PRIMARY IMMUNODEFICIENCY GENE PANEL DG 2.18 (419 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ACD	100%	99,90%	100%	100%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACP5	99,80%	98,30%	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACTB	99,70%	96,10%	100%	100%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADA	100%	99,70%	100%	100%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	100%	99,00%	100%	100%	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	99,90%	99,00%	100%	100%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAR	100%	99,80%	100%	100%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	100%	100%	100%	100%	Aspartylglucosaminuria, 208400
AICDA	100%	100%	100%	100%	Immunodeficiency with hyper-IgM, type 2, 605258
AIRE	100%	99,80%	100%	100%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK2	98,80%	94,50%	100%	100%	Reticular dysgenesis, 267500
ALG13	98,40%	92,60%	100%	99,60%	Epileptic encephalopathy, early infantile, 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
ALPI	100%	99,50%	100%	100%	No OMIM disease ID
AP1S3	90,40%	90,10%	90,50%	90,50%	No OMIM disease ID
AP3B1	99,20%	95,80%	100%	100%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	99,80%	98,60%	100%	100%	?Hermansky-Pudlak syndrome 10, 617050
APOL1	100%	100%	100%	100%	No OMIM disease ID
ARHGEF1	99,90%	98,40%	100%	100%	?Immunodeficiency 62, 618459
ARPC1B	100%	100%	100%	100%	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
ATM	99,80%	98,10%	100%	100%	Ataxia-telangiectasia, 208900 Lymphoma, mantle cell, somatic, 0

					Lymphoma, B-cell non-Hodgkin, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0
ATP6AP1	98,20%	92,10%	100%	100%	Immunodeficiency 47, 300972
B2M	100%	100%	100%	100%	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
BACH2	100%	100%	100%	100%	Immunodeficiency 60, 618394
BCL10	100%	100%	100%	100%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245
BCL11B	99,10%	95,60%	98,80%	97,30%	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BLK	100%	100%	100%	100%	Maturity-onset diabetes of the young, type 11, 613375
BLM	99,80%	98,30%	100%	100%	Bloom syndrome, 210900
BLNK	97,10%	95,50%	100%	100%	?Agammaglobulinemia 4, 613502
BLOC1S6	99,90%	97,10%	94,90%	94,90%	?Hermansky-pudlak syndrome 9, 614171
BTK	100%	99,90%	100%	99,90%	Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 Agammaglobulinemia, X-linked 1, 300755
C17orf62	99,60%	97,00%	100%	100%	No OMIM disease ID
C1QA	100%	100%	100%	100%	C1q deficiency, 613652
C1QB	100%	100%	100%	100%	C1q deficiency, 613652
C1QC	100%	99,20%	100%	100%	C1q deficiency, 613652
C1R	100%	100%	99,00%	96,90%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	99,90%	99,00%	99,50%	97,70%	Ehlers-Danlos syndrome, periodontal type, 2, 617174 C1s deficiency, 613783
C2	100%	100%	100%	100%	C2 deficiency, 217000
C3	99,90%	99,20%	100%	100%	C3 deficiency, 613779
C5	99,90%	98,50%	100%	100%	C5 deficiency, 609536
C6	100%	99,70%	100%	100%	C6 deficiency, 612446 Combined C6/C7 deficiency, 0
C7	100%	98,90%	100%	100%	C7 deficiency, 610102
C8A	100%	99,60%	100%	100%	C8 deficiency, type I, 613790
C8B	100%	99,20%	100%	100%	C8 deficiency, type II, 613789
C8G	100%	100%	100%	100%	No OMIM disease ID
C9	99,90%	99,50%	100%	100%	C9 deficiency, 613825
CA2	100%	100%	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730

