Az ORIGIN PRÉMIUM
által vizsgált betegségek listája

Gene	OMIM	Condition title
AAAS	605378	Triple-A syndrome (achalasia-addisonianism-alacrimia)
ABAT	137150	GABA-transaminase deficiency
ABCA12	607800	Ichthyosis congenital autosomal recessive type 4A and 4B (harlequin)
ABCA3	601615	Surfactant metabolism dysfunction, pulmonary, type 3
ABCC8	600509	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
ABCD1	300371	Adrenoleukodystrophy
ABHD12	613599	PHARC syndrome (polyneuropathy, hearing loss, ataxia, retinitis pigmentosa and cataract)
ACAD9	611103	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)
ACADM	607008	Medium-chain acyl-CoA dehydrogenase deficiency
ACADS	606885	Short-chain acyl-CoA dehydrogenase deficiency
ACADVL	609575	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
ACAT1	607809	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)
ACE	106180	Renal tubular dysgenesis
ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency
ACTA1	102610	Nemaline myopathy 3; Congenital fiber-type disproportion myopathy 1
ADA	608958	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)
ADAMTS10	608990	Weill-Marchesani syndrome, type 1 recessive
ADAMTS13	604134	Thrombotic thrombocytopenic purpura, familial (Schulman-Upshaw syndrome)
AFF2	300806	Intellectual developmental disorder, X-linked 109
AGA	613228	Aspartylglucosaminuria (glycosylasparaginase deficiency)
AGL	610860	Glycogen storage disease, type 3
AGPAT2	603100	Congenital generalized lipodystrophy (Berardinelli-Seip syndrome)
AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3
AGRN	103320	Myasthenic syndrome, congenital, type 8
AGXT	604285	Hyperoxaluria, primary, type 1
AHI1	608894	Joubert syndrome, type 3
AIMP1	603605	Leukodystrophy, hypomyelinating, type 3
AIRE	607358	Autoimmune polyendocrinopathy syndrome, type 1
ALDH18A1	138250	Spastic paraplegia, type 9B, autosomal recessive; Cutis laxa, type 3A (De Barsy syndrome)
ALDH3A2	609523	Sjogren-Larsson syndrome
ALDH5A1	610045	Succinic semialdehyde dehydrogenase deficiency
ALDH7A1	107323	Epilepsy, pyridoxine-dependent
ALDOB	612724	Fructose intolerance, hereditary
ALG12	607144	Congenital disorder of glycosylation, type 1G
ALG6	604566	Congenital disorder of glycosylation, type 1C
ALMS1	606844	Alström syndrome
ALOX12B	603741	Ichthyosis, congenital, autosomal recessive, type 2
ALPL	171760	Hypophosphatasia, infantile/childhood

AMACR	604489	Bile acid synthesis defect, congenital, type 4; Alpha-methylacyl-CoA racemase deficiency
AMT	238310	Glycine encephalopathy
ANO10	613726	Spinocerebellar ataxia, autosomal recessive, type 10
ANTXR2	608041	Hyaline fibromatosis syndrome
AP1S2	300629	Pettigrew syndrome
APTX	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia
AQP2	107777	Diabetes insipidus, nephrogenic, type 2
AR	313700	Androgen insensitivity syndrome, complete
ARFGEF2	605371	Periventricular heterotopia with microcephaly
ARL13B	608922	Joubert syndrome type 8
ARL6	608845	Bardet-Biedl syndrome, type 3
ARSA	607574	Metachromatic leukodystrophy
ARSB	611542	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)
ARSE	300180	Chondrodysplasia punctata, X-linked recessive
ARX	300382	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders
ASL	608310	Argininosuccinic aciduria
ASPA	608034	Canavan disease
ASPM	605481	Primary microcephaly type 5 autosomal recessive
ASS1	603470	Citrullinemia, type 1
ATM	607585	Ataxia-telangiectasia
ATP6V0A2	611716	Cutis laxa, autosomal recessive, type 2A; Wrinkly skin syndrome
ATP7A	300011	Menkes disease; Occipital horn syndrome
ATP7B	606882	Wilson disease
ATR	601215	Seckel syndrome, type 1
ATRX	300032	Intellectual disability-hypotonic facies syndrome, X-linked; Alpha-thalassemia/intellectual developmental disorder syndrome
