Panel Inmunológico

Es nuestra solución para la inmunodeficiencia y los trastornos de inmunodeficiencia combinada grave (SCID). Nuestro panel incluye genes dirigidos a la inmunodeficiencia combinada grave, la neutropenia congénita, la deficiencia primaria de anticuerpos, la inmunodeficiencia común variable, la enfermedad granulomatosa crónica, la linfoproliferación autoinmune y la agammaglobulinemia.

Nº de genes:	326
Entrega:	25 días
Cobertura:	≥99.5% ≥20x Cobertura media con profundidad ≥ 150 x
Detalles:	Análisis CNV incluido

Síndromes y trastornos comunes cubiertos

Agammaglobulinemia

Síndrome linfoproliferativo autoinmune

Inmunodeficiencia combinada severa de células B negativas

Inmunodeficiencia combinada severa con células B positivas

Enfermedad granulomatosa crónica

Inmunodeficiencia común variable

Afibrinogenemia congénita

Sínd Síndrome de Hermasky-Pudlak

Síndromes de neutropenia congénita

Susceptibilidad mendeliana a enfermedades micobacterianas

Síndrome de fiebre periódica

Deficiencia primaria de anticuerpos

Discinesia ciliar primaria

Inmunodeficiencias primarias (IDP)

Inmunodeficiencia combinada severa

Genes	OMIM (Genes)	Enfermedades asociadas (OMIM)	Herencia
ACD	609377	Dyskeratosis congenita, AD 6;Dyskeratosis congenita, AR 7	AD, AR
ACP5	171640	Spondyloenchondrodysplasia with immune dysregulation	AR
ACTB	102630	Baraitser-Winter syndrome 1;?Dystonia, juvenile-onset	AD
ADA	608958	Adenosine deaminase deficiency, partial;Severe combined immunodeficiency due to ADA deficiency	AR, SM
ADA2	607575	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome;?Sneddon syndrome	AR
ADAR	146920	Aicardi-Goutieres syndrome 6;Dyschromatosis symmetrica hereditaria	AR, AD
AICDA	605257	Immunodeficiency with hyper-IgM, type 2	AR
AIRE	607358	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	AD, AR
AK2	103020	Reticular dysgenesis	AR

AP1S3	615781	Psoriasis 15, pustular, susceptibility to	AD
AP3B1	603401	Hermansky-Pudlak syndrome 2	AR
ARPC1B	604223	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia	AR
ATM	607585	Breast cancer, susceptibility to;Ataxia-telangiectasia	AD, SM, AR
ATP6AP1	300197	Immunodeficiency 47	XLR
B2M	109700	Immunodeficiency 43;?Amyloidosis, familial visceral	AR, AD
BACH2	605394	Immunodeficiency 60	AD
BCL11B	606558	Immunodeficiency 49;Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities	AD
BLM	604610	Bloom syndrome	AR
BLNK	604515	Agammaglobulinemia 4	AR
BLOC1S3	609762	Hermansky-Pudlak syndrome 8	AR
BLOC1S6	604310	Hermansky-pudlak syndrome 9	AR
BTK	300300	Agammaglobulinemia, XL 1;Isolated growth hormone deficiency, type III, with agammaglobulinemia	XLR
C1QA	120550	C1q deficiency	AR
C1QB	120570	C1q deficiency	AR
C1QC	120575	C1q deficiency	AR
C1R	613785	Ehlers-Danlos syndrome, periodontal type, 1	AD