CARD11	100%	99,90%	100%	100%	Immunodeficiency 11B with atopic dermatitis, 617638 B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206
CARD14	100%	99,10%	100%	100%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	99,90%	98,40%	100%	100%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	96,30%	94,50%	99,70%	98,20%	Immunodeficiency 58, 618131
CASP10	99,50%	97,30%	100%	100%	Lymphoma, non-Hodgkin, somatic, 605027 Gastric cancer, somatic, 613659 Autoimmune lymphoproliferative syndrome, type II, 603909
CASP8	95,60%	95,40%	95,60%	95,60%	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550
CAVIN1	100%	100%	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
CCBE1	99,80%	98,80%	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD19	100%	100%	100%	100%	Immunodeficiency, common variable, 3, 613493
CD247	100%	100%	100%	100%	?Immunodeficiency 25, 610163
CD27	99,90%	96,90%	100%	100%	Lymphoproliferative syndrome 2, 615122
CD3D	100%	100%	100%	100%	Immunodeficiency 19, 615617
CD3E	100%	99,50%	100%	100%	Immunodeficiency 18, SCID variant, 615615 Immunodeficiency 18, 615615
CD3G	100%	100%	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	100%	100%	100%	100%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	97,30%	88,10%	100%	100%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	99,90%	99,40%	100%	100%	No OMIM disease ID
CD55	92,20%	84,30%	99,20%	97,10%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	95,10%	86,60%	79,50%	79,50%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	99,80%	97,70%	100%	100%	Lymphoproliferative syndrome 3, 618261
CD79A	100%	100%	100%	100%	Agammaglobulinemia 3, 613501
CD79B	100%	100%	100%	100%	Agammaglobulinemia 6, 612692
CD81	100%	99,90%	100%	100%	Immunodeficiency, common variable, 6, 613496
CD8A	100%	99,80%	100%	100%	CD8 deficiency, familial, 608957
CDCA7	100%	99,60%	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDKN2B	100%	99,90%	100%	100%	No OMIM disease ID
CEBPE	100%	100%	100%	100%	Specific granule deficiency, 245480
CFB	100%	100%	100%	100%	?Complement factor B deficiency, 615561
CFD	89,30%	83,70%	100%	100%	Complement factor D deficiency, 613912

CFH	99,90%	99,00%	100%	99,90%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	96,40%	94,90%	95,40%	93,80%	No OMIM disease ID
CFHR2	99,80%	97,60%	100%	100%	No OMIM disease ID
CFHR3	94,00%	92,20%	96,00%	95,20%	No OMIM disease ID
CFHR4	100%	99,90%	100%	100%	No OMIM disease ID
CFHR5	99,60%	98,40%	100%	100%	Nephropathy due to CFHR5 deficiency, 614809
CFI	99,20%	96,80%	100%	100%	Complement factor I deficiency, 610984
CFP	100%	99,00%	100%	100%	Properdin deficiency, X-linked, 312060
CFTR	99,60%	97,90%	100%	100%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF, 0
CHD7	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CIB1	97,30%	93,60%	100%	100%	Epidermodysplasia verruciformis 3, 618267
CIITA	100%	99,50%	100%	100%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CLCN7	99,70%	98,40%	100%	100%	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600 Hypopigmentation, organomegaly, and delayed myelination and development, 618541
CLEC4D	100%	99,80%	100%	100%	No OMIM disease ID
CLEC7A	100%	100%	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108
CLPB	100%	99,90%	100%	100%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COLEC11	100%	100%	100%	100%	3MC syndrome 2, 265050
COPA	100%	99,20%	100%	100%	No OMIM disease ID
CORO1A	100%	98,60%	100%	100%	Immunodeficiency 8, 615401
CR2	100%	99,80%	100%	100%	Immunodeficiency, common variable, 7, 614699
CREBBP	99,70%	98,50%	100%	100%	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CSF2RA	89,90%	87,50%	95,60%	92,10%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	100%	99,00%	100%	100%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	99,60%	98,20%	100%	100%	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	100%	99,60%	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100%	100%	100%	100%	Autoimmune lymphoproliferative syndrome, type V, 616100
CTPS1	100%	100%	100%	100%	Immunodeficiency 24, 615897
CTSC	100%	100%	100%	100%	Periodontitis 1, juvenile, 170650 Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010