AUH	600529	3-methylglutaconic aciduria, type 1
BBS1	209901	Bardet-Biedl syndrome, type 1
BBS10	610148	Bardet-Biedl syndrome, type 10
BBS12	610683	Bardet-Biedl syndrome, type 12
BBS2	606151	Bardet-Biedl syndrome, type 2
BBS7	607590	Bardet-Biedl syndrome, type 7
BBS9	607968	Bardet-Biedl syndrome, type 9
BCKDHA	608348	Maple syrup urine disease, type 1A
BCKDHB	248611	Maple syrup urine disease, type 1B
BCS1L	603647	Mitochondrial complex III deficiency nuclear type 1; GRACILE syndrome; Bjornstad syndrome
BEST1	607854	Bestrophinopathy, AR
BLM	604610	Bloom syndrome
BRWD3	300553	Intellectual developmental disorder, X-linked 93
BSCL2	606158	Congenital generalized lipodystrophy, type 2; Encephalopathy, progressive, with or without lipodystrophy
BTD	609019	Biotinidase deficiency
BUB1B	602860	Mosaic variegated aneuploidy syndrome 1
CAPN3	114240	Limb-girdle muscular dystrophy, type 1 (LGMD R1)
CBS	613381	Homocystinuria due to cystathionine beta-synthase

CC2D2A	612013	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2
CCDC88C	611204	Hydrocephalus, congenital, type 1
CD40LG	300386	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)
CDAN1	607465	Dyserythropoietic anemia, congenital, type 1A
CDH23	605516	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D
CDK5RAP2	608201	Primary microcephaly type 3 autosomal recessive
CENPJ	#N/D	Primary microcephaly type 6 autosomal recessive
CEP152	613529	Primary microcephaly type 9 autosomal recessive
CEP290	610142	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10
CFL2	601443	Nemaline myopathy, type 7 autosomal recessive
CFTR	602421	Cystic fibrosis
CHAT	118490	Myasthenic syndrome, congenital, type 6 presynaptic
CHM	300390	Choroideremia
CHRND	100720	Myasthenic syndrome, congenital, type 3B, fast-channel; Multiple pterygium syndrome, lethal type
CHRNE	100725	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency
CHRNG	100730	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type
CHST14	608429	Ehlers-Danlos syndrome, musculocontractural, type 1
CHST3	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations
CLCN1	118425	Myotonia congenita, recessive
CLN3	607042	Ceroid lipofuscinosis, neuronal, type 3
CLN5	608102	Ceroid lipofuscinosis, neuronal, type 5
CLN6	606725	Ceroid lipofuscinosis, neuronal, type 6
CLN8	607837	Ceroid lipofuscinosis, neuronal, type 8
CLRN1	606397	Usher syndrome, type 3A
CNGB3	605080	Achromatopsia, type 3
COG4	606976	Congenital disorder of glycosylation, type 2J
COL11A2	120290	Otospondylomegaepiphyseal dysplasia, autosomal recessive
COL17A1	113811	Epidermolysis bullosa, junctional, non-Herlitz type
COL4A3	120070	Alport syndrome, autosomal recessive, type 2
COL4A4	120131	Alport syndrome, autosomal recessive, type 2
COL4A5	303630	Alport syndrome, X-linked
COL7A1	120120	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial
COLQ	603033	Myasthenic syndrome, congenital, type 5
COQ2	609825	Primary coenzyme Q10 deficiency, type 1
CORO1A	605000	Immunodeficiency, type 8
CPS1	608307	Carbamoylphosphate synthetase 1 deficiency
CPT1A	600528	Carnitine palmitoyltransferase type 1A deficiency, hepatic
CPT2	600650	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile
CRB1	604210	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8
CRLF1	604237	Cold-induced sweating syndrome type 1
CRTAP	605497	Osteogenesis imperfecta, type 7
CSTB	601145	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts

CTNS	606272	Nephropathic cystinosis
CTSD	116840	Ceroid lipofuscinosis, neuronal, type 10
CTSK	601105	Pycnodysostosis
CUL4B	300304	Intellectual developmental disorder, X-linked syndromic, Cabezas type
CUL7	609577	3M syndrome 1
