.3%	Specific granule deficiency 2, 617475
SNORA31					{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 10}, 619396
SNX10	89.3%	89.3%	100%	99.8%	Osteopetrosis, autosomal recessive 8, 615085
SOCS1	100%	100%	100%	94.4%	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375
SOCS4	100%	100%	100%	99.5%	
SP110	100%	100%	100%	99.3%	{Mycobacterium tuberculosis, susceptibility to}, 607948;Hepatic venoocclusive disease with immunodeficiency, 235550
SPI1	100%	100%	100%	98.3%	Agammaglobulinemia 10, autosomal dominant, 619707

SPINK5	100%	100%	100%	99.8%	Netherton syndrome, 256500
SPPL2A	100%	100%	100%	99.9%	Immunodeficiency 86, mycobacteriosis, 619549
SRP54	100%	100%	100%	99.8%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	100%	100%	100%	99.5%	Bone marrow failure syndrome 1, 614675
STAT1	95.9%	95.9%	100%	99.7%	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%	100%	99.4%	Pseudo-TORCH syndrome 3, 618886;Immunodeficiency 44, 616636
STAT3	100%	100%	100%	99.7%	Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060;Autoimmune disease, multisystem, infantile-onset, 1, 615952

STAT4	100%	100%	100%	99.9%	Disabling pansclerotic morphea of childhood, 620443;{Systemic lupus erythematosus, susceptibility to, 11}, 612253
STAT5B	100%	100%	100%	99.2%	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590;Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985
STAT6	100%	100%	100%	98.6%	Hyper-IgE syndrome 6, autosomal dominant, with recurrent infections, 620532
STIM1	100%	99.8%	100%	99.2%	Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
STING1	100%	100%	100%	99.4%	STING-associated vasculopathy, infantile-onset, 615934
STK4	100%	100%	100%	99.8%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	100%	100%	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552

STXBP2	100%	100%	100%	98.2%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
SYK	100%	100%	100%	99.8%	Immunodeficiency 82 with systemic inflammation, 619381
TAFAZZIN	100%	99.6%	97.2%	66.7%	Barth syndrome, 302060
TAP1	99.7%	97.1%	100%	99.5%	MHC class I deficiency 1, 604571
TAP2	98.1%	97.9%	100%	98%	MHC class I deficiency 2, 620813
TAPBP	88.8%	88.8%	100%	98%	?MHC class I deficiency 3, 620814
TBX1	96.8%	93%	99.7%	89.1%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430
TBX21	100%	100%	100%	98.3%	Asthma and nasal polyps, 208550;?Immunodeficiency 88, 619630;{Asthma, aspirin-induced, susceptibility to}, 208550
TCF3	100%	100%	100%	99%	Agammaglobulinemia 8B, autosomal recessive, 619824;Agammaglobulinemia 8A, autosomal dominant, 616941

TCIRG1	100%	100%	100%	99%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	94.2%	94.2%	100%	99.1%	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%	100%	100%	98.4%	Dyskeratosis congenita, autosomal dominant 2, 613989;Dyskeratosis congenita, autosomal recessive 4, 613989;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742;{Melanoma, cutaneous malignant, 9}, 615134;{Leukemia, acute myeloid}, 601626
TET2	100%	100%	100%	99.6%	Myelodysplastic syndrome, somatic, 614286;Immunodeficiency 75, 619126
TFRC	95.5%	95.5%	100%	99.2%	Immunodeficiency 46, 616740

TGFB1	100%	99.8%	100%	96.7%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213;Camurati-Engelmann disease, 131300;{Cystic fibrosis lung disease, modifier of}, 219700
THBD	100%	100%	100%	96.6%	Thrombophilia 12 due to thrombomodulin defect, 614486;{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TICAM1	100%	100%	100%	99.4%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850
TINF2	100%	100%	100%	99.4%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
TIRAP	100%	100%	100%	99.5%	{Malaria, protection against}, 611162;{Tuberculosis, protection against}, 607948;{Bacteremia, protection against}, 614382
TLR3	100%	100%	100%	100%	{HIV1 infection, resistance to}, 609423;{Immunodeficiency 83, susceptibility to viral infections}, 613002
TLR4	100%	100%	100%	99.8%	