CXCR4	100%	100%	100%	100%	WHIM syndrome, 193670 Myelokathexis, isolated, 0
CYBA	95,00%	82,40%	100%	100%	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690
CYBB	99,90%	99,30%	100%	100%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400
DBR1	100%	99,30%	100%	100%	No OMIM disease ID
DCLRE1C	100%	99,40%	100%	100%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDX58	99,90%	99,00%	100%	100%	Singleton-Merten syndrome 2, 616298
DEF6	96,70%	93,80%	100%	99,90%	No OMIM disease ID
DHFR	92,10%	78,90%	100%	100%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	99,80%	98,70%	100%	99,70%	Dyskeratosis congenita, X-linked, 305000
DNASE1	100%	99,90%	100%	100%	No OMIM disease ID
DNASE1L3	100%	100%	100%	100%	Systemic lupus erythematosus 16, 614420
DNASE2	99,70%	97,10%	100%	100%	No OMIM disease ID
DNMT3B	100%	100%	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	100%	99,60%	100%	100%	Immunodeficiency 40, 616433
DOCK8	100%	99,60%	100%	100%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
ELANE	99,70%	97,40%	100%	100%	Neutropenia, severe congenital 1, autosomal dominant, 202700 Neutropenia, cyclic, 162800
ELF4	100%	99,70%	100%	100%	No OMIM disease ID
EPG5	99,50%	98,50%	100%	100%	Vici syndrome, 242840
ERCC2	100%	99,70%	100%	100%	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	100%	99,40%	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
EXTL3	100%	100%	100%	100%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
F12	99,90%	98,80%	100%	100%	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
FAAP24	99,30%	96,70%	100%	100%	No OMIM disease ID
FADD	100%	100%	100%	100%	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAS	100%	99,60%	100%	100%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0
FASLG	100%	99,60%	100%	100%	Autoimmune lymphoproliferative syndrome, type IB, 601859

FAT4	100%	100%	100%	100%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FCGR1A	46,80%	44,10%	100%	100%	No OMIM disease ID
FCGR2A	100%	100%	100%	100%	No OMIM disease ID
FCGR2B	99,50%	95,40%	100%	100%	No OMIM disease ID
FCGR3A	99,00%	97,10%	100%	100%	Immunodeficiency 20, 615707
FCGR3B	99,40%	98,20%	98,10%	98,00%	Neutropenia, alloimmune neonatal, 0
FCHO1	98,90%	97,70%	100%	100%	No OMIM disease ID
FCN3	100%	100%	100%	100%	Immunodeficiency due to ficolin 3 deficiency, 613860
FERMT3	100%	100%	100%	100%	Leukocyte adhesion deficiency, type III, 612840
FOXN1	100%	99,60%	100%	100%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	99,20%	95,50%	100%	100%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FPR1	100%	100%	100%	100%	No OMIM disease ID
G6PC	100%	100%	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	100%	99,90%	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	99,30%	98,10%	100%	99,30%	Hemolytic anemia, G6PD deficient (favism), 300908
GATA2	100%	98,30%	100%	100%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GFI1	100%	99,20%	100%	100%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GINS1	99,30%	94,90%	100%	100%	Immunodeficiency 55, 617827
GJC2	78,20%	58,70%	96,90%	91,40%	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GRHL2	100%	100%	100%	100%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GTF2H5	100%	99,60%	100%	100%	Trichothiodystrophy 3, photosensitive, 616395
HAVCR2	100%	100%	100%	100%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100%	100%	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HELLS	97,80%	92,10%	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HMOX1	98,40%	89,90%	100%	100%	Heme oxygenase-1 deficiency, 614034
HYOU1	100%	99,50%	100%	100%	?Immunodeficiency 59 and hypoglycemia, 233600
ICOS	99,90%	99,80%	100%	100%	Immunodeficiency, common variable, 1, 607594
ICOSLG	99,50%	98,80%	100%	100%	No OMIM disease ID