CYBB	300481	Chronic granulomatous disease, X-linked
CYP11A1	118485	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency
CYP1B1	601771	Glaucoma, primary congenital, type 3A
CYP21A2	613815	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
CYP27A1	606530	Cerebrotendinous xanthomatosis
CYP27B1	609506	Vitamin D-dependent rickets, type 1
D2HGDH	609186	D-2-hydroxyglutaric aciduria
DBH	609312	Dopamine beta-hydroxylase deficiency
DBT	248610	Maple syrup urine disease, type 2
DCLRE1C	605988	Omenn syndrome; Severe combined immunodeficiency, Athabascan type
DCX	300121	Lissencephaly, X-linked, type 1
DDC	107930	Aromatic L-amino acid decarboxylase deficiency
DGUOK	601465	DGUOK-related mitochondrial DNA depletion syndrome
DHCR7	602858	Smith-Lemli-Opitz syndrome
DHDDS	608172	Retinitis pigmentosa, type 59
DHH	605423	46,XY complete gonadal dysgenesis
DLAT	608770	Pyruvate dehydrogenase E2 deficiency
DLG3	300189	Intellectual developmental disorder, X-linked 90
DMD	300377	Duchenne/Becker muscular dystrophy
DMP1	600980	Hypophosphatemic rickets, autosomal recessive
DNAH11	603339	Ciliary dyskinesia, primary, type 7 with or without situs inversus
DNAH5	603335	Ciliary dyskinesia, primary, type 3 with or without situs inversus
DNAL1	610062	Ciliary dyskinesia, primary, type 16
DOCK8	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive
DOK7	610285	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10
DPAGT1	191350	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13
DPYD	612779	Dihydropyrimidine dehydrogenase deficiency
DYNC2H1	603297	Short-rib thoracic dysplasia, type 3 with or without polydactyly
EDA	300451	Ectodermal dysplasia, type 1 hypohidrotic, X-linked
EDNRB	131244	ABCD syndrome
EIF2AK3	604032	Wolcott-Rallison syndrome
EIF2B2	606454	Leukoencephalopathy with vanishing white matter (VWM)
EIF2B3	606273	Leukoencephalopathy with vanishing white matter (VWM)
EIF2B4	606687	Leukoencephalopathy with vanishing white matter (VWM)
EIF2B5	603945	Leukoencephalopathy with vanishing white matter (VWM)
EMD	300384	Emery-Dreifuss muscular dystrophy, type 1 X-linked
ENPP1	173335	Arterial calcification, generalized, of infancy, type 1
EPG5	615068	Vici syndrome
EPM2A	607566	Epilepsy, progressive myoclonic, type 2A (Lafora)
ERCC2	126340	Trichothiodystrophy, type 1

ERCC4	133520	Fanconi anemia, complementation group Q
ERCC5	133530	Cerebrooculofacioskeletal syndrome, type 3
ERCC6	609413	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1
ERCC8	609412	Cockayne syndrome, type A
ESCO2	609353	Roberts syndrome; Juberg-Hayward syndrome
ETFDH	231675	Glutaric acidemia, type 2C
ETHE1	608451	Ethylmalonic encephalopathy
EVC	604831	Ellis-van Creveld syndrome
EVC2	607261	Ellis-van Creveld syndrome
EXOSC3	606489	Pontocerebellar hypoplasia, type 1B
F11	264900	Factor XI deficiency
F8	300841	Hemophilia A
F9	300746	Hemophilia B
FA2H	611026	Spastic paraplegia, type 35 autosomal recessive
FAH	613871	Tyrosinemia, type 1
FAM20C	611061	Raine syndrome
FANCA	607139	Fanconi anemia, complementation group A
FANCC	613899	Fanconi anemia, complementation group C
FANCF	613897	Fanconi anemia, complementation group F
FANCG	602956	Fanconi anemia, complementation group G
FANCI	611360	Fanconi anemia, complementation group I
FGD1	300546	Aarskog-Scott syndrome; intellectual developmental disorder, X-linked syndromic, type 16
FKRP	606596	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])
FKTN	607440	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])
FMO3	136132	Trimethylaminuria
FMR1	309550	Fragile X syndrome
FRAS1	607830	Fraser syndrome, type 1
FTSJ1	300499	Intellectual developmental disorder, X-linked 9
FUCA1	612280	Fucosidosis
G6PC	613742	Glycogen storage disease, type 1A
G6PD	305900	Hemolytic anemia, G6PD deficient (favism)
GAA	606800	Glycogen storage disease, type 2
GALC	606890	Krabbe disease
GALNS	612222	Mucopolysaccharidosis, type 4A
GALT	606999	Galactosemia
GAMT	601240	Cerebral creatine deficiency syndrome, type 2
GAN	605379	Giant axonal neuropathy, type 1
GATM	602360	Cerebral creatine deficiency syndrome, type 3
GBA	606463	Gaucher Disease
GBE1	607839	Glycogen storage disease, type 4
GCDH	608801	Glutaricaciduria, type 1
GFM1	606639	Combined oxidative phosphorylation deficiency, type 1
GFPT1	138292	Myasthenia, congenital, type 12 with tubular aggregates
GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1
GJB2	121011	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6

GJB6	604418	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6
GLA	300644	Fabry disease
GLB1	611458	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)
GLDC	238300	Glycine encephalopathy
GLRA1	138491	Hyperekplexia, type 1
GNPTAB	607840	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta
GNRHR	138850	Hypogonadotropic hypogonadism, type 7 without anosmia
GOSR2	604027	Epilepsy, progressive myoclonic, type 6
GP1BA	606672	Bernard-Soulier syndrome, type A1
GP1BB	138720	Bernard-Soulier syndrome, type B
GPHN	603930	Molybdenum cofactor deficiency C
GPR143	300808	Ocular albinism, type 1 (Nettleship-Falls type)
GRHPR	604296	Hyperoxaluria, primary, type 2
GRIP1	604597	Fraser syndrome 3
HADH	601609	3-hydroxyacyl-CoA dehydrogenase deficiency
HADHA	600890	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency
HBA1	141800	Alpha-thalassemia
HBA2	141850	Alpha-thalassemia
HBB	141900	Beta-thalassemia, Sickle cell anemia and other HBB-related hemoglobinopathies
HEXA	606869	Tay-Sachs disease
HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms
HGSNAT	610453	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)
HIBCH	610690	3-hydroxyisobutryl-CoA hydrolase deficiency
HLCS	609018	Holocarboxylase synthetase deficiency
HPS3	606118	Hermansky-Pudlak syndrome, type 3
HPS4	606682	Hermansky-Pudlak syndrome, type 4
HPS6	607522	Hermansky-Pudlak syndrome, type 6
HSD17B3	605573	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
HSD17B4	601860	D-bifunctional protein deficiency
HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency
HSPG2	142461	Dyssegmental dysplasia, Silverman-Handmaker type
IDS	300823	Mucopolysaccharidosis, type 2
IDUA	252800	Mucopolysaccharidosis type 1
IGHMBP2	600502	Charcot-Marie-Tooth disease, axonal, type 2S
IL1RAPL1	300206	Intellectual developmental disorder, X-linked 21
IL2RG	308380	Severe combined immunodeficiency, X-linked
IL7R	146661	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type
INPP5E	613037	Joubert syndrome, type 1
INSR	147670	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, type A
ITGB4	147557	Epidermolysis bullosa, junctional, with pyloric atresia
IVD	607036	Isovaleric acidemia
JAK3	600173	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type
KCNJ1	600359	Bartter syndrome, type 2

KCNJ13	603208	Leber congenital amaurosis, type 16
KCTD7	611725	Epilepsy, progressive myoclonic, type 3 with or without intracellular inclusions
KDM5C	314690	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type
KIF7	611254	Acrocallosal syndrome; Joubert syndrome, type 12
L1CAM	308840	L1 Syndrome
LAMA2	156225	LAMA2-related muscular dystrophy
LAMA3	600805	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMB2	150325	Pierson syndrome; Nephrotic syndrome, type 5 with or without ocular abnormalities
LAMB3	150310	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMC2	150292	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LIFR	151443	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome
LMNA	150330	Cardiomyopathy, dilated, 1A
LPL	609708	Lipoprotein lipase deficiency
LRP2	600073	Donnai-Barrow syndrome
LRPPRC	607544	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)
LTBP4	604710	Cutis laxa, autosomal recessive, type 1C
MAN2B1	609458	Alpha-mannosidosis
MCCC2	609014	3-Methylcrotonyl-CoA carboxylase deficiency, type 2
MCOLN1	605248	Mucolipidosis type 4
MCPH1	607117	Microcephaly type 1 primary, autosomal recessive
MEFV	608107	Familial Mediterranean fever
MFN2	608507	Charcot-Marie-Tooth disease, axonal, type 2A2B
MFSD8	611124	Ceroid lipofuscinosis, neuronal, type 7
MID1	300552	Opitz GBBB syndrome, type 1
MKS1	609883	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28
MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, type cblB
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type
MMADHC	611935	Homocystinuria, cblD type, variant 1
MMUT	609058	Methylmalonic aciduria, mut(0) type
MPDU1	604041	Congenital disorder of glycosylation, type 1F
MPI	154550	Congenital disorder of glycosylation, type 1B
MPV17	137960	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE
MPZ	159440	Dejerine-Sottas disease
MTHFR	607093	Homocystinuria due to MTHFR deficiency
MTM1	300415	Myotubular myopathy, X-linked
MTO1	614667	Combined oxidative phosphorylation deficiency 10
MTR	156570	Homocystinuria-megaloblastic anemia, cblG complementation type
MUSK	601296	Fetal akinesia deformation sequence, type 1; Myasthenic syndrome, congenital, type 9 associated with acetylcholine receptor deficiency
MVK	251170	Mevalonic aciduria
MYO15A	602666	Deafness, autosomal recessive, type 3
MYO5A	160777	Griscelli syndrome, type 1
MYO5B	606540	Microvillus inclusion disease
MYO7A	276903	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2

NAGA	104170	Schindler disease, type I
NBN	602667	Nijmegen breakage syndrome
NDP	300658	Norrie disease
NEB	161650	Nemaline myopathy type 2
NEUROG3	604882	Diarrhea 4 malabsorptive, congenital
NHLRC1	608072	Epilepsy, progressive myoclonic, type 2B (Lafora)
NPC1	607623	Niemann-Pick disease, type C1
NPC2	601015	Niemann-pick disease, type C2
NPHP1	607100	Joubert syndrome type 4
NPHP3	608002	Meckel syndrome type 7
NPHP4	607215	Nephronophthisis type 4
NPHS1	602716	Nephrotic syndrome, type 1
NPHS2	604766	Nephrotic syndrome, type 2
NR0B1	300473	Adrenal hypoplasia, congenital
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis
NUP62	605815	Striatonigral degeneration, infantile
OBSL1	610991	3M syndrome 2
OCA2	611409	Oculocutaneous albinism type 2
OCRL	300535	Lowe Syndrome; Dent disease type 2
OPHN1	300127	Intellectual developmental disorder, X-linked syndromic, Billuart type
OTC	300461	Ornithine transcarbamylase deficiency
OTOF	603681	Deafness, autosomal recessive, type 9
PAH	612349	Phenylketonuria
PAK3	300142	Intellectual developmental disorder, X-linked 30
PC	608786	Pyruvate carboxylase deficiency
PCCA	232000	Propionic acidemia
PCDH15	605514	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency
PDHX	608769	Lacticacidemia due to PDX1 deficiency
PEX1	602136	Heimler syndrome type 1
PEX10	602859	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B
PEX16	603360	Peroxisome biogenesis disorder, type 8A (Zellweger syndrome); Peroxisome biogenesis disorder, type 8B
PEX7	601757	Rhizomelic chondrodysplasia punctata, type 1
PGK1	311800	Phosphoglycerate kinase 1 deficiency
PHF8	300560	Intellectual developmental disorder, X-linked syndromic, Siderius type
PKHD1	606702	Polycystic kidney disease type 4
PKP1	601975	Ectodermal dysplasia/skin fragility syndrome
PLA2G6	603604	Infantile neuroaxonal dystrophy type 1
PLEC	601282	Epidermolysis bullosa simplex with muscular dystrophy
PLOD1	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1
PLP1	300401	Pelizaeus-Merzbacher disease
PMM2	601785	Congenital disorder of glycosylation, type 1A
PNPLA1	612121	Ichthyosis, congenital, autosomal recessive, type 10
PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency

POLG	174763	POLG-related disorders
POLR1C	610060	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])
POMT1	607423	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	607439	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	173110	Pituitary hormone deficiency, combined, type 1
POU3F4	300039	Deafness, X-linked, type 2
PPT1	600722	Ceroid lipofuscinosis, neuronal, type 1
PQBP1	300463	Renpenning syndrome
PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, type 2
PRICKLE1	608500	Epilepsy, progressive myoclonic, type 1B
PROP1	601538	Pituitary hormone deficiency, combined, type 2
PRPS1	311850	Phosphoribosylpyrophosphate synthetase (PRS) deficiency
PSAP	176801	Combined SAP deficiency
PYGL	613741	Glycogen storage disease, type 6
RAB3GAP1	602536	Warburg micro syndrome; Martsolf syndrome
RAB3GAP2	609275	Warburg micro syndrome; Martsolf syndrome
RAD51C	602774	Fanconi anemia, complementation group O
RAG1	179615	Omenn syndrome; Severe combined immunodeficiency, B cell-negative
RAG2	179616	Omenn syndrome; Severe combined immunodeficiency, B cell-negative
RAPSN	601592	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11 associated with AChR deficiency
RARS2	611524	Pontocerebellar hypoplasia, type 6
RELN	600514	Lissencephaly 2 (Norman-Roberts type)
RMRP	157660	Anauxetic dysplasia 1
RNASEH2B	610326	Aicardi-Goutieres syndrome, type 2
ROR2	602337	Robinow syndrome, autosomal recessive
RP2	300757	Retinitis pigmentosa, type 2 X-linked
RPGR	312610	Retinitis pigmentosa, type 3 X-linked; Cone-rod dystrophy, X-linked, 1
RPGRIP1	605446	Leber congenital amaurosis, type 6
RPGRIP1L	610937	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome
RS1	300839	Retinoschisis
RTEL1	608833	Dyskeratosis congenita, autosomal recessive type 5
RYR1	180901	RYR1 related congenitalmyopathy
SACS	604490	Spastic ataxia, Charlevoix-Saguenay, type
SAG	181031	Oguchi disease, type 1
SBDS	607444	Shwachman-Diamond syndrome
SCN4A	603967	Myasthenic syndrome, congenital, type 16
SCN9A	603415	Indifference to pain and autosomal recessive hereditary sensory neuropathy type 2D
SCNN1B	600760	Pseudohypoaldosteronism, type 1
SCO2	604272	Mitochondrial complex IV deficiency, nuclear type 2
SERPINA1	107400	Alpha-1 antitrypsin deficiency

SGCA	600119	Limb-girdle muscular dystrophy, type 3 (LGMD R3)
SGCB	600900	Limb-girdle muscular dystrophy, type 4 (LGMD R4)
SH3TC2	608206	Charcot-Marie-Tooth disease, type 4C
SIL1	608005	Marinesco-Sjogren syndrome
SLC12A1	600839	Bartter syndrome, type 1
SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy
SLC16A2	300095	Allan-Herndon-Dudley syndrome
SLC17A5	604322	Salla disease
SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome
SLC19A3	606152	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)
SLC25A19	606521	Microcephaly, Amish type; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)
SLC25A22	609302	Developmental and epileptic encephalopathy 3
SLC26A2	606718	Achondrogenesis, type 1B (diastrophic dysplasia)
SLC26A4	605646	Deafness, autosomal recessive, type 4; Pendred syndrome
SLC2A2	138160	Fanconi-Bickel syndrome
SLC35A1	605634	Congenital disorder of glycosylation, type 2F
SLC35C1	605881	Congenital disorder of glycosylation, type 2C
SLC37A4	602671	Glycogen storage disease, type 1B
SLC3A1	104614	Cystinuria
SLC45A2	606202	Albinism, oculocutaneous, type 4
SLC4A11	610206	Corneal endothelial dystrophy, autosomal recessive
SLC6A8	300036	Cerebral creatine deficiency syndrome, type 1
SMN1	600354	Spinal muscular atrophy
SMPD1	607608	Niemann-Pick disease, type A; Niemann-Pick disease, type B
SNAP29	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome
SP110	604457	Hepatic venoocclusive disease with immunodeficiency
SPATA7	609868	Leber congenital amaurosis, type 3
SPG7	602783	Spastic paraplegia, type 7 autosomal recessive
SRD5A2	607306	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)
SRD5A3	611715	Congenital disorder of glycosylation, type 1Q; Kahrizi syndrome
ST3GAL5	604402	Salt and pepper developmental regression syndrome
STAR	600617	Lipoid adrenal hyperplasia
STIL	181590	Microcephaly, type 7 primary, autosomal recessive
STRA6	610745	Microphthalmia, isolated, with coloboma, type 8
SUOX	606887	Sulfite oxidase deficiency
SURF1	185620	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency
SYN1	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
TCTN1	609863	Joubert syndrome, type 13
TCTN2	613846	Joubert syndrome, type 24; ?Meckel syndrome, type 8
TF	190000	Atransferrinemia
TGM1	190195	Ichthyosis, congenital, autosomal recessive, type 1
TH	191290	Segawa syndrome, recessive
THOC2	300395	intellectual developmental disorder, X-linked 12
TJP2	607709	Cholestasis, progressive familial intrahepatic, type 4

TMEM138	614459	Joubert syndrome 16
TMEM216	613277	Joubert syndrome, type 2; Meckel syndrome, type 2
TMEM237	614423	Joubert syndrome, type 14
TMEM67	609884	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome
TMPRSS3	605511	Deafness, autosomal recessive, type 45573
TNNT1	191041	Nemaline myopathy , type 5 Amish type
TNXB	600985	Ehlers-Danlos syndrome, classic-like
TPK1	606370	Episodic encephalopathy due to thiamine pyrophosphokinase deficiency
TPP1	607998	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7
TREX1	606609	Aicardi-Goutieres syndrome, type 1
TRIM32	602290	Limb-girdle muscular dystrophy, type 8 (LGMD R8)
TSEN2	608753	Pontocerebellar hypoplasia, type 2B
TSEN34	608754	Pontocerebellar hypoplasia type 2C
TSEN54	608755	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4
TSFM	604723	Combined oxidative phosphorylation deficiency, type 3
TSHR	603372	Hypothyroidism, congenital, nongoitrous, type 1
TTC21B	612014	Short-rib thoracic dysplasia, type 4 with or without polydactyly
TTC8	608132	Bardet-Biedl syndrome, type 8
TTN	188840	Limb-girdle muscular dystrophy type 10 (LGMDR10); Early-onset myopathy with fatal cardiomyopathy (Salih myopathy)
TTPA	600415	Ataxia with isolated vitamin E deficiency
TYR	606933	Oculocutaneous albinism (OCA) type 1A; OCA type 1B
UBR1	605981	Johanson-Blizzard syndrome
UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, type 3
UPF3B	300298	Intellectual developmental disorder, X-linked syndromic 14
USH1C	605242	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A
USH1G	607696	Usher syndrome, type 1G
USH2A	608400	Usher syndrome, type 2A
VLDLR	192977	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1
VPS13B	607817	Cohen syndrome
VPS33B	608552	Arthrogryposis, renal dysfunction and cholestasis, type 1
WAS	300392	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked
WDR62	613583	Microcephaly, type 2 primary, autosomal recessive, with or without cortical malformations
WFS1	606201	Wolfram syndrome, type 1
WNT7A	601570	Fuhrmann syndrome
WRN	604611	Werner syndrome
XPA	611153	Xeroderma pigmentosum, group A
XPC	613208	Xeroderma pigmentosum, group C
ZAP70	176947	Autoimmune disease, multisystem, infantile-onset, type 2; Immunodeficiency, type 48
ZDHHC9	300646	Intellectual developmental disorder, X-linked syndromic, Raymond type
ZMPSTE24	606480	Mandibuloacral dysplasia with, type B lipodystrophy
ZNF711	314990	Intellectual developmental disorder, X-linked 97