TLR5	100%	100%	100%	99.7%	{Melioidosis, susceptibility to}, 615557;{Systemic lupus erythematosus, susceptibility to, 1}, 601744;{Systemic lupus erythematosus, resistance to}, 601744;{Legionnaire disease, susceptibility to}, 608556
TLR7	100%	100%	99.1%	75.2%	Immunodeficiency 74, COVID19-related, X-linked, 301051;Systemic lupus erythematosus 17, 301080
TLR8	100%	100%	99.1%	75.4%	Immunodeficiency 98 with autoinflammation, X-linked, 301078
TMC6	100%	100%	100%	98.8%	{Epidermodysplasia verruciformis, susceptibility to, 1}, 226400
TMC8	100%	100%	100%	99%	{Epidermodysplasia verruciformis, susceptibility to, 2}, 618231
TNFAIP3	100%	100%	100%	99.5%	Autoinflammatory syndrome, familial, Behcet-like 1, 616744
TNFRSF11A	99.9%	99.2%	100%	98.3%	Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810

TNFRSF13B	100%	100%	100%	98.6%	Immunodeficiency, common variable, 2, 240500;Immunoglobulin A deficiency 2, 609529
TNFRSF13C	100%	100%	100%	98.9%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	100%	100%	100%	98.7%	{Multiple sclerosis, susceptibility to, 5}, 614810;Periodic fever, familial, 142680
TNFRSF4	100%	100%	100%	98%	?Immunodeficiency 16, 615593
TNFRSF9	100%	100%	100%	99.7%	Immunodeficiency 109 with lymphoproliferation, 620282
TNFSF11	100%	100%	100%	98.6%	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	100%	100%	100%	99.2%	
TNFSF13	100%	99.9%	100%	98.7%	
TOM1	100%	100%	100%	99.3%	?Immunodeficiency 85 and autoimmunity, 619510
TOP2B	100%	100%	100%	99.5%	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296
TPP2	100%	100%	100%	99.7%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220
TRAC	100%	100%	100%	99.2%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387

TRAF3	100%	100%	100%	97.9%	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849
TRAF3IP2	99.7%	97.9%	100%	99.4%	?Candidiasis, familial, 8, 615527;{Psoriasis susceptibility 13}, 614070
TREX1	100%	100%	100%	99.7%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448
TRIM22	100%	100%	100%	99.4%	
TRNT1	91.9%	91.8%	100%	99.9%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959
TTC7A	100%	100%	100%	98.8%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	100%	100%	100%	98.1%	Immunodeficiency 35, 611521

UBA1	99.9%	99.3%	98.7%	68.8%	Spinal muscular atrophy, X-linked 2, infantile, 301830;VEXAS syndrome, somatic, 301054
UNC13D	100%	100%	100%	99.2%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	100%	99.6%	100%	95.9%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551
UNG	96.5%	96.4%	100%	99%	Immunodeficiency with hyper IgM, type 5, 608106
USB1	95.5%	93.2%	100%	99.2%	Poikiloderma with neutropenia, 604173
USP18	100%	100%	100%	98.7%	Pseudo-TORCH syndrome 2, 617397
VAV1	98.3%	98.3%	100%	98.6%	
VPS13B	100%	100%	100%	99.7%	Cohen syndrome, 216550
VPS45	87.3%	86.8%	100%	99.6%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	97.7%	89.9%	97.7%	65.4%	Wiskott-Aldrich syndrome, 301000;Neutropenia, severe congenital, X-linked, 300299;Thrombocytopenia, X-linked, intermittent, 313900;Thrombocytopenia, X-linked, 313900

WDR1	100%	100%	100%	98.7%	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550
WIPF1	100%	100%	100%	98.8%	Wiskott-Aldrich syndrome 2, 614493
WRAP53	100%	100%	100%	98.8%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	100%	99.9%	99.2%	74%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	100%	100%	100%	99.3%	Immunodeficiency 48, 269840;Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB24	100%	100%	100%	99.1%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZNF341	100%	100%	100%	98.5%	Hyper-IgE syndrome 3, autosomal recessive, with recurrent infections, 618282
ZNFX1	100%	100%	100%	99.5%	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 4.0.0

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