Panel de anemia/insuficiencia de la médula ósea

Nuestro **panel de anemia/insuficiencia de la médula ósea** está destinado a pacientes con anomalías en más de 2 tipos de células sanguíneas (glóbulos rojos, glóbulos blancos y plaquetas) que presentan síntomas de letargo, infecciones recurrentes, sangrado excesivo, pigmentación anormal, agrandamiento del bazo, y malignidades. Algunos trastornos específicos detectados con este panel son linfohistiocitosis hemofagocítica, síndrome de Seckel, trombocitopenia, anemia de Fanconi, disqueratosis congénita, síndrome de Shwachman Diamond así como otros tipos de anemias, como talasemia alfa y beta, enfermedad de células falciformes, esferocitosis, anemia megaloblástica, anemia congénita anemia sideroblástica y diseritropoyética.

Nº de genes:	211
Entrega:	25 días
Cobertura:	≥99.5% ≥20x Cobertura media con profundidad ≥ 150 x
Detalles:	Análisis CNV incluido

SÍNDROMES Y TRASTORNOS COMUNES CUBIERTOS

Síndrome de insuficiencia de la médula ósea

Anemia diseritropoyética congénita

Anemia sideroblástica congénita

Anemia de Diamond-Blackfan

Anemia de Fanconi

Linfohistiocitosis hemofagocítica

Esferocitosis hereditaria

Trombocitopenia

ByGENS

Resumen de genes y enfermedades asociadas

Genes	OMIM (gen)	Enfermedades asociadas (OMIM)	Herencia
ABCB6	605452	Microphthalmia, isolated, with coloboma 7;[Blood group, Langereis system];Dyschromatosis universalis hereditaria 3;Pseudohyperkalemia, familial, 2, due to red cell leak	AD
ABCB7	300135	Anemia, sideroblastic, with ataxia	XLR
ABCG5	605459	Sitosterolemia 2	AR
ABCG8	605460	Gallbladder disease 4;Sitosterolemia 1	AR
ACD	609377	?Dyskeratosis congenita, AD 6;?Dyskeratosis congenita, AR 7	AD, AR
ACTN1	102575	Bleeding disorder, platelet-type, 15	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
ADAMTS13	604134	Thrombotic thrombocytopenic purpura, hereditary	AR
AK1	103000	Hemolytic anemia due to adenylate kinase deficiency	AR
AK2	103020	Reticular dysgenesis	AR
ALAS2	301300	Protoporphyria, erythropoietic, XL;Anemia, sideroblastic, 1	XL, XLR
ALDOA	103850	Glycogen storage disease XII	AR
AMMECR1	300195	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis	XLR
AMN	605799	Imerslund-Grasbeck syndrome 2	AR
ANK1	612641	Spherocytosis, type 1	AD, AR
ANKRD26	610855	Thrombocytopenia 2	AD
AP3B1	603401	Hermansky-Pudlak syndrome 2	AR
ATM	607585	Breast cancer, susceptibility to;Ataxia-telangiectasia	AD, SM, AR
ATRX	300032	Mental retardation-hypotonic facies syndrome, XL;Alpha-thalassemia/mental retardation syndrome;Alpha-thalassemia myelodysplasia syndrome, somatic	XLR, XLD
BLM	604610	Bloom syndrome	AR
BLOC1S3	609762	Hermansky-Pudlak syndrome 8	AR
BRCA1	113705	Breast-ovarian cancer, familial, 1;Pancreatic cancer, susceptibility to, 4;Fanconi anemia, complementation group S	AD, mi, AR
BRCA2	600185	Prostate cancer;Breast-ovarian cancer, familial, 2;Glioblastoma 3;Wilms tumor;Pancreatic cancer 2;Breast cancer, male, susceptibility to;Fanconi anemia, complementation group D1;Medulloblastoma	AD, SM, AD, AR, AD, AR, SM