IFIH1	99,70%	98,40%	100%	100%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFNAR1	100%	99,30%	100%	100%	No OMIM disease ID
IFNAR2	100%	99,70%	100%	100%	?Immunodeficiency 45, 616669
IFNGR1	100%	99,40%	100%	100%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978
IFNGR2	93,30%	93,20%	100%	99,80%	Immunodeficiency 28, mycobacteriosis, 614889
IGHM	100%	100%	100%	100%	Agammaglobulinemia 1, 601495
IGLL1	99,90%	96,90%	100%	100%	Agammaglobulinemia 2, 613500
IKBKB	99,70%	96,50%	97,20%	97,20%	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IKBKG	84,10%	77,20%	100%	100%	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Immunodeficiency, isolated, 300584 Ectodermal dysplasia and immunodeficiency 1, 300291 Invasive pneumococcal disease, recurrent isolated, 2, 300640 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301
IKZF1	100%	100%	100%	100%	Immunodeficiency, common variable, 13, 616873
IL10	99,80%	98,20%	100%	100%	No OMIM disease ID
IL10RA	100%	100%	100%	100%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	99,80%	98,00%	100%	100%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL12B	100%	99,30%	100%	100%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	98,90%	96,30%	94,10%	94,10%	Immunodeficiency 30, 614891
IL17F	99,90%	97,20%	100%	100%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100%	99,40%	100%	100%	Immunodeficiency 51, 613953
IL17RC	100%	99,90%	100%	100%	Candidiasis, familial, 9, 616445
IL18BP	100%	100%	100%	100%	No OMIM disease ID
IL1RN	100%	100%	100%	100%	Interleukin 1 receptor antagonist deficiency, 612852
IL2	94,50%	88,00%	100%	100%	No OMIM disease ID
IL21	99,40%	95,70%	100%	100%	?Immunodeficiency, common variable, 11, 615767
IL21R	100%	100%	100%	100%	Immunodeficiency 56, 615207
IL2RA	100%	99,70%	100%	100%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL2RB	100%	99,70%	100%	100%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL2RG	99,80%	97,10%	100%	100%	Severe combined immunodeficiency, X-linked, 300400 Combined immunodeficiency, X-linked, moderate, 312863
IL36RN	100%	100%	100%	100%	Psoriasis 14, pustular, 614204
IL6R	98,40%	94,20%	92,80%	92,70%	No OMIM disease ID

IL6ST	96,40%	90,30%	100%	100%	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IL7R	100%	99,80%	100%	100%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
INO80	100%	99,10%	100%	100%	No OMIM disease ID
INSR	97,80%	94,70%	99,90%	99,20%	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200
MR *E11*	98,90%	93,30%	100%	100%	Ataxia-telangiectasia-like disorder 1, 604391
IRAK1	99,30%	94,90%	99,90%	99,40%	No OMIM disease ID
IRAK4	99,80%	97,70%	100%	100%	IRAK4 deficiency, 607676 Invasive pneumococcal disease, recurrent isolated, 1, 610799
IRF2BP2	93,90%	77,70%	100%	99,90%	?Immunodeficiency, common variable, 14, 617765
IRF3	100%	99,80%	100%	100%	No OMIM disease ID
IRF4	100%	100%	100%	100%	No OMIM disease ID
IRF7	100%	99,90%	100%	100%	?Immunodeficiency 39, 616345
IRF8	99,00%	95,70%	100%	100%	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893
IRF9	100%	100%	100%	100%	Immunodeficiency 65, susceptibility to viral infections, 618648
ISG15	100%	100%	100%	100%	Immunodeficiency 38, 616126
ITCH	95,40%	95,20%	100%	99,00%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGB2	100%	100%	100%	100%	Leukocyte adhesion deficiency, 116920
ITK	100%	98,90%	100%	100%	Lymphoproliferative syndrome 1, 613011
JAGN1	100%	100%	100%	100%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	100%	99,80%	100%	100%	No OMIM disease ID
JAK2	98,10%	95,80%	100%	100%	Myelofibrosis, somatic, 254450 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300 Leukemia, acute myeloid, somatic, 601626 Erythrocytosis, somatic, 133100
JAK3	99,90%	98,70%	100%	100%	SCID, autosomal recessive, T-negative/B-positive type, 600802
KDM6A	96,10%	88,70%	100%	99,90%	Kabuki syndrome 2, 300867
KMT2D	100%	99,40%	100%	100%	Kabuki syndrome 1, 147920
LACC1	100%	99,40%	100%	100%	Juvenile arthritis, 618795
LAMTOR2	100%	99,70%	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAT	100%	99,20%	100%	100%	Immunodeficiency 52, 617514
LCK	98,90%	96,60%	100%	100%	?Immunodeficiency 22, 615758