C1S	120580	C1s deficiency;Ehlers-Danlos syndrome, periodontal type, 2	AD
C2	613927	Macular degeneration, age-related, 14, reduced risk of;C2 deficiency	DD, AR
C3	120700	Macular degeneration, age-related, 9;C3 deficiency;Hemolytic uremic syndrome, atypical, susceptibility to, 5	AR, AD
C5	120900	C5 deficiency;[Eculizumab, poor response to]	AR, AD
C6	217050	C6 deficiency	
C7	217070	C7 deficiency	
C8A	120950	C8 deficiency, type I	AR
C8B	120960	C8 deficiency, type II	AR
C9	120940	Macular degeneration, age-related, 15, susceptibility to;C9 deficiency	
CARD11	607210	Immunodeficiency 11A;Immunodeficiency 11B with atopic dermatitis;B-cell expansion with NFKB and T-cell anergy	AR, AD
CARD14	607211	Psoriasis 2;Pityriasis rubra pilaris	AD
CARD9	607212	Candidiasis, familial, 2, AR	AR
CARMIL2	610859	Immunodeficiency 58	AR
CASP10	601762	Autoimmune lymphoproliferative syndrome, type II;Gastric cancer, somatic;Lymphoma, non-Hodgkin, somatic	AD

CASP8	601763	Breast cancer, protection against;Lung cancer, protection against;?Autoimmune lymphoproliferative syndrome, type IIB;Hepatocellular carcinoma, somatic	AD, SM, AR
CCBE1	612753	Hennekam lymphangiectasia-lymphedema syndrome 1	AR
CCDC103	614677	Ciliary dyskinesia, primary, 17	AR
CCDC39	613798	Ciliary dyskinesia, primary, 14	AR
CCDC40	613799	Ciliary dyskinesia, primary, 15	AR
CCDC65	611088	Ciliary dyskinesia, primary, 27	AR
CCNO	607752	Ciliary dyskinesia, primary, 29	AR
CD19	107265	Immunodeficiency, common variable, 3	AR
CD247	186780	?Immunodeficiency 25	AR
CD27	186711	Lymphoproliferative syndrome 2	AR
CD3D	186790	Immunodeficiency 19	AR
CD3E	186830	Immunodeficiency 18, SCID variant;Immunodeficiency 18	AR
CD3G	186740	Immunodeficiency 17, CD3 gamma deficient	AR
CD40	109535	Immunodeficiency with hyper-IgM, type 3	AR
CD40LG	300386	Immunodeficiency, XL, with hyper-IgM	XLR
CD46	120920	Hemolytic uremic syndrome, atypical, susceptibility to, 2	AD, AR

CD55	125240	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy;[Blood group Cromer]	AR
CD59	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	AR
CD70	602840	Lymphoproliferative syndrome 3	AR
CD79A	112205	Agammaglobulinemia 3	AR
CD79B	147245	Agammaglobulinemia 6	AR
CD81	186845	Immunodeficiency, common variable, 6	AR
CD8A	186910	CD8 deficiency, familial	AR
CDCA7	609937	Immunodeficiency-centromeric instability-facial anomalies syndrome 3	AR
CEBPE	600749	Specific granule deficiency	AR
CFAP298	615494	Ciliary dyskinesia, primary, 26	AR
CFB	138470	?Complement factor B deficiency;Hemolytic uremic syndrome, atypical, susceptibility to, 4;Macular degeneration, age-related, 14, reduced risk of	AR, AD, DD
CFD	134350	Complement factor D deficiency	AR
CFH	134370	Basal laminar drusen;Macular degeneration, age-related, 4;Hemolytic uremic syndrome, atypical, susceptibility to, 1;Complement factor H deficiency	AD, AD, AR

CFHR1	134371	Macular degeneration, age-related, reduced risk of;Hemolytic uremic syndrome, atypical, susceptibility to	AD, AD, AR
CFI	217030	Complement factor I deficiency;Macular degeneration, age-related, 13, susceptibility to;Hemolytic uremic syndrome, atypical, susceptibility to, 3	AR, AD
CFP	300383	Properdin deficiency, XL	XLR
CFTR	602421	Congenital bilateral absence of vas deferens;Pancreatitis, hereditary;Bronchiectasis with or without elevated sweat chloride 1, modifier of;Cystic fibrosis	AR, AD
CHD7	608892	CHARGE syndrome;Hypogonadotropic hypogonadism 5 with or without anosmia	AD
CIITA	600005	Rheumatoid arthritis, susceptibility to;Bare lymphocyte syndrome, type II, complementation group A	AR
CLCN7	602727	Osteopetrosis, AR 4;Osteopetrosis, AD 2;Hypopigmentation, organomegaly, and delayed myelination and development	AR, AD
CLPB	616254	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia	AR
COPA	601924	Autoimmune interstitial lung, joint, and kidney disease	AD