BRIP1	605882	Fanconi anemia, complementation group J;Breast cancer, early-onset, susceptibility to	AD, SM
CASP10	601762	Autoimmune lymphoproliferative syndrome, type II;Gastric cancer, somatic;Lymphoma, non-Hodgkin, somatic	AD
CBL	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia;?Juvenile myelomonocytic leukemia	AD, AD, SM
CBLIF	609342	Intrinsic factor deficiency	AR
CD36	173510	Malaria, cerebral, reduced risk of;Coronary heart disease, susceptibility to, 7;Platelet glycoprotein IV deficiency;Malaria, cerebral, susceptibility to	AR
CD40LG	300386	Immunodeficiency, XL, with hyper-IgM	XLR
CD59	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	AR
CDAN1	607465	Dyserythropoietic anemia, congenital, type Ia	AR
CDC42	116952	Takenouchi-Kosaki syndrome	AD
CENPJ	609279	?Seckel syndrome 4;Microcephaly 6, primary, AR	AR
CEP152	613529	Microcephaly 9, primary, AR;Seckel syndrome 5	AR
CHEK2	604373	Prostate cancer, familial, susceptibility to;Colorectal cancer, susceptibility to;Breast cancer, susceptibility to;Osteosarcoma, somatic;Li-Fraumeni syndrome	AD, SM
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
COL4A1	120130	?Retinal arteries, tortuosity of;Hemorrhage, intracerebral, susceptibility to;Microangiopathy and leukoencephalopathy, pontine, AD;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps;Brain small vessel disease with or without ocular anomalies	AD
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
CUBN	602997	[Proteinuria, chronic benign];Imerslund-Grasbeck syndrome 1	AR
CXCR4	162643	WHIM syndrome	AD
CYB5R3	613213	Methemoglobinemia, type I;Methemoglobinemia, type II	AR

CYCS	123970	Thrombocytopenia 4	AD
DHFR	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency	AR
DIAPH1	602121	Seizures, cortical blindness, microcephaly syndrome;Deafness, AD 1, with or without thrombocytopenia	AR, AD
DKC1	300126	Dyskeratosis congenita, XL	XLR
DNAJC21	617048	Bone marrow failure syndrome 3	AR
DTNBP1	607145	Hermansky-Pudlak syndrome 7	AR
EFL1	617538	Shwachman-Diamond syndrome 2	AR
ELANE	130130	Neutropenia, cyclic;Neutropenia, severe congenital 1, AD	AD
EPB41	130500	Elliptocytosis-1	AD, AR
EPB42	177070	Spherocytosis, type 5	
ERCC4	133520	XFE progeroid syndrome;Xeroderma pigmentosum, type F/Cockayne syndrome;Xeroderma pigmentosum, group F;Fanconi anemia, complementation group Q	AR
ETV6	600618	Leukemia, acute myeloid, somatic;Thrombocytopenia 5	AD
FANCA	607139	Fanconi anemia, complementation group A	AR
FANCB	300515	Fanconi anemia, complementation group B	XLR
FANCC	613899	Fanconi anemia, complementation group C	AR
FANCD2	613984	Fanconi anemia, complementation group D2	AR
FANCE	613976	Fanconi anemia, complementation group E	AR
FANCF	613897	Fanconi anemia, complementation group F	AR
FANCG	602956	Fanconi anemia, complementation group G	AR
FANCI	611360	Fanconi anemia, complementation group I	AR
FANCL	608111	Fanconi anemia, complementation group L	AR
FANCM	609644	Spermatogenic failure 28;?Premature ovarian failure 15	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
FCGR2C	612169		
FLI1	193067	Bleeding disorder, platelet-type, 21	AD, AR
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

GCLC	606857	Myocardial infarction, susceptibility to;Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency	AR
GFI1	600871	Neutropenia, severe congenital 2, AD;?Neutropenia, nonimmune chronic idiopathic, of adults	AD
GFI1B	604383	Bleeding disorder, platelet-type, 17	AD, AR
GLRX5	609588	Spasticity, childhood-onset, with hyperglycinemia;Anemia, sideroblastic, 3, pyridoxine-refractory	AR
GNE	603824	Nonaka myopathy;Sialuria	AR, AD
GP1BA	606672	Bernard-Soulier syndrome, type A1 (recessive);von Willebrand disease, platelet-type;Bernard-Soulier syndrome, type A2 (dominant);Nonarteritic anterior ischemic optic neuropathy, susceptibility to	AR, AD
GP1BB	138720	Giant platelet disorder, isolated;Bernard-Soulier syndrome, type B	AR
GP9	173515	Bernard-Soulier syndrome, type C	AR
GPI	172400	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency	AR
GPX1	138320	Hemolytic anemia due to glutathione peroxidase deficiency	AR
GSR	138300	Hemolytic anemia due to glutathione reductase deficiency	AR
GSS	601002	Glutathione synthetase deficiency;Hemolytic anemia due to glutathione synthetase deficiency	AR
GYPC	110750	Malaria, resistance to;[Blood group, Gerbich]	
HAX1	605998	Neutropenia, severe congenital 3, AR	AR
HBA1	141800	Methemoglobinemia, alpha type;Heinz body anemias, alpha-;Erythrocytosis 7;Thalassemias, alpha-;Hemoglobin H disease, nondeletional	AD
HBA2	141850	Thalassemia, alpha-;Erythrocytosis 7;Heinz body anemia;Hemoglobin H disease, deletional and nondeletional	AD
HBB	141900	Erythrocytosis 6;Methemoglobinemia, beta type;Delta-beta thalassemia;Thalassemia-beta, dominant inclusion-body;Sickle cell anemia;Hereditary persistence of fetal hemoglobin;Malaria, resistance to;Thalassemia, beta;Heinz body anemia	AD, AR
HBD	142000		
HFE	613609	Porphyria variegata, susceptibility to;Alzheimer disease, susceptibility to;Hemochromatosis;[Transferrin serum level QTL2];Porphyria cutanea tarda, susceptibility to;Microvascular complications of diabetes 7	AD, AR, AD, AR

HK1	142600	Retinitis pigmentosa 79;Hemolytic anemia due to hexokinase deficiency;Neurodevelopmental disorder with visual defects and brain anomalies;Neuropathy, hereditary motor and sensory, Russe type	AD, AR
HMOX1	141250	Heme oxygenase-1 deficiency;Pulmonary disease, chronic obstructive, susceptibility to	AR
HOXA11	142958	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1	AD
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
HSPA9	600548	Even-plus syndrome;Anemia, sideroblastic, 4	AR, AD
IKZF1	603023	Immunodeficiency, common variable, 13	AD
IL2RG	308380	Severe combined immunodeficiency, XL;Combined immunodeficiency, XL, moderate	XLR
ITGA2B	607759	Glanzmann thrombasthenia;Bleeding disorder, platelet-type, 16, AD	AR, AD
ITGB3	173470	Glanzmann thrombasthenia 2;Glanzmann thrombasthenia;Bleeding disorder, platelet-type, 16, AD;Myocardial infarction, susceptibility to;Bleeding disorder, platelet-type, 24, AD	AR, AD
ITK	186973	Lymphoproliferative syndrome 1	AR
JAGN1	616012	Neutropenia, severe congenital, 6, AR	AR
KCNN4	602754	Dehydrated hereditary stomatocytosis 2	AD
KDM1A	609132	Cleft palate, psychomotor retardation, and distinctive facial features	AD
KDSR	136440	Erythrokeratodermia variabilis et progressiva 4	AR
KIT	164920	Gastrointestinal stromal tumor, familial;Germ cell tumors, somatic;Piebaldism;Leukemia, acute myeloid, somatic;Mastocytosis, systemic, somatic;Mastocytosis, cutaneous	AD, IC, AD
KLF1	600599	Dyserythropoietic anemia, congenital, type IV;Blood group--Lutheran inhibitor;[Hereditary persistence of fetal hemoglobin]	AD
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

LIG4	601837	Multiple myeloma, resistance to;LIG4 syndrome	SM, AR
LPIN2	605519	Majeed syndrome	
LYST	606897	Chediak-Higashi syndrome	AR
MECOM	165215	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2	AD
MLH1	120436	Mismatch repair cancer syndrome 1;Muir-Torre syndrome;Colorectal cancer, hereditary nonpolyposis, type 2	AR, AD
MPIG6B	606520	?Thrombocytopenia, anemia, and myelofibrosis	AR
MPL	159530	Myelofibrosis with myeloid metaplasia, somatic;Thrombocytopenia, congenital amegakaryocytic;Thrombocythemia 2	AR, AD, SM
MRE11	600814	Ataxia-telangiectasia-like disorder 1	AR
MSH2	609309	Muir-Torre syndrome;Colorectal cancer, hereditary nonpolyposis, type 1;Mismatch repair cancer syndrome 2	AD, AR
MSH6	600678	Colorectal cancer, hereditary nonpolyposis, type 5;Endometrial cancer, familial;Mismatch repair cancer syndrome 3	AD, AD, SM, AR
MTR	156570	Neural tube defects, folate-sensitive, susceptibility to;Homocystinuria-megaloblastic anemia, cblG complementation type	AR
MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type;Neural tube defects, folate-sensitive, susceptibility to	AR
MYH9	160775	Deafness, AD 17;Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss	AD
NBEAL2	614169	Gray platelet syndrome	AR
NBN	602667	Aplastic anemia;Leukemia, acute lymphoblastic;Nijmegen breakage syndrome	AR
NF1	613113	Watson syndrome;Leukemia, juvenile myelomonocytic;Neurofibromatosis, type 1;Neurofibromatosis, familial spinal;Neurofibromatosis-Noonan syndrome	AD, AD, SM
NFKB1	164011	Immunodeficiency, common variable, 12	AD
NHP2	606470	Dyskeratosis congenita, AR 2	AR
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
NT5C3A	606224	Anemia, hemolytic, due to UMPH1 deficiency	AR

PALB2	610355	Pancreatic cancer, susceptibility to, 3;Fanconi anemia, complementation group N;Breast cancer, susceptibility to	AD, SM
PARN	604212	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4;Dyskeratosis congenita, AR 6	AD, AR
PC	608786	Pyruvate carboxylase deficiency	AR
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency	XLD
PDHX	608769	Lacticacidemia due to PDX1 deficiency	AR
PFKM	610681	Glycogen storage disease VII	AR
PGK1	311800	Phosphoglycerate kinase 1 deficiency	XLR
PIEZO1	611184	Lymphatic malformation 6;Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema	AR, AD
PKLR	609712	Adenosine triphosphate, elevated, of erythrocytes;Pyruvate kinase deficiency	AD, AR
PLAU	191840	Quebec platelet disorder;Alzheimer disease, late-onset, susceptibility to	AD
PMS2	600259	Colorectal cancer, hereditary nonpolyposis, type 4;Mismatch repair cancer syndrome 4	AR
PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, 2;Lymphoma, non-Hodgkin;Aplastic anemia	AR
PTPN11	176876	Leukemia, juvenile myelomonocytic, somatic;LEOPARD syndrome 1;Metachondromatosis;Noonan syndrome 1	AD
PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1	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
RAD51	179617	Breast cancer, susceptibility to;Mirror movements 2;Fanconi anemia, complementation group R	AD, SM, AD
RAD51C	602774	Fanconi anemia, complementation group O;Breast-ovarian cancer, familial, susceptibility to, 3	AR
RBBP8	604124	Seckel syndrome 2;Jawad syndrome	AR
RBM8A	605313	Thrombocytopenia-absent radius syndrome	AR
REN	179820	Hyperuricemic nephropathy, familial juvenile 2;Renal tubular dysgenesis	AD, AR
RHAG	180297	Overhydrated hereditary stomatocytosis;Anemia, hemolytic, Rh-, regulator type	AD
RIT1	609591	Noonan syndrome 8	AD
RPL11	604175	Diamond-Blackfan anemia 7	AD
RPL15	604174	?Diamond-Blackfan anemia 12	AD

RPL35A	180468	Diamond-Blackfan anemia 5	AD
RPL5	603634	Diamond-Blackfan anemia 6	AD
RPS10	603632	Diamond-Blackfan anemia 9	AD
RPS19	603474	Diamond-Blackfan anemia 1	AD
RPS24	602412	Diamond-blackfan anemia 3	AD
RPS26	603701	Diamond-Blackfan anemia 10	AD
RPS28	603685	Diamond Blackfan anemia 15 with mandibulofacial dysostosis	AD
RPS29	603633	Diamond-Blackfan anemia 13	AD
RPS7	603658	Diamond-Blackfan anemia 8	AD
RTEL1	608833	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3;Dyskeratosis congenita, AD 4;Dyskeratosis congenita, AR 5	AD, AD, AR
RUNX1	151385	Leukemia, acute myeloid;Platelet disorder, familial, with associated myeloid malignancy	AD, SM, AD
SAMD9	610456	Monosomy 7 myelodysplasia and leukemia syndrome 2;MIRAGE syndrome;Tumoral calcinosis, familial, normophosphatemic	AD, AR
SBDS	607444	Aplastic anemia, susceptibility to;Shwachman-Diamond syndrome	AR
SEC23B	610512	Dyserythropoietic anemia, congenital, type II;?Cowden syndrome 7	AR, AD
SH2D1A	300490	Lymphoproliferative syndrome, XL, 1	XLR
SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome	AR
SLC19A3	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	AR
SLC25A19	606521	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type);Microcephaly, Amish type	AR
SLC25A38	610819	Anemia, sideroblastic, 2, pyridoxine-refractory	AR
SLC2A1	138140	GLUT1 deficiency syndrome 2, childhood onset;GLUT1 deficiency syndrome 1, infantile onset, severe;Epilepsy, idiopathic generalized, susceptibility to, 12;Stomatin-deficient cryohydrocytosis with neurologic defects;Dystonia 9	AD, AD, AR
SLC35C1	605881	Congenital disorder of glycosylation, type IIc	AR
SLC4A1	109270	[Blood group, Wright];Distal renal tubular acidosis 1;Ovalocytosis, SA type;[Malaria, resistance to];[Blood group, Swann];[Blood group, Froese];[Blood group, Waldner];Cryohydrocytosis;[Blood group, Diego];Distal renal tubular acidosis 4 with hemolytic anemia;Spherocytosis, type 4	AD, AR
SLFN14	614958	Bleeding disorder, platelet-type, 20	AD
SLX4	613278	Fanconi anemia, complementation group P	AR

SPTA1	182860	Pyropoikilocytosis;Elliptocytosis-2;Spherocytosis, type 3	AR, AD
SPTB	182870	Spherocytosis, type 2;Elliptocytosis-3;Anemia, neonatal hemolytic, fatal or near-fatal	AD
SRP72	602122	Bone marrow failure syndrome 1	AD
STAT3	102582	Hyper-IgE recurrent infection syndrome;Autoimmune disease, multisystem, infantile-onset, 1	AD
STIM1	605921	Myopathy, tubular aggregate, 1;Stormorken syndrome;Immunodeficiency 10	AD, AR
STX11	605014	Hemophagocytic lymphohistiocytosis, familial, 4	AR
STXBP2	601717	Hemophagocytic lymphohistiocytosis, familial, 5	
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
THPO	600044	Thrombocythemia 1	AD
TINF2	604319	Revesz syndrome;Dyskeratosis congenita, AD 3	AD
TMPRSS6	609862	Iron-refractory iron deficiency anemia	AR
TP53	191170	Hepatocellular carcinoma, somatic;Bone marrow failure syndrome 5;Basal cell carcinoma 7;Nasopharyngeal carcinoma, somatic;Choroid plexus papilloma;Glioma susceptibility 1;Pancreatic cancer, somatic;Breast cancer, somatic;Li-Fraumeni syndrome;Adrenocortical carcinoma, pediatric;Osteosarcoma;Colorectal cancer	AD, AD, SM, SM
TPI1	190450	Hemolytic anemia due to triosephosphate isomerase deficiency	AR
TPK1	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	AR
TRNT1	612907	Retinitis pigmentosa and erythrocytic microcytosis;Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay	AR
UBE2T	610538	Fanconi anemia, complementation group T	AR
UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, 3	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
WNT4	603490	Mullerian aplasia and hyperandrogenism;?SERKAL syndrome	AD, AR
WRAP53	612661	Dyskeratosis congenita, AR 3	AR
XIAP	300079	Lymphoproliferative syndrome, XL, 2	XLR

XK	314850	McLeod syndrome with or without chronic granulomatous disease	XL
XRCC2	600375	Spermatogenic failure;?Fanconi anemia, complementation group U;?Premature ovarian failure 17	AR
YARS2	610957	Myopathy, lactic acidosis, and sideroblastic anemia 2	AR