LIG1	100%	99,70%	100%	100%	No OMIM disease ID
LIG4	100%	99,90%	100%	100%	LIG4 syndrome, 606593
LPIN2	100%	100%	100%	100%	Majeed syndrome, 609628
LRBA	99,90%	99,60%	100%	100%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRRC8A	100%	99,80%	100%	100%	?Agammaglobulinemia 5, 613506
LTBP3	99,60%	98,10%	100%	100%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LYST	99,60%	98,30%	100%	100%	Chediak-Higashi syndrome, 214500
MAGT1	98,50%	96,50%	98,70%	98,70%	Congenital disorder of glycosylation, type Icc, 301031 Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853
MAL2	99,90%	98,00%	100%	99,90%	No OMIM disease ID
MALT1	91,20%	89,40%	100%	100%	Immunodeficiency 12, 615468
MAN2B1	99,80%	97,90%	100%	100%	Mannosidosis, alpha-, types I and II, 248500
MANBA	99,80%	98,40%	100%	100%	Mannosidosis, beta, 248510
MAP3K14	99,30%	99,30%	99,40%	99,30%	No OMIM disease ID
MASP2	100%	99,60%	100%	100%	MASP2 deficiency, 613791
MBL2	100%	99,80%	100%	100%	No OMIM disease ID
MC2R	99,90%	98,30%	100%	100%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM4	100%	99,50%	100%	100%	Immunodeficiency 54, 609981
MEFV	99,90%	98,60%	96,40%	96,40%	Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MKL1	98,50%	97,20%	100%	100%	?Immunodeficiency 66, 618847
MOGS	100%	99,90%	100%	100%	Congenital disorder of glycosylation, type IIb, 606056
MS4A1	99,80%	98,80%	100%	100%	Immunodeficiency, common variable, 5, 613495
MSN	99,00%	95,70%	100%	100%	Immunodeficiency 50, 300988
MTHFD1	100%	99,50%	100%	100%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MVK	90,90%	90,50%	90,50%	90,50%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYD88	100%	100%	100%	100%	Macroglobulinemia, Waldenstrom, somatic, 153600 Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260
MYSM1	100%	99,10%	100%	100%	Bone marrow failure syndrome 4, 618116
NBAS	100%	99,60%	100%	100%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBN	99,90%	98,60%	100%	100%	Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260 Leukemia, acute lymphoblastic, 613065

NCF1	26,00%	25,80%	100%	99,80%	Chronic granulomatous disease due to deficiency of NCF-1, 233700
NCF2	99,90%	98,30%	100%	100%	Chronic granulomatous disease due to deficiency of NCF-2, 233710
NCF4	100%	100%	100%	100%	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960
NCSTN	100%	99,80%	100%	100%	Acne inversa, familial, 1, 142690
NFAT5	99,80%	99,10%	100%	100%	No OMIM disease ID
NFE2L2	100%	99,90%	100%	100%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFKB1	100%	99,40%	100%	100%	Immunodeficiency, common variable, 12, 616576
NFKB2	98,80%	95,60%	100%	100%	Immunodeficiency, common variable, 10, 615577
NFKBIA	95,20%	88,00%	100%	100%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHEJ1	100%	96,20%	100%	100%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHP2	100%	100%	100%	100%	Dyskeratosis congenita, autosomal recessive 2, 613987
NLRC4	100%	100%	100%	100%	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115
NLRP1	99,60%	98,00%	100%	100%	Palmoplantar carcinoma, multiple self-healing, 615225 Autoinflammation with arthritis and dyskeratosis, 617388 ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803
NLRP12	100%	99,90%	100%	100%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	100%	99,90%	100%	100%	Familial cold inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Keratoendothelitis fugax hereditaria, 148200
NLRP7	100%	99,60%	100%	100%	Hydatidiform mole, recurrent, 1, 231090
NOD2	100%	99,90%	100%	100%	Blau syndrome, 186580
NOP10	100%	99,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NRAS	100%	100%	100%	100%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
NSMCE3	100%	100%	100%	100%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
OAS1	100%	100%	100%	100%	No OMIM disease ID
ORAI1	95,80%	92,80%	97,20%	92,40%	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782