CORO1A	605000	Immunodeficiency 8	AR
CR2	120650	Immunodeficiency, common variable, 7;Systemic lupus erythematosus, susceptibility to, 9	AR
CSF2RB	138981	Surfactant metabolism dysfunction, pulmonary, 5	AR
CSF3R	138971	Neutropenia, severe congenital, 7, AR	AR
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts	AR
CTLA4	123890	Systemic lupus erythematosus, susceptibility to;Autoimmune lymphoproliferative syndrome, type V;Celiac disease, susceptibility to, 3;Hashimoto thyroiditis;Diabetes mellitus, insulin-dependent, 12	AD
CTPS1	123860	Immunodeficiency 24	AR
CTSC	602365	Periodontitis 1, juvenile;Haim-Munk syndrome;Papillon-Lefevre syndrome	AR
CXCR4	162643	WHIM syndrome	AD
CYBA	608508	Chronic granulomatous disease 4, AR	AR
CYBB	300481	Immunodeficiency 34, mycobacteriosis, XL;Chronic granulomatous disease, XL	XLR
DCLRE1C	605988	Omenn syndrome;Severe combined immunodeficiency, Athabascan type	AR
DDX58	609631	Singleton-Merten syndrome 2	AD

DGKE	601440	Nephrotic syndrome, type 7;Hemolytic uremic syndrome, atypical, susceptibility to, 7	AR
DKC1	300126	Dyskeratosis congenita, XL	XLR
DNAAF1	613190	Ciliary dyskinesia, primary, 13	AR
DNAAF2	612517	Ciliary dyskinesia, primary, 10	AR
DNAAF3	614566	Ciliary dyskinesia, primary, 2	AR
DNAAF4	608706	Dyslexia, susceptibility to, 1;Ciliary dyskinesia, primary, 25	AD, AR
DNAAF5	614864	Ciliary dyskinesia, primary, 18	AR
DNAH11	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus	AR
DNAH5	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus	AR
DNAI1	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus	AR
DNAI2	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus	AR
DNAJC21	617048	Bone marrow failure syndrome 3	AR
DNAL1	610062	Ciliary dyskinesia, primary, 16	AR
DNASE1L3	602244	Systemic lupus erythematosus 16	AR
DNMT3B	602900	Facioscapulohumeral muscular dystrophy 4, digenic;Immunodeficiency-centromeric instability-facial anomalies syndrome 1	DD, AR
DOCK2	603122	Immunodeficiency 40	AR
DOCK8	611432	Hyper-IgE recurrent infection syndrome, AR	AR

DRC1	615288	Ciliary dyskinesia, primary, 21	AR
DTNBP1	607145	Hermansky-Pudlak syndrome 7	AR
ELANE	130130	Neutropenia, cyclic;Neutropenia, severe congenital 1, AD	AD
EPG5	615068	Vici syndrome	AR
ERCC6L2	615667	Bone marrow failure syndrome 2	AR
EXTL3	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities	AR
F12	610619	Factor XII deficiency;Angioedema, hereditary, type III	AR, AD
FADD	602457	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations	AR
FAS	134637	Autoimmune lymphoproliferative syndrome, type IA;Autoimmune lymphoproliferative syndrome	AD
FASLG	134638	Autoimmune lymphoproliferative syndrome, type IB;Lung cancer, susceptibility to	AD, AD, SM
FAT4	612411	Hennekam lymphangiectasia-lymphedema syndrome 2;Van Maldergem syndrome 2	AR
FCGR3A	146740	Immunodeficiency 20	AR
FCN3	604973	Immunodeficiency due to ficolin 3 deficiency	AR
FERMT3	607901	Leukocyte adhesion deficiency, type III	AR

