eurofins | Genoma

Genescreen Easy-Donor: gene list v.1

N.B. A question mark (?) indicates that the relationship between the condition and gene is provisional.

Gene name	Condition
AAAS	Triple-A syndrome (achalasia-addisonianism-alacrimia)
ABAT	GABA-transaminase deficiency
ABCA12	Ichthyosis congenital autosomal recessive type 4A and 4B (harlequin)
ABCA3	Surfactant metabolism dysfunction, pulmonary, type 3
ABCC8	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
ABCD1	Adrenoleukodystrophy
ABHD12	PHARC syndrome (polyneuropathy, hearing loss, ataxia, retinitis pigmentosa and cataract)
ACAD9	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency
ACADS	Short-chain acyl-CoA dehydrogenase deficiency
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
ACAT1	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)
ACE	Renal tubular dysgenesis
ACOX1	Peroxisomal acyl-CoA oxidase deficiency
ACTA1	Nemaline myopathy 3; Congenital fiber-type disproportion myopathy 1
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)
ADAMTS10	Weill-Marchesani syndrome, type 1 recessive
ADAMTS13	Thrombotic thrombocytopenic purpura, familial (Schulman-Upshaw syndrome)
AFF2	Intellectual developmental disorder, X-linked 109
AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)
AGL	Glycogen storage disease, type 3
AGPAT2	Congenital generalized lipodystrophy (Berardinelli-Seip syndrome)
AGPS	Rhizomelic chondrodysplasia punctata, type 3
AGRN	Myasthenic syndrome, congenital, type 8
AGXT	Hyperoxaluria, primary, type 1
AHI1	Joubert syndrome, type 3
AIMP1	Leukodystrophy, hypomyelinating, type 3
AIRE	Autoimmune polyendocrinopathy syndrome, type 1
ALDH18A1	Spastic paraplegia, type 9B, autosomal recessive; Cutis laxa, type 3A (De Barsy syndrome)
ALDH3A2	Sjogren-Larsson syndrome
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency
ALDH7A1	Epilepsy, pyridoxine-dependent
ALDOB	Fructose intolerance, hereditary
ALG12	Congenital disorder of glycosylation, type 1G
ALG6	Congenital disorder of glycosylation, type 1C
ALMS1	Alström syndrome
ALOX12B	Ichthyosis, congenital, autosomal recessive, type 2

ALPL	Hypophosphatasia, infantile/childhood
AMACR	Bile acid synthesis defect, congenital, type 4; Alpha-methylacyl-CoA racemase deficiency
AMT	Glycine encephalopathy
ANO10	Spinocerebellar ataxia, autosomal recessive, type 10
ANTXR2	Hyaline fibromatosis syndrome
AP1S2	Pettigrew syndrome
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia
AQP2	Diabetes insipidus, nephrogenic, type 2
ARFGEF2	Periventricular heterotopia with microcephaly
ARL13B	Joubert syndrome type 8
ARL6	Bardet-Biedl syndrome, type 3
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)
ARSE	Chondrodysplasia punctata, X-linked recessive
ARX	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders
ASL	Argininosuccinic aciduria
ASPA	Canavan disease
ASPM	Primary microcephaly type 5 autosomal recessive
ASS1	Citrullinemia, type 1
ATM	Ataxia-telangiectasia
ATP6V0A2	Cutis laxa, autosomal recessive, type 2A; Wrinkly skin syndrome
ATP7A	Menkes disease; Occipital horn syndrome
ATP7B	Wilson disease
ATR	Seckel syndrome, type 1
ATRX	Intellectual disability-hypotonic facies syndrome, X-linked; Alpha-thalassemia/intellectual developmental disorder syndrome
AUH	3-methylglutaconic aciduria, type 1
BBS1	Bardet-Biedl syndrome, type 1
BBS10	Bardet-Biedl syndrome, type 10
BBS12	Bardet-Biedl syndrome, type 12
BBS2	Bardet-Biedl syndrome, type 2
BBS7	Bardet-Biedl syndrome, type 7
BBS9	Bardet-Biedl syndrome, type 9
BCKDHA	Maple syrup urine disease, type 1A
BCKDHB	Maple syrup urine disease, type 1B
BCS1L	Mitochondrial complex III deficiency nuclear type 1; GRACILE syndrome; Bjornstad syndrome
BEST1	Bestrophinopathy, AR
BLM	Bloom syndrome
BRWD3	Intellectual developmental disorder, X-linked 93
BSCL2	Congenital generalized lipodystrophy, type 2; Encephalopathy, progressive, with or without lipodystrophy
BTD	Biotinidase deficiency
BUB1B	Mosaic variegated aneuploidy syndrome 1
CBS	Homocystinuria due to cystathionine beta-synthase
CC2D2A	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2
CCDC88C	Hydrocephalus, congenital, type 1