OSTM1	98,60%	94,00%	100%	100%	Osteopetrosis, autosomal recessive 5, 259720
OTULIN	92,60%	86,50%	99,20%	95,00%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
PARN	100%	99,90%	100%	100%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
PAX5	98,70%	96,10%	100%	100%	No OMIM disease ID
PBX1	100%	99,40%	100%	100%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCCA	99,50%	96,70%	100%	100%	Propionicacidemia, 606054
PCCB	97,90%	96,00%	98,70%	96,20%	Propionicacidemia, 606054
PEPD	100%	98,80%	100%	100%	Prolidase deficiency, 170100
PGM3	100%	99,80%	100%	100%	Immunodeficiency 23, 615816
TAP1	100%	99,20%	100%	100%	Bare lymphocyte syndrome, type I, 604571
TAP2	99,90%	99,30%	100%	100%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,50%	95,50%	96,60%	96,60%	Bare lymphocyte syndrome, type I, 604571
TAZ	99,20%	96,50%	100%	100%	Barth syndrome, 302060
PIGA	93,80%	86,70%	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIK3CD	98,80%	96,90%	100%	100%	Immunodeficiency 14, 615513
PIK3R1	99,80%	99,00%	100%	100%	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PLCG2	100%	99,80%	100%	100%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLEKHM1	100%	99,80%	100%	100%	Osteopetrosis, autosomal dominant 3, 618107 ?Osteopetrosis, autosomal recessive 6, 611497
PLG	87,80%	87,50%	100%	100%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PMM2	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNP	99,80%	98,90%	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POLA1	99,30%	95,40%	100%	99,90%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLE2	97,30%	89,80%	100%	100%	No OMIM disease ID
POMP	100%	99,10%	100%	100%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POT1	99,90%	99,00%	100%	100%	No OMIM disease ID
PRF1	91,20%	90,80%	100%	100%	Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027 Hemophagocytic lymphohistiocytosis, familial, 2, 603553

PRKCD	100%	100%	100%	100%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	99,70%	98,00%	100%	100%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRPS1	100%	100%	100%	100%	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Deafness, X-linked 1, 304500 Arts syndrome, 301835 Gout, PRPS-related, 300661
PSENEN	100%	100%	100%	100%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMA3	99,80%	97,20%	100%	100%	No OMIM disease ID
PSMB4	100%	100%	100%	100%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	99,90%	98,50%	100%	100%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	99,90%	97,70%	100%	100%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMG2	100%	98,90%	100%	100%	No OMIM disease ID
PSTPIP1	100%	99,10%	100%	99,90%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTPN22	99,50%	97,10%	100%	100%	No OMIM disease ID
PTPRC	99,00%	95,10%	100%	100%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
RAB27A	100%	100%	100%	100%	Griscelli syndrome, type 2, 607624
RAC2	99,90%	98,30%	100%	100%	Neutrophil immunodeficiency syndrome, 608203
RAG1	100%	100%	100%	100%	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650
RAG2	100%	100%	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RANBP2	49,70%	49,30%	100%	100%	No OMIM disease ID
RASGRP1	100%	99,60%	100%	100%	Immunodeficiency 64, 618534
RASGRP2	99,70%	97,30%	100%	100%	?Bleeding disorder, platelet-type, 18, 615888
RBCK1	99,90%	98,20%	100%	100%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RECQL4	99,80%	98,10%	100%	99,90%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
RELB	98,80%	88,70%	100%	100%	?Immunodeficiency 53, 617585
RFX5	99,70%	98,10%	100%	100%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFXANK	100%	99,50%	100%	100%	MHC class II deficiency, complementation group B, 209920
RFXAP	99,30%	97,00%	100%	99,90%	Bare lymphocyte syndrome, type II, complementation group D, 209920