FGA	134820	Hypodysfibrinogenemia, congenital;Afibrinogenemia, congenital;Amyloidosis, familial visceral;Dysfibrinogenemia, congenital	AR, AD
FGB	134830	Afibrinogenemia, congenital;Dysfibrinogenemia, congenital;Hypofibrinogenemia, congenital	AR
FGG	134850	Dysfibrinogenemia, congenital;Afibrinogenemia, congenital;Hypofibrinogenemia, congenital;Hypodysfibrinogenemia	AR
FOXN1	600838	T-cell lymphopenia, infantile, with or without nail dystrophy, AD;T-cell immunodeficiency, congenital alopecia, and nail dystrophy	AD, AR
FOXP3	300292	Immunodysregulation, polyendocrinopathy, and enteropathy, XL	XLR
G6PC3	611045	Dursun syndrome;Neutropenia, severe congenital 4, AR	AR
G6PD	305900	Resistance to malaria due to G6PD deficiency;Hemolytic anemia, G6PD deficient (favism)	XLD

GATA1	305371	Thrombocytopenia with beta-thalassemia, XL;Thrombocytopenia, XL, with or without dyserythropoietic anemia;Anemia, XL, with/without neutropenia and/or platelet abnormalities;Leukemia, megakaryoblastic, with or without Down syndrome, somatic	XLR
GATA2	137295	Myelodysplastic syndrome, susceptibility to;Leukemia, acute myeloid, susceptibility to;Emberger syndrome;Immunodeficiency 21	AD, SM, AD
GFI1	600871	Neutropenia, severe congenital 2, AD;?Neutropenia, nonimmune chronic idiopathic, of adults	AD
GINS1	610608	Immunodeficiency 55	AR
GUCY2C	601330	Meconium ileus;Diarrhea 6	AR, AD
HAX1	605998	Neutropenia, severe congenital 3, AR	AR
HELLS	603946	Immunodeficiency-centromeric instability-facial anomalies syndrome 4	AR
HPS1	604982	Hermansky-Pudlak syndrome 1	AR
HPS3	606118	Hermansky-Pudlak syndrome 3	AR
HPS4	606682	Hermansky-Pudlak syndrome 4	AR

HPS5	607521	Hermansky-Pudlak syndrome 5	AR
HPS6	607522	Hermansky-Pudlak syndrome 6	AR
HTRA2	606441	Parkinson disease 13;3-methylglutaconic aciduria, type VIII	AR
HYDIN	610812	Ciliary dyskinesia, primary, 5	AR
ICOS	604558	Immunodeficiency, common variable, 1	AR
IFIH1	606951	Aicardi-Goutieres syndrome 7;Singleton-Merten syndrome 1	AD
IFNGR1	107470	Immunodeficiency 27A, mycobacteriosis, AR;Hepatitis B virus infection, susceptibility to;Tuberculosis, susceptibility to;Tuberculosis infection, protection against;H. pylori infection, susceptibility to;Immunodeficiency 27B, mycobacteriosis, AD	AR, AD
IFNGR2	147569	Immunodeficiency 28, mycobacteriosis	AR
IGLL1	146770	Agammaglobulinemia 2	AR
IKBKB	603258	Immunodeficiency 15B;Immunodeficiency 15A	AR, AD
IKZF1	603023	Immunodeficiency, common variable, 13	AD
IL10	124092	Graft-versus-host disease, protection against;Rheumatoid arthritis, progression of;HIV-1, susceptibility to	
IL10RA	146933	Inflammatory bowel disease 28, early onset, AR	AR