CD40LG	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)
CDAN1	Dyserythropoietic anemia, congenital, type 1A
CDH23	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D
CDK5RAP2	Primary microcephaly type 3 autosomal recessive
CENPJ	Primary microcephaly type 6 autosomal recessive
CEP152	Primary microcephaly type 9 autosomal recessive
CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10
CFL2	Nemaline myopathy, type 7 autosomal recessive
CFTR	Cystic fibrosis
CHAT	Myasthenic syndrome, congenital, type 6 presynaptic
CHM	Choroideremia
CHRND	Myasthenic syndrome, congenital, type 3B, fast-channel; Multiple pterygium syndrome, lethal type
CHRNE	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency
CHRNG	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type
CHST14	Ehlers-Danlos syndrome, musculocontractural, type 1
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations
CLCN1	Myotonia congenita, recessive
CLN3	Ceroid lipofuscinosis, neuronal, type 3
CLN5	Ceroid lipofuscinosis, neuronal, type 5
CLN6	Ceroid lipofuscinosis, neuronal, type 6
CLN8	Ceroid lipofuscinosis, neuronal, type 8
CLRN1	Usher syndrome, type 3A
CNGB3	Achromatopsia, type 3
COG4	Congenital disorder of glycosylation, type 2J
COL11A2	Otospondylomegaepiphyseal dysplasia, autosomal recessive
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type
COL4A3	Alport syndrome, autosomal recessive, type 2
COL4A5	Alport syndrome, X-linked
COL7A1	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial
COLQ	Myasthenic syndrome, congenital, type 5
COQ2	Primary coenzyme Q10 deficiency, type 1
CORO1A	Immunodeficiency, type 8
CPS1	Carbamoylphosphate synthetase 1 deficiency
CPT1A	Carnitine palmitoyltransferase type 1A deficiency, hepatic
CPT2	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile
CRLF1	Cold-induced sweating syndrome type 1
CRTAP	Osteogenesis imperfecta, type 7
CSTB	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)
CTC1	Cerebroretinal microangiopathy with calcifications and cysts
CTNS	Nephropathic cystinosis
CTSD	Ceroid lipofuscinosis, neuronal, type 10
CTSK	Pycnodysostosis
CUL4B	Intellectual developmental disorder, X-linked syndromic, Cabezas type
CUL7	3M syndrome 1

CYBB	Chronic granulomatous disease, X-linked
CYP11A1	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency
CYP1B1	Glaucoma, primary congenital, type 3A
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
CYP27A1	Cerebrotendinous xanthomatosis
CYP27B1	Vitamin D-dependent rickets, type 1
D2HGDH	D-2-hydroxyglutaric aciduria
DBH	Dopamine beta-hydroxylase deficiency
DBT	Maple syrup urine disease, type 2
DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type
DCX	Lissencephaly, X-linked, type 1
DDC	Aromatic L-amino acid decarboxylase deficiency
DGUOK	DGUOK-related mitochondrial DNA depletion syndrome
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Retinitis pigmentosa, type 59
DHH	46,XY complete gonadal dysgenesis
DLAT	Pyruvate dehydrogenase E2 deficiency
DLG3	Intellectual developmental disorder, X-linked 90
DMD	Duchenne/Becker muscular dystrophy
DMD	Duchenne/Becker muscular dystrophy
DMP1	Hypophosphatemic rickets, autosomal recessive
DNAH11	Ciliary dyskinesia, primary, type 7 with or without situs inversus
DNAH5	Ciliary dyskinesia, primary, type 3 with or without situs inversus
DNAL1	Ciliary dyskinesia, primary, type 16
DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive
DOK7	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10
DPAGT1	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13
DPYD	Dihydropyrimidine dehydrogenase deficiency
DYNC2H1	Short-rib thoracic dysplasia, type 3 with or without polydactyly
EDA	Ectodermal dysplasia, type 1 hypohidrotic, X-linked
EDNRB	ABCD syndrome
EIF2AK3	Wolcott-Rallison syndrome
EIF2B2	Leukoencephalopathy with vanishing white matter (VWM)
EIF2B3	Leukoencephalopathy with vanishing white matter (VWM)
EIF2B4	Leukoencephalopathy with vanishing white matter (VWM)
EIF2B5	Leukoencephalopathy with vanishing white matter (VWM)
EMD	Emery-Dreifuss muscular dystrophy, type 1 X-linked
ENPP1	Arterial calcification, generalized, of infancy, type 1
EPG5	Vici syndrome
EPM2A	Epilepsy, progressive myoclonic, type 2A (Lafora)
ERCC2	Trichothiodystrophy, type 1
ERCC4	Fanconi anemia, complementation group Q
ERCC5	Cerebrooculofacioskeletal syndrome, type 3
ERCC6	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1
ERCC8	Cockayne syndrome, type A
ESCO2	Roberts syndrome; Juberg-Hayward syndrome
ETFDH	Glutaric acidemia, type 2C

ETHE1	Ethylmalonic encephalopathy
EVC	Ellis-van Creveld syndrome
EVC2	Ellis-van Creveld syndrome
EXOSC3	Pontocerebellar hypoplasia, type 1B
F11	Factor XI deficiency
F8	Hemophilia A
F9	Hemophilia B
FA2H	Spastic paraplegia, type 35 autosomal recessive
FAH	Tyrosinemia, type 1
FAM20C	Raine syndrome
FANCA	Fanconi anemia, complementation group A
FANCC	Fanconi anemia, complementation group C
FANCF	Fanconi anemia, complementation group F
FANCG	Fanconi anemia, complementation group G
FANCI	Fanconi anemia, complementation group I
FGD1	Aarskog-Scott syndrome; intellectual developmental disorder, X-linked syndromic, type 16
FKRP	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])
FKTN	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])
FMO3	Trimethylaminuria
FMR1	Fragile X syndrome
FRAS1	Fraser syndrome, type 1
FTSJ1	Intellectual developmental disorder, X-linked 9
FUCA1	Fucosidosis
G6PC	Glycogen storage disease, type 1A
G6PD	Hemolytic anemia, G6PD deficient (favism)
GAA	Glycogen storage disease, type 2
GALC	Krabbe disease
GALT	Galactosemia
GAMT	Cerebral creatine deficiency syndrome, type 2
GAN	Giant axonal neuropathy, type 1
GATM	Cerebral creatine deficiency syndrome, type 3
GBA	Gaucher Disease
GBE1	Glycogen storage disease, type 4
GCDH	Glutaricaciduria, type 1
GFM1	Combined oxidative phosphorylation deficiency, type 1
GFPT1	Myasthenia, congenital, type 12 with tubular aggregates
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1
GJB2	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6
GJB6	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6
GLA	Fabry disease
GLB1	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)
GLDC	Glycine encephalopathy
GLRA1	Hyperekplexia, type 1
GNPTAB	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta
GNRHR	Hypogonadotropic hypogonadism, type 7 without anosmia

GOSR2	Epilepsy, progressive myoclonic, type 6
GP1BA	Bernard-Soulier syndrome, type A1
GP1BB	Bernard-Soulier syndrome, type B
GPHN	Molybdenum cofactor deficiency C
GPR143	Ocular albinism, type 1 (Nettleship-Falls type)
GRHPR	Hyperoxaluria, primary, type 2
GRIP1	Fraser syndrome 3
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency
HADHA	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency
HBA1	Alpha-thalassemia
HBA2	Alpha-thalassemia
HBB	Beta-thalassemia, Sickle cell anemia and other HBB-related hemoglobinopathies
HEXA	Tay-Sachs disease
HEXB	Sandhoff disease, infantile, juvenile, and adult forms
HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency
HLCS	Holocarboxylase synthetase deficiency
HPS3	Hermansky-Pudlak syndrome, type 3
HPS4	Hermansky-Pudlak syndrome, type 4
HPS6	Hermansky-Pudlak syndrome, type 6
HSD17B3	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
HSD17B4	D-bifunctional protein deficiency
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type
IDS	Mucopolysaccharidosis, type 2
IDUA	Mucopolysaccharidosis type 1
IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S
IL1RAPL1	Intellectual developmental disorder, X-linked 21
IL2RG	Severe combined immunodeficiency, X-linked
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type
INPP5E	Joubert syndrome, type 1
INSR	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, type A
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia
IVD	Isovaleric acidemia
JAK3	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type
KCNJ1	Bartter syndrome, type 2
KCNJ13	Leber congenital amaurosis, type 16
KCTD7	Epilepsy, progressive myoclonic, type 3 with or without intracellular inclusions
KDM5C	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type
KIF7	Acrocallosal syndrome; Joubert syndrome, type 12
L1CAM	L1 Syndrome
LAMA2	LAMA2-related muscular dystrophy
LAMA3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMB2	Pierson syndrome; Nephrotic syndrome, type 5 with or without ocular abnormalities

LAMB3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMC2	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LIFR	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome
LMNA	Cardiomyopathy, dilated, 1A
LPL	Lipoprotein lipase deficiency
LRP2	Donnai-Barrow syndrome
LRPPRC	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)
LTBP4	Cutis laxa, autosomal recessive, type 1C
MAN2B1	Alpha-mannosidosis
MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency, type 2
MCOLN1	Mucolipidosis type 4
MCPH1	Microcephaly type 1 primary, autosomal recessive
MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2B
MFSD8	Ceroid lipofuscinosis, neuronal, type 7
MID1	Opitz GBBB syndrome, type 1
MKS1	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts
MMAB	Methylmalonic aciduria, vitamin B12-responsive, type cblB
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
MMADHC	Homocystinuria, cblD type, variant 1
MMUT	Methylmalonic aciduria, mut(0) type
MPDU1	Congenital disorder of glycosylation, type 1F
MPI	Congenital disorder of glycosylation, type 1B
MPV17	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE
MPZ	Dejerine-Sottas disease
MTHFR	Homocystinuria due to MTHFR deficiency
MTM1	Myotubular myopathy, X-linked
MTO1	Combined oxidative phosphorylation deficiency 10
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type
MUSK	Fetal akinesia deformation sequence, type 1; Myasthenic syndrome, congenital, type 9 associated with acetylcholine receptor deficiency
MVK	Mevalonic aciduria
MYO15A	Deafness, autosomal recessive, type 3
MYO5A	Griscelli syndrome, type 1
MYO5B	Microvillus inclusion disease
MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2
NAGA	Schindler disease, type I
NBN	Nijmegen breakage syndrome
NDP	Norrie disease
NEB	Nemaline myopathy type 2
NEUROG3	Diarrhea 4 malabsorptive, congenital
NHLRC1	Epilepsy, progressive myoclonic, type 2B (Lafora)
NPC1	Niemann-Pick disease, type C1
NPC2	Niemann-pick disease, type C2
NPHP1	Joubert syndrome type 4
NPHP3	Meckel syndrome type 7

NPHP4	Nephronophthisis type 4
NPHS1	Nephrotic syndrome, type 1
NPHS2	Nephrotic syndrome, type 2
NR0B1	Adrenal hypoplasia, congenital
NTRK1	Insensitivity to pain, congenital, with anhidrosis
NUP62	Striatonigral degeneration, infantile
OBSL1	3M syndrome 2
OCA2	Oculocutaneous albinism type 2
OCRL	Lowe Syndrome; Dent disease type 2
OPHN1	Intellectual developmental disorder, X-linked syndromic, Billuart type
OTC	Ornithine transcarbamylase deficiency
PAH	Phenylketonuria
PAK3	Intellectual developmental disorder, X-linked 30
PC	Pyruvate carboxylase deficiency
PCCA	Propionic acidemia
PCDH15	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency
PDHB	Pyruvate dehydrogenase E1-beta deficiency
PDHX	Lacticacidemia due to PDX1 deficiency
PEX1	Heimler syndrome type 1
PEX10	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B
PEX16	Peroxisome biogenesis disorder, type 8A (Zellweger syndrome); Peroxisome biogenesis disorder, type 8B
PEX7	Rhizomelic chondrodysplasia punctata, type 1
PGK1	Phosphoglycerate kinase 1 deficiency
PHF8	Intellectual developmental disorder, X-linked syndromic, Siderius type
PKHD1	Polycystic kidney disease type 4
PKP1	Ectodermal dysplasia/skin fragility syndrome
PLA2G6	Infantile neuroaxonal dystrophy type 1
PLEC	Epidermolysis bullosa simplex with muscular dystrophy
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1
PLP1	Pelizaeus-Merzbacher disease
PMM2	Congenital disorder of glycosylation, type 1A
PNPLA1	Ichthyosis, congenital, autosomal recessive, type 10
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency
POLG	POLG-related disorders
POLR1C	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3
POMGNT1	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])
POMT1	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11]) muscular dystrophy, type 11 [LGMD R11])
POMT2	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14]) muscular dystrophy, type 14 [LGMD R14])
POU1F1	Pituitary hormone deficiency, combined, type 1
POU3F4	Deafness, X-linked, type 2

PPT1	Ceroid lipofuscinosis, neuronal, type 1
PQBP1	Renpenning syndrome
PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2
PRICKLE1	Epilepsy, progressive myoclonic, type 1B
PROP1	Pituitary hormone deficiency, combined, type 2
PRPS1	Phosphoribosylpyrophosphate synthetase (PRS) deficiency
PSAP	Combined SAP deficiency
PYGL	Glycogen storage disease, type 6
RAB3GAP1	Warburg micro syndrome; Martsolf syndrome
RAB3GAP2	Warburg micro syndrome; Martsolf syndrome
RAD51C	Fanconi anemia, complementation group O
RAG1	Omenn syndrome; Severe combined immunodeficiency, B cell-negative
RAG2	Omenn syndrome; Severe combined immunodeficiency, B cell-negative
RAPSN	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11 associated with AChR deficiency
RARS2	Pontocerebellar hypoplasia, type 6
RELN	Lissencephaly 2 (Norman-Roberts type)
RMRP	Anauxetic dysplasia 1
RNASEH2B	Aicardi-Goutieres syndrome, type 2
ROR2	Robinow syndrome, autosomal recessive
RP2	Retinitis pigmentosa, type 2 X-linked
RPGR	Retinitis pigmentosa, type 3 X-linked; Cone-rod dystrophy, X-linked, 1
RPGRIP1	Leber congenital amaurosis, type 6
RPGRIP1L	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome
RS1	Retinoschisis
RTEL1	Dyskeratosis congenita, autosomal recessive type 5
RYR1	RYR1 related congenitalmyopathy
SACS	Spastic ataxia, Charlevoix-Saguenay, type
SBDS	Shwachman-Diamond syndrome
SCN4A	Myasthenic syndrome, congenital, type 16
SCN9A	Indifference to pain and autosomal recessive hereditary sensory neuropathy type 2D
SCNN1B	Pseudohypoaldosteronism, type 1
SCO2	Mitochondrial complex IV deficiency, nuclear type 2
SGCA	Limb-girdle muscular dystrophy, type 3 (LGMD R3)
SGCB	Limb-girdle muscular dystrophy, type 4 (LGMD R4)
SH3TC2	Charcot-Marie-Tooth disease, type 4C
SIL1	Marinesco-Sjogren syndrome
SLC12A1	Bartter syndrome, type 1
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy
SLC16A2	Allan-Herndon-Dudley syndrome
SLC17A5	Salla disease
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome
SLC19A3	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)
SLC25A19	Microcephaly, Amish type; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)
SLC25A22	Developmental and epileptic encephalopathy 3
SLC26A2	Achondrogenesis, type 1B (diastrophic dysplasia)

SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome
SLC2A2	Fanconi-Bickel syndrome
SLC35A1	Congenital disorder of glycosylation, type 2F
SLC35C1	Congenital disorder of glycosylation, type 2C
SLC37A4	Glycogen storage disease, type 1B
SLC3A1	Cystinuria
SLC45A2	Albinism, oculocutaneous, type 4
SLC4A11	Corneal endothelial dystrophy, autosomal recessive
SLC6A8	Cerebral creatine deficiency syndrome, type 1
SMN1	Spinal muscular atrophy
SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome
SP110	Hepatic venoocclusive disease with immunodeficiency
SPATA7	Leber congenital amaurosis, type 3
SRD5A2	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)
SRD5A3	Congenital disorder of glycosylation, type 1Q; Kahrizi syndrome
ST3GAL5	Salt and pepper developmental regression syndrome
STAR	Lipoid adrenal hyperplasia
STIL	Microcephaly, type 7 primary, autosomal recessive
STRA6	Microphthalmia, isolated, with coloboma, type 8
SUOX	Sulfite oxidase deficiency
SURF1	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
TCTN1	Joubert syndrome, type 13
TCTN2	Joubert syndrome, type 24; ?Meckel syndrome, type 8
TF	Atransferrinemia
TGM1	Ichthyosis, congenital, autosomal recessive, type 1
TH	Segawa syndrome, recessive
THOC2	intellectual developmental disorder, X-linked 12
TJP2	Cholestasis, progressive familial intrahepatic, type 4
TMEM138	Joubert syndrome 16
TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2
TMEM237	Joubert syndrome, type 14
TMEM67	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome
TNNT1	Nemaline myopathy , type 5 Amish type
TNXB	Ehlers-Danlos syndrome, classic-like
TPK1	Episodic encephalopathy due to thiamine pyrophosphokinase deficiency
TPP1	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7
TREX1	Aicardi-Goutieres syndrome, type 1
TRIM32	Limb-girdle muscular dystrophy, type 8 (LGMD R8)
TSEN2	Pontocerebellar hypoplasia, type 2B
TSEN34	Pontocerebellar hypoplasia type 2C
TSEN54	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4
TSFM	Combined oxidative phosphorylation deficiency, type 3
TTC21B	Short-rib thoracic dysplasia, type 4 with or without polydactyly

TTC8	Bardet-Biedl syndrome, type 8
TTN	Limb-girdle muscular dystrophy type 10 (LGMDR10); Early-onset myopathy with fatal cardiomyopathy (Salih myopathy)
TTPA	Ataxia with isolated vitamin E deficiency
TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B
UBR1	Johanson-Blizzard syndrome
UNC13D	Hemophagocytic lymphohistiocytosis, familial, type 3
UPF3B	Intellectual developmental disorder, X-linked syndromic 14
USH1C	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A
USH1G	Usher syndrome, type 1G
USH2A	Usher syndrome, type 2A
VLDLR	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1
VPS13B	Cohen syndrome
VPS33B	Arthrogryposis, renal dysfunction and cholestasis, type 1
WAS	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked
WDR62	Microcephaly, type 2 primary, autosomal recessive, with or without cortical malformations
WFS1	Wolfram syndrome, type 1
WNT7A	Fuhrmann syndrome
WRN	Werner syndrome
XPA	Xeroderma pigmentosum, group A
XPC	Xeroderma pigmentosum, group C
ZAP70	Autoimmune disease, multisystem, infantile-onset, type 2; Immunodeficiency, type 48
ZDHHC9	Intellectual developmental disorder, X-linked syndromic, Raymond type
ZMPSTE24	Mandibuloacral dysplasia with, type B lipodystrophy
ZNF711	Intellectual developmental disorder, X-linked 97