RHOH	100%	100%	100%	100%	No OMIM disease ID
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RIPK1	100%	99,00%	100%	100%	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852
RNASEH2A	100%	100%	100%	100%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	96,00%	92,50%	100%	99,80%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	99,50%	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNF168	100%	99,80%	100%	100%	RIDDLE syndrome, 611943
RNF31	99,90%	99,00%	100%	100%	No OMIM disease ID
RNU4ATAC	NC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
RORC	100%	100%	100%	100%	Immunodeficiency 42, 616622
RPSA	100%	99,80%	100%	100%	Asplenia, isolated congenital, 271400
RSPH9	99,90%	97,90%	100%	100%	Ciliary dyskinesia, primary, 12, 612650
RTEL1	99,50%	96,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
SAMD9	100%	99,80%	100%	100%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	100%	100%	100%	100%	Ataxia-pancytopenia syndrome, 159550
SAMHD1	100%	99,60%	100%	100%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SBDS	100%	100%	100%	100%	Shwachman-Diamond syndrome, 260400
SEC61A1	100%	100%	100%	100%	Hyperuricemic nephropathy, familial juvenile, 4, 617056
SEMA3E	100%	99,60%	100%	100%	?CHARGE syndrome, 214800
SERAC1	99,90%	99,50%	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPING1	99,70%	97,50%	100%	100%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SH2B3	99,40%	95,10%	100%	99,90%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SH2D1A	97,20%	94,00%	100%	100%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	91,40%	91,20%	97,00%	95,30%	Cherubism, 118400
SH3KBP1	99,70%	95,90%	100%	100%	?Immunodeficiency 61, 300310
SKIV2L	100%	99,80%	100%	100%	Trichohepatoenteric syndrome 2, 614602

SLC29A3	100%	99,60%	100%	100%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC35A1	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type IIf, 603585
SLC35C1	99,90%	98,70%	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC37A4	100%	99,20%	100%	100%	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC39A4	99,50%	95,50%	100%	100%	Acrodermatitis enteropathica, 201100
SLC39A7	100%	100%	100%	100%	No OMIM disease ID
SLC46A1	99,90%	98,50%	100%	100%	Folate malabsorption, hereditary, 229050
SLC7A7	100%	99,90%	100%	100%	Lysinuric protein intolerance, 222700
SMARCAL1	100%	99,90%	100%	100%	Schimke immunoosseous dysplasia, 242900
SMARCD2	87,00%	85,90%	99,60%	97,00%	Specific granule deficiency 2, 617475
SNX10	96,20%	95,70%	100%	99,60%	Osteopetrosis, autosomal recessive 8, 615085
SOCS4	99,90%	99,20%	100%	100%	No OMIM disease ID
SP110	100%	100%	100%	100%	Hepatic venoocclusive disease with immunodeficiency, 235550
SPINK5	99,90%	99,50%	100%	100%	Netherton syndrome, 256500
SPPL2A	85,90%	74,60%	100%	100%	No OMIM disease ID
SRP54	99,50%	96,50%	100%	100%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	97,60%	89,70%	100%	100%	Bone marrow failure syndrome 1, 614675
STAT1	99,90%	98,80%	100%	100%	Immunodeficiency 31C, autosomal dominant, 614162 Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796
STAT2	100%	99,90%	100%	100%	Immunodeficiency 44, 616636
STAT3	100%	99,80%	100%	100%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT4	99,90%	99,60%	100%	100%	No OMIM disease ID
STAT5B	100%	98,50%	100%	100%	Leukemia, acute promyelocytic, somatic, 102578 Growth hormone insensitivity with immunodeficiency, 245590
STAT6	100%	99,80%	100%	100%	No OMIM disease ID
STIM1	99,80%	98,00%	100%	100%	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STK4	100%	99,80%	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STX11	100%	100%	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	82,10%	79,70%	99,30%	97,10%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TBX1	86,90%	79,50%	94,10%	90,80%	Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400