IL10RB	123889	Inflammatory bowel disease 25, early onset, AR;Hepatitis B virus, susceptibility to	AR
IL12B	161561	Immunodeficiency 29, mycobacteriosis	AR
IL12RB1	601604	Immunodeficiency 30	AR
IL12RB2	601642		
IL17RA	605461	Immunodeficiency 51	AR
IL17RC	610925	Candidiasis, familial, 9	AR
IL1RN	147679	Microvascular complications of diabetes 4;Gastric cancer risk after H. pylori infection;Interleukin 1 receptor antagonist deficiency	AR, AD
IL21R	605383	[IgE, elevated level of];Immunodeficiency 56	AD, AR
IL2RA	147730	Diabetes, mellitus, insulin-dependent, susceptibility to, 10;Immunodeficiency 41 with lymphoproliferation and autoimmunity	AR
IL2RG	308380	Severe combined immunodeficiency, XL;Combined immunodeficiency, XL, moderate	XLR
IL36RN	605507	Psoriasis 14, pustular	AR
IL7R	146661	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	AR
INO80	610169		
IRAK4	606883	Immunodeficiency 67	AR

IRF3	603734	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7	AD
IRF8	601565	Immunodeficiency 32B, monocyte and dendritic cell deficiency, AR;Immunodeficiency 32A, mycobacteriosis, AD	AR, AD
ISG15	147571	Immunodeficiency 38	AR
ITCH	606409	Autoimmune disease, multisystem, with facial dysmorphism	AR
ITGB2	600065	Leukocyte adhesion deficiency	AR
ITK	186973	Lymphoproliferative syndrome 1	AR
JAGN1	616012	Neutropenia, severe congenital, 6, AR	AR
JAK3	600173	SCID, AR, T-negative/B-positive type	AR

KRAS	190070	Arteriovenous malformation of the brain, somatic;Gastric cancer, somatic;Oculoectodermal syndrome, somatic;RAS-associated autoimmune leukoproliferative disorder;Pancreatic carcinoma, somatic;Lung cancer, somatic;Cardiofaciocutaneous syndrome 2;Bladder cancer, somatic;Leukemia, acute myeloid, somatic;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Breast cancer, somatic;Noonan syndrome 3	AD
LAMTOR2	610389	Immunodeficiency due to defect in MAPBP-interacting protein	AR
LAT	602354	Immunodeficiency 52	AR
LIG1	126391		
LIG4	601837	Multiple myeloma, resistance to;LIG4 syndrome	SM, AR
LPIN2	605519	Majeed syndrome	
LRBA	606453	Immunodeficiency, common variable, 8, with autoimmunity	AR
LYST	606897	Chediak-Higashi syndrome	AR
MAGT1	300715	Congenital disorder of glycosylation, type Icc;Immunodeficiency, XL, with magnesium defect, Epstein-Barr virus infection and neoplasia	XLR
MALT1	604860	Immunodeficiency 12	AR

MASP2	605102	MASP2 deficiency	AR
MBL2	154545	Chronic infections, due to MBL deficiency	AD
MCM4	602638	Immunodeficiency 54	AR
MEFV	608107	Neutrophilic dermatosis, acute febrile;Familial Mediterranean fever, AR;Familial Mediterranean fever, AD	AD, AR
MOGS	601336	Congenital disorder of glycosylation, type IIb	AR
MS4A1	112210	?Immunodeficiency, common variable, 5	AR
MSN	309845	Immunodeficiency 50	XLR
MTHFD1	172460	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia;Neural tube defects, folate-sensitive, susceptibility to	AR
MVK	251170	Mevalonic aciduria;Hyper-IgD syndrome;Porokeratosis 3, multiple types	AR, AD
MYD88	602170	Macroglobulinemia, Waldenstrom, somatic;Immunodeficiency 68	AR
MYSM1	612176	Bone marrow failure syndrome 4	AR
NBN	602667	Aplastic anemia;Leukemia, acute lymphoblastic;Nijmegen breakage syndrome	AR
NCF1	608512	Chronic granulomatous disease 1, AR	AR
NCF2	608515	Chronic granulomatous disease 2, AR	AR
NCF4	601488	Chronic granulomatous disease 3, AR	AR
NCSTN	605254	Acne inversa, familial, 1	AD

NFKB1	164011	Immunodeficiency, common variable, 12	AD
NFKB2	164012	Immunodeficiency, common variable, 10	AD
NFKBIA	164008	Ectodermal dysplasia and immunodeficiency 2	AD
NHEJ1	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	
NHP2	606470	Dyskeratosis congenita, AR 2	AR
NLRC4	606831	Autoinflammation with infantile enterocolitis;?Familial cold autoinflammatory syndrome 4	AD
NLRP1	606636	Autoinflammation with arthritis and dyskeratosis;?Respiratory papillomatosis, juvenile recurrent, congenital;Palmoplantar carcinoma, multiple self-healing;Vitiligo-associated multiple autoimmune disease susceptibility 1	AD, AR, AR, AD
NLRP12	609648	Familial cold autoinflammatory syndrome 2	AD
NLRP3	606416	CINCA syndrome;Deafness, AD 34, with or without inflammation;Keratoendothelitis fugax hereditaria;Familial cold inflammatory syndrome 1;Muckle-Wells syndrome	AD
NME8	607421	Ciliary dyskinesia, primary, 6	AR
NOD2	605956	Yao syndrome;Blau syndrome;Inflammatory bowel disease 1, Crohn disease	mi, AD

NOP10	606471	Dyskeratosis congenita, AR 1	AR
NRAS	164790	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Colorectal cancer, somatic;Neurocutaneous melanosis, somatic;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic;Noonan syndrome 6;Melanocytic nevus syndrome, congenital, somatic;Thyroid carcinoma, follicular, somatic;Epidermal nevus, somatic	AD
NSMCE3	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome	AR
OFD1	300170	Joubert syndrome 10;Simpson-Golabi-Behmel syndrome, type 2;?Retinitis pigmentosa 23;Orofaciodigital syndrome I	XLR, XLD
OSTM1	607649	Osteopetrosis, AR 5	AR
OTULIN	615712	Autoinflammation, panniculitis, and dermatosis syndrome	AR
PARN	604212	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4;Dyskeratosis congenita, AR 6	AD, AR
PEPD	613230	Prolidase deficiency	AR
PGM3	172100	Immunodeficiency 23	AR
PIK3CD	602839	Immunodeficiency 14;?Roifman-Chitayat syndrome, digenic;Immunodeficiency 14B, AR	AD, DR, AR

PIK3R1	171833	?Agammaglobulinemia 7, AR;Immunodeficiency 36;SHORT syndrome	AR, AD
PLCG2	600220	Familial cold autoinflammatory syndrome 3;Autoinflammation, antibody deficiency, and immune dysregulation syndrome	AD
PMM2	601785	Congenital disorder of glycosylation, type Ia	AR
PMS2	600259	Colorectal cancer, hereditary nonpolyposis, type 4;Mismatch repair cancer syndrome 4	AR
PNP	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency	AR
POLA1	312040	Van Esch-O'Driscoll syndrome;Pigmentary disorder, reticulate, with systemic manifestations, XL	XLR
POLE	174762	Colorectal cancer, susceptibility to, 12;IMAGE-I syndrome;FILS syndrome	AD, AR
PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, 2;Lymphoma, non-Hodgkin;Aplastic anemia	AR
PRKCD	176977	Autoimmune lymphoproliferative syndrome, type III	AR
PSENEN	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease	AD
PSMB4	602177	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms	AR

PSMB8	177046	Proteasome-associated autoinflammatory syndrome 1 and digenic forms	AR
PSTPIP1	606347	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne	AD
PTEN	601728	Cowden syndrome 1;Lhermitte-Duclos syndrome;Macrocephaly/autism syndrome;Glioma susceptibility 2;Meningioma;Prostate cancer, somatic	AD
PTPRC	151460	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive;Hepatitis C virus, susceptibility to	AR
RAB27A	603868	Griscelli syndrome, type 2	AR
RAC2	602049	?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia;Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis;Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia	AR, AD

RAG1	179615	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity;Omenn syndrome;Severe combined immunodeficiency, B cell-negative;Combined cellular and humoral immune defects with granulomas	AR
RAG2	179616	Omenn syndrome;Combined cellular and humoral immune defects with granulomas;Severe combined immunodeficiency, B cell-negative	AR
RANBP2	601181	Encephalopathy, acute, infection-induced, 3, susceptibility to	AD
RASGRP1	603962	Immunodeficiency 64	AR
RBCK1	610924	Polyglucosan body myopathy 1 with or without immunodeficiency	AR
RFX5	601863	Bare lymphocyte syndrome, type II, complementation group E;Bare lymphocyte syndrome, type II, complementation group C	AR
RFXANK	603200	MHC class II deficiency, complementation group B	AR
RFXAP	601861	Bare lymphocyte syndrome, type II, complementation group D	AR

RIPK1	603453	Autoinflammation with episodic fever and lymphadenopathy;Immunodeficiency 57 with autoinflammation	AD, AR
RNASEH2A	606034	Aicardi-Goutieres syndrome 4	AR
RNASEH2B	610326	Aicardi-Goutieres syndrome 2	AR
RNASEH2C	610330	Aicardi-Goutieres syndrome 3	AR
RNF168	612688	RIDDLE syndrome	AR
RNF31	612487		
RORC	602943	Immunodeficiency 42	AR
RSPH1	609314	Ciliary dyskinesia, primary, 24	AR
RSPH4A	612647	Ciliary dyskinesia, primary, 11	
RSPH9	612648	Ciliary dyskinesia, primary, 12	
RTEL1	608833	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3;Dyskeratosis congenita, AD 4;Dyskeratosis congenita, AR 5	AD, AD, AR
SAMD9	610456	Monosomy 7 myelodysplasia and leukemia syndrome 2;MIRAGE syndrome;Tumoral calcinosis, familial, normophosphatemic	AD, AR
SAMD9L	611170	Ataxia-pancytopenia syndrome;Monosomy 7 myelodysplasia and leukemia syndrome 1	AD
SAMHD1	606754	?Chilblain lupus 2;Aicardi-Goutieres syndrome 5	AD, AR

SBDS	607444	Aplastic anemia, susceptibility to;Shwachman-Diamond syndrome	AR
SEMA3E	608166	?CHARGE syndrome	AD
SERPING1	606860	Complement component 4, partial deficiency of;Angioedema, hereditary, types I and II	AD, AD, AR
SH2D1A	300490	Lymphoproliferative syndrome, XL, 1	XLR
SKIV2L	600478	Trichohepatoenteric syndrome 2	AR
SLC29A3	612373	Histiocytosis-lymphadenopathy plus syndrome	AR
SLC35C1	605881	Congenital disorder of glycosylation, type IIc	AR
SLC7A7	603593	Lysinuric protein intolerance	AR
SMARCAL1	606622	Schimke immunoosseous dysplasia	AR
SMARCD2	601736	Specific granule deficiency 2	AR
SP110	604457	Mycobacterium tuberculosis, susceptibility to;Hepatic venoocclusive disease with immunodeficiency	AR
SPAG1	603395	Ciliary dyskinesia, primary, 28	AR
SPINK5	605010	Netherton syndrome	AR
SRP54	604857	Neutropenia, severe congenital, 8, AD	AD
SRP72	602122	Bone marrow failure syndrome 1	AD

STAT1	600555	Immunodeficiency 31C, chronic mucocutaneous candidiasis, AD;Immunodeficiency 31B, mycobacterial and viral infections, AR;Immunodeficiency 31A, mycobacteriosis, AD	AD, AR
STAT2	600556	Pseudo-TORCH syndrome 3;Immunodeficiency 44	AR
STAT3	102582	Hyper-IgE recurrent infection syndrome;Autoimmune disease, multisystem, infantile-onset, 1	AD
STAT5B	604260	Growth hormone insensitivity with immune dysregulation 2, AD;Growth hormone insensitivity with immune dysregulation 1, AR;Leukemia, acute promyelocytic, somatic	AD, AR
STIM1	605921	Myopathy, tubular aggregate, 1;Stormorken syndrome;Immunodeficiency 10	AD, AR
STING1	612374	STING-associated vasculopathy, infantile-onset	AD
STK4	604965	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	
STX11	605014	Hemophagocytic lymphohistiocytosis, familial, 4	AR
STXBP2	601717	Hemophagocytic lymphohistiocytosis, familial, 5	
TAFAZZIN	300394	Barth syndrome	XLR

TAP1	170260	Bare lymphocyte syndrome, type I	AR
TAP2	170261	Bare lymphocyte syndrome, type I, due to TAP2 deficiency	AR
TAPBP	601962	Bare lymphocyte syndrome, type I	AR
TBK1	604834	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4;Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8	AD
TBX1	602054	Velocardiofacial syndrome;Tetralogy of Fallot;DiGeorge syndrome;Conotruncal anomaly face syndrome	AD
TCF3	147141	Agammaglobulinemia 8, AD	AD
TCIRG1	604592	Osteopetrosis, AR 1	AR
TCN2	613441	Transcobalamin II deficiency	AR
TERT	187270	Melanoma, cutaneous malignant, 9;Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1;Dyskeratosis congenita, AD 2;Leukemia, acute myeloid;Dyskeratosis congenita, AR 4	AD, AD, AR, AD, SM
TFRC	190010	Immunodeficiency 46	AR
THBD	188040	Thrombophilia due to thrombomodulin defect;Hemolytic uremic syndrome, atypical, susceptibility to, 6	AD

TICAM1	607601	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6	AD, AR
TINF2	604319	Revesz syndrome;Dyskeratosis congenita, AD 3	AD
TLR3	603029	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2;HIV1 infection, resistance to	AD, AR
TMC6	605828	Epidermodysplasia verruciformis	AR
TMC8	605829	Epidermodysplasia verruciformis 2	AR
TNFAIP3	191163	Autoinflammatory syndrome, familial, Behcet-like	AD
TNFRSF11A	603499	Paget disease of bone 2, early-onset;Osteopetrosis, AR 7;Osteolysis, familial expansile	AD, AR
TNFRSF13B	604907	Immunodeficiency, common variable, 2;Immunoglobulin A deficiency 2	AD, AR
TNFRSF13C	606269	Immunodeficiency, common variable, 4	AR
TNFRSF1A	191190	Periodic fever, familial;Multiple sclerosis, susceptibility to, 5	AD
TNFSF11	602642	Osteopetrosis, AR 2	AR
TPP2	190470	Immunodeficiency 78 with autoimmunity and developmental delay	AR

TREX1	606609	Aicardi-Goutieres syndrome 1, dominant and recessive;Chilblain lupus;Vasculopathy, retinal, with cerebral leukodystrophy;Systemic lupus erythematosus, susceptibility to	AD, AR, AD
TRNT1	612907	Retinitis pigmentosa and erythrocytic microcytosis;Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay	AR
TTC37	614589	Trichohepatoenteric syndrome 1	AR
TTC7A	609332	Gastrointestinal defects and immunodeficiency syndrome	AR
TYK2	176941	Immunodeficiency 35	AR
UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, 3	AR
UNG	191525	Immunodeficiency with hyper IgM, type 5	AR
USB1	613276	Poikiloderma with neutropenia	AR
USP18	607057	Pseudo-TORCH syndrome 2	AR
VPS13B	607817	Cohen syndrome	AR
VPS45	610035	Neutropenia, severe congenital, 5, AR	AR
WAS	300392	Wiskott-Aldrich syndrome;Thrombocytopenia, XL;Neutropenia, severe congenital, XL;Thrombocytopenia, XL, intermittent	XLR

WDR1	604734	Periodic fever, immunodeficiency, and thrombocytopenia syndrome	AR
WRAP53	612661	Dyskeratosis congenita, AR 3	AR
XIAP	300079	Lymphoproliferative syndrome, XL, 2	XLR
ZAP70	176947	Autoimmune disease, multisystem, infantile-onset, 2;Immunodeficiency 48	AR
ZBTB24	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2	AR
ZMYND10	607070	Ciliary dyskinesia, primary, 22	AR