					Tetralogy of Fallot, 187500 Conotruncal anomaly face syndrome, 217095
TCF3	97,10%	94,00%	100%	100%	Agammaglobulinemia 8, autosomal dominant, 616941
TCIRG1	97,60%	90,10%	100%	100%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100%	100%	100%	100%	Transcobalamin II deficiency, 275350
TERC	NC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	96,20%	94,50%	100%	100%	No OMIM disease ID
TFRC	100%	99,80%	100%	100%	Immunodeficiency 46, 616740
TGFB1	100%	99,90%	100%	100%	Camurati-Engelmann disease, 131300 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213
THBD	100%	99,70%	100%	100%	Thrombophilia due to thrombomodulin defect, 614486
TICAM1	100%	99,90%	100%	100%	No OMIM disease ID
TINF2	100%	100%	100%	100%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TIRAP	100%	100%	100%	100%	No OMIM disease ID
TLR3	100%	99,60%	100%	100%	No OMIM disease ID
TLR4	100%	99,80%	100%	100%	No OMIM disease ID
TMC6	100%	99,30%	100%	100%	Epidermodysplasia verruciformis, 226400
TMC8	100%	98,70%	100%	100%	Epidermodysplasia verruciformis 2, 618231
TMEM173	99,70%	95,30%	100%	100%	STING-associated vasculopathy, infantile-onset, 615934
TNFAIP3	100%	100%	100%	100%	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF11A	94,60%	93,30%	99,20%	98,00%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF13B	100%	100%	100%	100%	Immunoglobulin A deficiency 2, 609529 Immunodeficiency, common variable, 2, 240500
TNFRSF13C	80,10%	75,40%	100%	99,90%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	90,60%	87,60%	92,80%	92,80%	Periodic fever, familial, 142680
TNFRSF4	99,40%	95,40%	100%	100%	?Immunodeficiency 16, 615593
TNFRSF9	100%	100%	100%	100%	No OMIM disease ID
TNFSF11	100%	99,90%	100%	100%	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	98,00%	93,60%	100%	100%	No OMIM disease ID
TOP2B	99,40%	96,30%	100%	100%	No OMIM disease ID
TPP2	99,20%	96,80%	100%	100%	No OMIM disease ID
TRAC	100%	100%	100%	100%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	100%	99,90%	100%	100%	No OMIM disease ID
TRAF3IP2	100%	99,30%	100%	100%	?Candidiasis, familial, 8, 615527

TREX1	100%	100%	100%	100%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM22	100%	100%	100%	100%	No OMIM disease ID
TRNT1	99,50%	96,50%	100%	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TTC37	100%	99,30%	100%	100%	Trichohepatoenteric syndrome 1, 222470
TTC7A	99,30%	95,40%	100%	100%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TYK2	99,90%	99,00%	100%	100%	Immunodeficiency 35, 611521
UNC13D	99,70%	98,10%	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC93B1	60,60%	58,80%	100%	100%	No OMIM disease ID
UNG	100%	98,80%	99,90%	99,30%	Immunodeficiency with hyper IgM, type 5, 608106
USB1	100%	99,40%	100%	100%	Poikiloderma with neutropenia, 604173
USP18	95,90%	95,90%	100%	100%	Pseudo-TORCH syndrome 2, 617397
VAV1	98,50%	97,10%	97,10%	97,10%	No OMIM disease ID
VPS13B	99,50%	98,20%	99,50%	99,40%	Cohen syndrome, 216550
VPS45	99,20%	95,60%	95,10%	95,10%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	95,90%	85,30%	100%	99,80%	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
WDR1	100%	99,60%	100%	100%	No OMIM disease ID
WIPF1	100%	99,90%	100%	100%	?Wiskott-Aldrich syndrome 2, 614493
WRAP53	100%	100%	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
XIAP	93,00%	88,80%	100%	100%	Lymphoproliferative syndrome, X-linked, 2, 300635
ZAP70	100%	99,30%	100%	100%	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB24	100%	100%	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZNF341	97,20%	95,00%	100%	100%	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282

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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. “No OMIM Disease ID” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors