Amniocentesi Virtuale

ALTAMEDICA — Istituto di Ricerca Scientifica

Elenco dei geni ricercati

Documento ufficiale di riferimento

VERSIONE 1.0 — 15 luglio 2026

Geni inclusi: 5.141

Fonte: Human Phenotype Ontology (HPO) — integra OMIM, Orphanet, DECIPHER

Questo documento elenca i geni analizzati dall'Amniocentesi Virtuale e le malattie a essi associate secondo i database internazionali di riferimento.

L'Amniocentesi Virtuale è un esame targhetizzato: indaga e refertata esclusivamente i geni riportati nel presente elenco. Nessun altro gene viene analizzato.

La presenza di un gene nell'elenco indica che la sua sequenza viene analizzata; non implica che ogni possibile variante di quel gene sia clinicamente interpretabile. L'esito dell'esame va sempre letto nell'ambito della consulenza genetica.

Il documento è richiamato nel consenso informato. La versione vigente è pubblicata su www.amniocentesivirtuale.it/geni-ricercati.php

Elenco geni e malattie associate

Totale geni: 5141. Fonte: Human Phenotype Ontology (HPO), file genes_to_disease.txt e phenotype.hpoa (integra OMIM, Orphanet, DECIPHER). Il codice della fonte è indicato tra parentesi accanto a ciascuna malattia. Geni senza associazione nota nel database HPO riportano "—".

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
A2M	—
A4GALT	OMIM:111400
A4GNT	—
AAAS	Achalasia-Addisonianism-Alacrima syndrome (OMIM:231550); Triple A syndrome (ORPHA:869)
AADAC	—
AADACL2	—
AAGAB	Keratoderma, palmoplantar, punctate type IA (OMIM:148600); Punctate palmoplantar keratoderma type 1 (ORPHA:79501)
AANAT	—
AARS1	Epileptic encephalopathy, early infantile, 29 (OMIM:616339); Leukoencephalopathy, hereditary diffuse, with spheroids 2 (OMIM:619661); Trichothiodystrophy 8, nonphotosensitive (OMIM:619691); Charcot-Marie-Tooth disease, axonal, type 2N (OMIM:613287); Trichothiodystrophy (ORPHA:33364); ORPHA:228174; Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
AARS2	Combined oxidative phosphorylation deficiency 8 (OMIM:614096); Leukoencephalopathy, progressive, with ovarian failure (OMIM:615889); ORPHA:99853; ORPHA:313808; ORPHA:319504
AASS	Hyperlysinemia, type I (OMIM:238700); Saccharopinuria (ORPHA:3124); Hyperlysinemia (ORPHA:2203)
ABALON	—
ABAT	GABA-transaminase deficiency (OMIM:613163); Gamma-aminobutyric acid transaminase deficiency (ORPHA:2066)
ABCA1	Tangier disease (OMIM:205400); HDL deficiency, familial, 1 (OMIM:604091); Tangier disease (ORPHA:31150); Apolipoprotein A-I deficiency (ORPHA:425)
ABCA10	—
ABCA12	Ichthyosis, congenital, autosomal recessive 4B (OMIM:242500); Ichthyosis, congenital, autosomal recessive 4A (OMIM:601277); Lamellar ichthyosis (ORPHA:313); Congenital ichthyosiform erythroderma (ORPHA:79394); Harlequin ichthyosis (ORPHA:457)
ABCA13	—
ABCA2	Intellectual developmental disorder with poor growth and with or without seizures or ataxia (OMIM:618808); ORPHA:88616
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3 (OMIM:610921); Pediatric acute respiratory distress syndrome (ORPHA:685082); Neonatal acute respiratory distress syndrome (ORPHA:217563); Idiopathic pulmonary fibrosis (ORPHA:2032); ORPHA:440402
ABCA4	Stargardt disease 1 (OMIM:248200); Macular degeneration, age-related, 2 (OMIM:153800); Cone-rod dystrophy 3 (OMIM:604116); Retinitis pigmentosa 19 (OMIM:601718); Cone rod dystrophy (ORPHA:1872); Retinitis pigmentosa (ORPHA:791); Stargardt disease (ORPHA:827)
ABCB1	OMIM:120080; Encephalopathy, acute transient (OMIM:620950); Inflammatory bowel disease 13 (OMIM:612244)
ABCB11	Cholestasis, progressive familial intrahepatic, 3 (OMIM:602347); Cholestasis, benign recurrent intrahepatic, 2 (OMIM:605479); Cholestasis, progressive familial intrahepatic 2 (OMIM:601847); ORPHA:99961; Intrahepatic cholestasis of pregnancy (ORPHA:69665); ORPHA:79304
ABCB4	Gallbladder disease 1 (OMIM:600803); Cholestasis, progressive familial intrahepatic, 3 (OMIM:602347); Cholestasis, intrahepatic, of pregnancy 3 (OMIM:614972); Low phospholipid-associated cholelithiasis (ORPHA:69663); Intrahepatic cholestasis of pregnancy (ORPHA:69665); ORPHA:79305
ABCB6	Pseudohyperkalemia, familial, 2, due to red cell leak (OMIM:609153); OMIM:111600; Dyschromatosis universalis hereditaria 3 (OMIM:615402); Microphthalmia, isolated, with coloboma 7 (OMIM:614497); Dyschromatosis universalis hereditaria (ORPHA:241); Familial pseudohyperkalemia (ORPHA:90044); ORPHA:98947; ORPHA:98943; ORPHA:98944; ORPHA:98945; ORPHA:98946; ORPHA:98942; ORPHA:98938
ABCB7	Anemia, sideroblastic, and spinocerebellar ataxia (OMIM:301310); X-linked sideroblastic anemia and spinocerebellar ataxia (ORPHA:2802)
ABCC1	Deafness, autosomal dominant 77 (OMIM:618915); ORPHA:90635
ABCC11	OMIM:117800
ABCC2	DUBIN-JOHNSON syndrome (OMIM:237500); Dubin-Johnson syndrome (ORPHA:234)
ABCC3	—
ABCC4	—
ABCC6	Pseudoxanthoma elasticum (OMIM:264800); Pseudoxanthoma elasticum, forme fruste (OMIM:177850); Arterial calcification, generalized, of infancy, 1 (OMIM:208000); Arterial calcification, generalized, of infancy, 2 (OMIM:614473); Pseudoxanthoma elasticum (ORPHA:758); Generalized arterial calcification of infancy (ORPHA:51608)
ABCC8	Diabetes mellitus, transient neonatal 2 (OMIM:610374); Hyperinsulinemic hypoglycemia, familial, 1 (OMIM:256450); Diabetes mellitus, noninsulin-dependent (OMIM:125853); Hypoglycemia of infancy, leucine-sensitive (OMIM:240800); Diabetes mellitus, permanent neonatal 3, with or without neurologic features (OMIM:618857); Maturity-onset diabetes of the young, type 12 (OMIM:621196); Isolated permanent neonatal diabetes mellitus (ORPHA:99885); Transient neonatal diabetes mellitus (ORPHA:99886); ORPHA:79643; Autosomal dominant hyperinsulinism due to SUR1 deficiency (ORPHA:276575); ORPHA:276598; MODY

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	(ORPHA:552); DEND syndrome (ORPHA:79134)
ABCC9	Hypertrichotic osteochondrodysplasia (OMIM:239850); Cardiomyopathy, dilated, 1O (OMIM:608569); Atrial fibrillation, familial, 12 (OMIM:614050); Intellectual disability and myopathy syndrome (OMIM:619719); Hereditary atrial fibrillation (ORPHA:334); Cantú syndrome (ORPHA:1517); Familial isolated dilated cardiomyopathy (ORPHA:154); Brugada syndrome (ORPHA:130)
ABCD1	Adrenoleukodystrophy (OMIM:300100); Hirschsprung disease (ORPHA:388); CADDS (ORPHA:369942); Adrenomyeloneuropathy (ORPHA:139399); X-linked cerebral adrenoleukodystrophy (ORPHA:139396)
ABCD3	Bile acid synthesis defect, congenital, 5 (OMIM:616278); Oculopharyngodistal myopathy 5 (OMIM:621446); Oculopharyngodistal myopathy (ORPHA:98897)
ABCD4	Methylmalonic aciduria and homocystinuria, Cblj type (OMIM:614857); ORPHA:369955
ABCG2	OMIM:614490; Uric acid concentration, serum, QTL1 (OMIM:138900)
ABCG5	Sitosterolemia 2 (OMIM:618666); Sitosterolemia (ORPHA:2882); Homozygous familial hypercholesterolemia (ORPHA:391665)
ABCG8	Gallbladder disease 4 (OMIM:611465); Sitosterolemia 1 (OMIM:210250); Sitosterolemia (ORPHA:2882); Homozygous familial hypercholesterolemia (ORPHA:391665)
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract (OMIM:612674); ORPHA:171848
ABHD14A	—
ABHD15	—
ABHD5	Chanarin-Dorfman syndrome (OMIM:275630); Neutral lipid storage disease with ichthyosis (ORPHA:98907)
ABI3BP	—
ABL1	Congenital heart defects and skeletal malformations syndrome (OMIM:617602); Leukemia, chronic myeloid (OMIM:608232); ORPHA:99861; ORPHA:585909; ORPHA:643503; Chronic myeloid leukemia (ORPHA:521)
ABL2	—
ABO	OMIM:616093
ABRAXAS1	—
ACAA1	—
ACACA	Acetyl-CoA carboxylase deficiency (OMIM:613933)
ACACB	—
ACAD10	—
ACAD11	—
ACAD8	Isobutyryl-CoA dehydrogenase deficiency (OMIM:611283); Isobutyryl-CoA dehydrogenase deficiency (ORPHA:79159)
ACAD9	Mitochondrial complex I deficiency, nuclear type 20 (OMIM:611126); Acyl-CoA dehydrogenase 9 deficiency (ORPHA:99901)
ACADL	—
ACADM	Acyl-Coa dehydrogenase, medium-chain, deficiency of (OMIM:201450); Medium chain acyl-CoA dehydrogenase deficiency (ORPHA:42)
ACADS	Acyl-Coa dehydrogenase, short-chain, deficiency of (OMIM:201470); Short chain acyl-CoA dehydrogenase deficiency (ORPHA:26792)
ACADSB	2-Methylbutyryl-Coa dehydrogenase deficiency (OMIM:610006); 2-methylbutyryl-CoA dehydrogenase deficiency (ORPHA:79157)
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency (OMIM:201475); Very long chain acyl-CoA dehydrogenase deficiency (ORPHA:26793)
ACAN	Spondyloepimetaphyseal dysplasia, Aggrecan type (OMIM:612813); Spondyloepiphyseal dysplasia, Kimberley type (OMIM:608361); Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans (OMIM:165800); Spondyloepimetaphyseal dysplasia, aggrecan type (ORPHA:171866); ORPHA:251262; Spondyloepiphyseal dysplasia, Kimberley type (ORPHA:93283); Short stature-advanced bone age-early-onset osteoarthritis syndrome (ORPHA:435804)
ACAT1	Alpha-methylacetoacetic aciduria (OMIM:203750); Beta-ketothiolase deficiency (ORPHA:134)
ACAT2	Acetyl-Coa acetyltransferase-2 deficiency (OMIM:614055)
ACBD5	Retinal dystrophy with leukodystrophy (OMIM:618863)
ACBD6	Neurodevelopmental disorder with progressive movement abnormalities (OMIM:620785)
ACCS	—
ACE	OMIM:621420; Renal tubular dysgenesis (OMIM:267430); OMIM:612624; ORPHA:97369
ACHE	OMIM:112100
ACKR1	OMIM:110700; OMIM:611862; OMIM:611162
ACLY	—
ACO2	Optic atrophy 9 (OMIM:616289); Infantile cerebellar-retinal degeneration (OMIM:614559); ORPHA:98676; ORPHA:313850
ACOX1	Mitchell syndrome (OMIM:618960); Peroxisomal acyl-CoA oxidase deficiency (OMIM:264470); ORPHA:631248; Peroxisomal acyl-CoA oxidase deficiency (ORPHA:2971)
ACP1	—
ACP2	OMIM:200950
ACP5	Spondyloenchondrodysplasia with immune dysregulation (OMIM:607944); Spondyloenchondrodysplasia (ORPHA:1855)
ACSF3	Combined malonic and methylmalonic aciduria (OMIM:614265); Combined malonic and methylmalonic acidemia (ORPHA:289504)
ACSL4	Intellectual developmental disorder, X-linked 63 (OMIM:300387); Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome (ORPHA:86818); ORPHA:777

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
ACSL5	Diarrhea 13 (OMIM:620357)
ACSL6	—
ACSM3	—
ACTA1	Nemaline myopathy 3 (OMIM:161800); Myopathy, scapulohumeroperoneal (OMIM:616852); Congenital myopathy 2B, severe infantile, autosomal recessive (OMIM:620265); Congenital myopathy 2C, severe infantile, autosomal dominant (OMIM:620278); Zebra body myopathy (ORPHA:97240); Rigid spine syndrome (ORPHA:97244); ORPHA:447977; Congenital fiber-type disproportion myopathy (ORPHA:2020); Typical nemaline myopathy (ORPHA:171436); Intermediate nemaline myopathy (ORPHA:171433); Severe congenital nemaline myopathy (ORPHA:171430); Childhood-onset nemaline myopathy (ORPHA:171439); ORPHA:98904
ACTA2	Aortic aneurysm, familial thoracic 6 (OMIM:611788); Moyamoya disease 5 (OMIM:614042); Multisystemic smooth muscle dysfunction syndrome (OMIM:613834); Moyamoya disease (ORPHA:2573); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); ORPHA:404463
ACTB	Baraitser-Winter syndrome 1 (OMIM:243310); Becker nevus syndrome (OMIM:604919); Thrombocytopenia 8, with dysmorphic features and developmental delay (OMIM:620475); Congenital smooth muscle hamartoma with or without hemihypertrophy (OMIM:620470); Dystonia, juvenile-onset (OMIM:607371); Becker nevus syndrome (ORPHA:64755); ORPHA:674653; Developmental malformations-deafness-dystonia syndrome (ORPHA:79107); Baraitser-Winter cerebrofrontofacial syndrome (ORPHA:2995); ORPHA:673556
ACTC1	Atrial septal defect 5 (OMIM:612794); Cardiomyopathy, dilated, 1R (OMIM:613424); Cardiomyopathy, familial hypertrophic, 11 (OMIM:612098); ORPHA:54260; Atrial septal defect, ostium secundum type (ORPHA:99103); Familial isolated dilated cardiomyopathy (ORPHA:154)
ACTG1	Deafness, autosomal dominant 20 (OMIM:604717); Baraitser-Winter syndrome 2 (OMIM:614583); ORPHA:90635; ORPHA:98944; ORPHA:98942; Baraitser-Winter cerebrofrontofacial syndrome (ORPHA:2995)
ACTN2	Cardiomyopathy, dilated, 1AA, with or without LVNC (OMIM:612158); Myopathy, congenital with structured cores and Z-line abnormalities (OMIM:618654); Myopathy, distal, 6, adult onset (OMIM:618655); Familial isolated dilated cardiomyopathy (ORPHA:154); ORPHA:708129; ORPHA:708133
ACTN4	Focal segmental glomerulosclerosis 1 (OMIM:603278); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
ACVR1	Fibrodysplasia ossificans progressiva (OMIM:135100); Fibrodysplasia ossificans progressiva (ORPHA:337)
ACVR1B	Pancreatic cancer (OMIM:260350)
ACVR1C	—
ACVR2A	—
ACVR2B	Heterotaxy, visceral, 4, autosomal (OMIM:613751); ORPHA:157769
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2 (OMIM:600376); ORPHA:275777; Hereditary hemorrhagic telangiectasia (ORPHA:774)
ADA	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-negative, due to adenosine deaminase deficiency (OMIM:102700); Severe combined immunodeficiency due to adenosine deaminase deficiency (ORPHA:277); Omenn syndrome (ORPHA:39041)
ADAM10	OMIM:615590; Reticulate acropigmentation of Kitamura (OMIM:615537); ORPHA:178307
ADAM12	—
ADAM17	Hypotichosis 16 (OMIM:621490); Inflammatory skin and bowel disease, neonatal, 1 (OMIM:614328); Neonatal erythroderma-autoinflammation-inflammatory bowel disease syndrome (ORPHA:294023)
ADAM19	—
ADAM22	Epileptic encephalopathy, early infantile, 61 (OMIM:617933)
ADAM23	—
ADAM33	—
ADAM7	—
ADAM9	Cone-Rod dystrophy 9 (OMIM:612775); Cone rod dystrophy (ORPHA:1872)
ADAMTS1	—
ADAMTS10	Weill-Marchesani syndrome 1 (OMIM:277600); Weill-Marchesani syndrome (ORPHA:3449)
ADAMTS13	Thrombotic thrombocytopenic purpura, hereditary (OMIM:274150); ORPHA:93583
ADAMTS17	Weill-Marchesani syndrome 4 (OMIM:613195); ORPHA:363992; Weill-Marchesani syndrome (ORPHA:3449)
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus (OMIM:615458); ORPHA:369970
ADAMTS19	Cardiac valvular dysplasia 2 (OMIM:620067)
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type (OMIM:225410); Dermatosparaxis Ehlers-Danlos syndrome (ORPHA:1901)
ADAMTSL2	Geleophysic dysplasia 1 (OMIM:231050); Geleophysic dysplasia (ORPHA:2623); Dermatosparaxis Ehlers-Danlos syndrome (ORPHA:1901)
ADAMTSL3	—
ADAMTSL4	Ectopia lentis et pupillae (OMIM:225200); Ectopia lentis (OMIM:225100); Isolated ectopia lentis (ORPHA:1885)
ADAR	Aicardi-Goutieres syndrome 6 (OMIM:615010); Dyschromatosis symmetrica hereditaria 1 (OMIM:127400); Dyschromatosis symmetrica hereditaria (ORPHA:41); Aicardi-Goutières syndrome (ORPHA:51); Familial infantile bilateral striatal necrosis (ORPHA:225154); ORPHA:694356
ADARB1	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures (OMIM:618862)
ADCY10	Hypercalciuria, absorptive, susceptibility to (OMIM:143870); Idiopathic hypercalciuria (ORPHA:2197)
ADCY3	Body mass index quantitative trait locus 19 (OMIM:617885)
ADCY5	Dyskinesia with orofacial involvement, autosomal recessive (OMIM:619647); Neurodevelopmental disorder with hyperkinetic movements and dyskinesia (OMIM:619651); Dyskinesia, familial, with facial myokymia (OMIM:606703); Benign hereditary chorea (ORPHA:1429); Familial dyskinesia and facial myokymia (ORPHA:324588)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
ADCY6	Lethal congenital contracture syndrome 8 (OMIM:616287); Hypomyelination neuropathy-arthrogryposis syndrome (ORPHA:2680)
ADCY9	—
ADD1	Hypertension, essential (OMIM:145500)
ADD2	—
ADD3	Cerebral palsy, spastic quadriplegic, 3 (OMIM:617008); ORPHA:210141
ADGRG1	Polymicrogyria, bilateral frontoparietal (OMIM:606854); Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian) (OMIM:615752); Bilateral frontoparietal polymicrogyria (ORPHA:101070); Bilateral perisylvian polymicrogyria (ORPHA:98889)
ADGRV1	Febrile seizures, familial, 4 (OMIM:604352); Usher syndrome, type IIC (OMIM:605472); Genetic epilepsy with febrile seizure plus (ORPHA:36387); Usher syndrome type 2 (ORPHA:231178)
ADH1B	Alcohol dependence (OMIM:103780)
ADH1C	Alcohol dependence (OMIM:103780); Parkinson disease, late-onset (OMIM:168600)
ADH4	—
ADH7	—
ADIPOQ	Adiponectin deficiency (OMIM:612556)
ADIPOR1	—
ADK	Hypermethioninemia due to adenosine kinase deficiency (OMIM:614300); ORPHA:289290
ADM	—
ADNP	Helsmoortel-van der Aa syndrome (OMIM:615873); Helsmoortel-Van der Aa syndrome (ORPHA:404448)
ADORA2A	Acute encephalopathy with biphasic seizures and late reduced diffusion (ORPHA:363549)
ADRA1A	—
ADRA2A	Lipodystrophy, familial partial, type 8 (OMIM:620679)
ADRA2B	Familial adult myoclonic epilepsy (ORPHA:86814)
ADRA2C	—
ADRB1	OMIM:607276; Short sleep, familial natural, 2 (OMIM:618591)
ADRB2	—
ADRB3	OBESITY (OMIM:601665)
ADSL	Adenylosuccinase deficiency (OMIM:103050); Adenylosuccinate lyase deficiency (ORPHA:46)
ADTRP	—
AFDN	—
AFF1	ORPHA:589595
AFF2	Intellectual developmental disorder, X-linked 109 (OMIM:309548); FRAXE intellectual disability (ORPHA:100973)
AFF3	KINSSHIP syndrome (OMIM:619297); ORPHA:632603
AFG3L2	Spinocerebellar ataxia 28 (OMIM:610246); Optic atrophy 12 (OMIM:618977); Spastic ataxia 5, autosomal recessive (OMIM:614487); Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome (ORPHA:313772); Spinocerebellar ataxia type 28 (ORPHA:101109)
AFP	Alpha-Fetoprotein deficiency (OMIM:615969); Alpha-Fetoprotein, hereditary persistence of (OMIM:615970); ORPHA:168615; ORPHA:168612
AGA	Aspartylglucosaminuria (OMIM:208400); Aspartylglucosaminuria (ORPHA:93)
AGBL1	Corneal dystrophy, fuchs endothelial, 8 (OMIM:615523); Fuchs endothelial corneal dystrophy (ORPHA:98974)
AGGF1	Capillary-lymphatic-venous malformation with segmental distribution (ORPHA:90308)
AGK	Sengers syndrome (OMIM:212350); Cataract, autosomal recessive congenital 5 (OMIM:614691); Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome (ORPHA:1369); ORPHA:98994
AGL	Glycogen storage disease III (OMIM:232400); Glycogen storage disease due to glycogen debranching enzyme deficiency (ORPHA:366)
AGMO	—
AGPAT2	Lipodystrophy, congenital generalized, type 1 (OMIM:608594); ORPHA:696189
AGPS	Rhizomelic chondrodysplasia punctata, type 3 (OMIM:600121); ORPHA:309803
AGRN	Myasthenic syndrome, congenital, 8 (OMIM:615120); Presynaptic congenital myasthenic syndromes (ORPHA:98914); Postsynaptic congenital myasthenic syndrome (ORPHA:98913)
AGRP	OBESITY (OMIM:601665)
AGT	Renal tubular dysgenesis (OMIM:267430); ORPHA:97369
AGTPBP1	Neurodegeneration, childhood-onset, with cerebellar atrophy (OMIM:618276); Pontocerebellar hypoplasia type 1 (ORPHA:2254)
AGTR1	Renal tubular dysgenesis (OMIM:267430); Hypertension, essential (OMIM:145500); ORPHA:97369
AGTR2	ORPHA:777
AGXT	Hyperoxaluria, primary, type I (OMIM:259900); Primary hyperoxaluria type 1 (ORPHA:93598)
AGXT2	Beta-aminoisobutyric acid, urinary excretion of (OMIM:210100)
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase (OMIM:613752); S-adenosylhomocysteine hydrolase deficiency (ORPHA:88618)
AHI1	Joubert syndrome 3 (OMIM:608629); Joubert syndrome with ocular defect (ORPHA:220493); Retinitis pigmentosa (ORPHA:791); Isolated Joubert syndrome (ORPHA:475)
AHNAK2	—
AHR	Retinitis pigmentosa 85 (OMIM:618345); Foveal hypoplasia 3 (OMIM:620958); Retinitis pigmentosa (ORPHA:791)
AHRR	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
AHSG	Alopecia-intellectual disability syndrome 1 (OMIM:203650); Alopecia-intellectual disability syndrome (ORPHA:2850)
AHSP	—
AICDA	Immunodeficiency with hyper-igm, type 2 (OMIM:605258); ORPHA:101089
AIFM1	Cowchock syndrome (OMIM:310490); Spondyloepimetaphyseal dysplasia, X-linked, with mental deterioration (OMIM:300232); Combined oxidative phosphorylation deficiency 6 (OMIM:300816); Deafness, X-linked 5 (OMIM:300614); X-linked Charcot-Marie-Tooth disease type 4 (ORPHA:101078); ORPHA:139583; Severe X-linked mitochondrial encephalomyopathy (ORPHA:238329); Leukoencephalopathy-spondyloepimetaphyseal dysplasia syndrome (ORPHA:83629)
AIMP1	Leukodystrophy, hypomyelinating, 3 (OMIM:260600); ORPHA:280293; ORPHA:88616
AIP	Pituitary adenoma, growth hormone-secreting, 1 (OMIM:102200); Pituitary adenoma 4, ACTH-secreting, somatic (OMIM:219090); Acromegaly (ORPHA:963); Prolactinoma (ORPHA:2965); ORPHA:314790; ORPHA:314786; ORPHA:314777; Pituitary gigantism (ORPHA:99725)
AIPL1	Leber congenital amaurosis 4 (OMIM:604393); Retinitis pigmentosa (OMIM:268000); Leber congenital amaurosis (ORPHA:65); Cone rod dystrophy (ORPHA:1872)
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia (OMIM:240300); ORPHA:189466; Autoimmune polyendocrinopathy type 1 (ORPHA:3453)
AJAP1	—
AK1	Adenylate kinase deficiency, hemolytic anemia due to (OMIM:612631); ORPHA:86817
AK2	Reticular dysgenesis (OMIM:267500); Reticular dysgenesis (ORPHA:33355)
AK7	Spermatogenic failure 27 (OMIM:617965); ORPHA:276234
AK8	—
AKAP10	—
AKAP9	Long QT syndrome 11 (OMIM:611820); Romano-Ward syndrome (ORPHA:101016); Brugada syndrome (ORPHA:130)
AKR1B1	—
AKR1C2	46,xy sex reversal 8 (OMIM:614279); ORPHA:443087
AKR1C3	—
AKR1C4	46,xy sex reversal 8 (OMIM:614279); ORPHA:443087
AKR1D1	Bile acid synthesis defect, congenital, 2 (OMIM:235555); Congenital bile acid synthesis defect type 2 (ORPHA:79303)
AKR7A2	—
AKT1	Cowden syndrome 6 (OMIM:615109); Breast cancer (OMIM:114480); Proteus syndrome, somatic (OMIM:176920); Colorectal cancer (OMIM:114500); Ovarian cancer (OMIM:167000); Cowden syndrome (ORPHA:201); Proteus syndrome (ORPHA:744); Meningioma (ORPHA:2495)
AKT2	Hypoinsulinemic hypoglycemia with hemihypertrophy (OMIM:240900); Diabetes mellitus, noninsulin-dependent (OMIM:125853); Hypoinsulinemic hypoglycemia and body hemihypertrophy (ORPHA:293964); AKT2-related familial partial lipodystrophy (ORPHA:79085)
AKT3	Megalencephaly-Polymicrogyria-Polydactyly-Hydrocephalus syndrome 2 (OMIM:615937); Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome (ORPHA:83473); Hemimegalencephaly (ORPHA:99802)
ALAD	Porphyria, acute hepatic (OMIM:612740); Porphyria due to ALA dehydratase deficiency (ORPHA:100924)
ALAS2	Anemia, sideroblastic, X-linked (OMIM:300751); Protoporphyria, erythropoietic, X-linked (OMIM:300752); ORPHA:443197; X-linked sideroblastic anemia (ORPHA:75563)
ALB	Familial dysalbuminemic hyperthyroxinemia (OMIM:615999); Analbuminemia (OMIM:616000); Congenital analbuminemia (ORPHA:86816)
ALDH16A1	—
ALDH18A1	Spastic paraplegia 9B, autosomal recessive (OMIM:616586); Cutis laxa, autosomal dominant 3 (OMIM:616603); Spastic paraplegia 9A, autosomal dominant (OMIM:601162); Cutis laxa, autosomal recessive, type IIIA (OMIM:219150); Autosomal dominant cutis laxa (ORPHA:90348); ALDH18A1-related De Barsy syndrome (ORPHA:35664); Autosomal dominant spastic paraplegia type 9B (ORPHA:447757); Autosomal dominant spastic paraplegia type 9A (ORPHA:447753); Autosomal recessive spastic paraplegia type 9B (ORPHA:447760)
ALDH1A1	—
ALDH1A2	Diaphragmatic hernia 4, with cardiovascular defects (OMIM:620025)
ALDH2	Alcohol sensitivity, acute (OMIM:610251)
ALDH3A2	Sjogren-Larsson syndrome (OMIM:270200); Sjögren-Larsson syndrome (ORPHA:816)
ALDH4A1	Hyperprolinemia, type II (OMIM:239510); Hyperprolinemia type 2 (ORPHA:79101)
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency (OMIM:271980); Succinic semialdehyde dehydrogenase deficiency (ORPHA:22)
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency (OMIM:614105); ORPHA:289307
ALDH7A1	Epilepsy, pyridoxine-dependent (OMIM:266100); Pyridoxine-dependent-developmental and epileptic encephalopathy (ORPHA:3006)
ALDOA	Glycogen storage disease XII (OMIM:611881); Glycogen storage disease due to aldolase A deficiency (ORPHA:57)
ALDOB	Fructose intolerance, hereditary (OMIM:229600); Hereditary fructose intolerance (ORPHA:469)
ALG1	Congenital disorder of glycosylation, type Ik (OMIM:608540); ALG1-CDG (ORPHA:79327)
ALG11	Congenital disorder of glycosylation, type Ip (OMIM:613661); ALG11-CDG (ORPHA:280071)
ALG12	Congenital disorder of glycosylation, type Ig (OMIM:607143); ALG12-CDG (ORPHA:79324)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
ALG13	Epileptic encephalopathy, early infantile, 36 (OMIM:300884); ALG13-CDG (ORPHA:324422); ORPHA:777
ALG14	Myasthenic syndrome, congenital, 15 (OMIM:616227); Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies (OMIM:619031); Myopathy, epilepsy, and progressive cerebral atrophy (OMIM:619036); Congenital myasthenic syndrome with glycosylation defect (ORPHA:353327)
ALG2	Myasthenic syndrome, congenital, 14 (OMIM:616228); Congenital disorder of glycosylation, type Ii (OMIM:607906); Congenital myasthenic syndrome with glycosylation defect (ORPHA:353327); ALG2-CDG (ORPHA:79326)
ALG3	Congenital disorder of glycosylation, type Id (OMIM:601110); ALG3-CDG (ORPHA:79321)
ALG5	Polycystic kidney disease 7 (OMIM:620056); Autosomal dominant polycystic kidney disease (ORPHA:730)
ALG6	Congenital disorder of glycosylation, type Ic (OMIM:603147); ALG6-CDG (ORPHA:79320)
ALG8	Congenital disorder of glycosylation, type Ih (OMIM:608104); Polycystic liver disease 3 with or without kidney cysts (OMIM:617874); Isolated polycystic liver disease (ORPHA:2924); ALG8-CDG (ORPHA:79325)
ALG9	Congenital disorder of glycosylation, type Il (OMIM:608776); Gillessen-Kaesbach-Nishimura syndrome (OMIM:263210); Autosomal dominant polycystic kidney disease (ORPHA:730); ALG9-CDG (ORPHA:79328)
ALK	OMIM:613014; ORPHA:178342; ORPHA:146; Ganglioneuroma (ORPHA:251992); Large/giant congenital melanocytic nevus (ORPHA:626); Neuroblastoma (ORPHA:635); ORPHA:251877; ORPHA:364043; ORPHA:300895
ALMS1	Alstrom syndrome (OMIM:203800); Alström syndrome (ORPHA:64)
ALOX12	—
ALOX12B	Ichthyosis, congenital, autosomal recessive 2 (OMIM:242100); Lamellar ichthyosis (ORPHA:313); Congenital ichthyosiform erythroderma (ORPHA:79394); Self-improving collodion baby (ORPHA:281122)
ALOX15	—
ALOX5	Asthma, susceptibility to (OMIM:600807)
ALOX5AP	Ischemic stroke, susceptibility to (OMIM:601367)
ALOXE3	Ichthyosis, congenital, autosomal recessive 2 (OMIM:242100); Ichthyosis, congenital, autosomal recessive 3 (OMIM:606545); Lamellar ichthyosis (ORPHA:313); Congenital ichthyosiform erythroderma (ORPHA:79394); Self-improving collodion baby (ORPHA:281122)
ALPL	Hypophosphatasia, adult (OMIM:146300); Hypophosphatasia, infantile (OMIM:241500); Hypophosphatasia, childhood (OMIM:241510); ORPHA:247685; ORPHA:247676; ORPHA:247667; ORPHA:247651; ORPHA:247638; ORPHA:247623
ALS2	Amyotrophic lateral sclerosis 2, juvenile (OMIM:205100); Spastic paralysis, infantile-onset ascending (OMIM:607225); Primary lateral sclerosis, juvenile (OMIM:606353); Juvenile amyotrophic lateral sclerosis (ORPHA:300605); Infantile-onset ascending hereditary spastic paralysis (ORPHA:293168); Juvenile primary lateral sclerosis (ORPHA:247604)
ALS2CL	—
ALX1	Frontonasal dysplasia 3 (OMIM:613456); Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome (ORPHA:306542)
ALX3	Frontonasal dysplasia 1 (OMIM:136760); Frontorhiny (ORPHA:391474)
ALX4	Craniosynostosis 5, susceptibility to (OMIM:615529); Parietal foramina 2 (OMIM:609597); Frontonasal dysplasia 2 (OMIM:613451); Frontonasal dysplasia-alopecia-genital anomalies syndrome (ORPHA:228390); Enlarged parietal foramina (ORPHA:60015); Non-syndromic sagittal craniosynostosis (ORPHA:35093); Potocki-Shaffer syndrome (ORPHA:52022)
AMACR	Alpha-methylacyl-CoA racemase deficiency (OMIM:614307); Bile acid synthesis defect, congenital, 4 (OMIM:214950); Congenital bile acid synthesis defect type 4 (ORPHA:79095)
AMBN	Amelogenesis imperfecta, type IF (OMIM:616270); ORPHA:100031
AMELX	Amelogenesis imperfecta, type IE (OMIM:301200); ORPHA:100033
AMELY	—
AMER1	Osteopathia striata with cranial sclerosis (OMIM:300373); Osteopathia striata-cranial sclerosis syndrome (ORPHA:2780)
AMH	Persistent mullerian duct syndrome, types I and II (OMIM:261550); Persistent Müllerian duct syndrome (ORPHA:2856)
AMHR2	Persistent mullerian duct syndrome, types I and II (OMIM:261550); Persistent Müllerian duct syndrome (ORPHA:2856)
AMN	Imerslund-Grasbeck syndrome 2 (OMIM:618882); Imerslund-Gräsbeck syndrome (ORPHA:35858)
AMPD1	Myopathy due to myoadenylate deaminase deficiency (OMIM:615511); Adenosine monophosphate deaminase deficiency (ORPHA:45)
AMPD3	AMP deaminase deficiency, erythrocytic (OMIM:612874); Adenosine monophosphate deaminase deficiency (ORPHA:45)
AMT	Glycine encephalopathy 2 (OMIM:620398); ORPHA:289857; ORPHA:289863; ORPHA:289860
ANAPC15	—
ANAPC4	—
ANGPT2	Lymphatic malformation 10 (OMIM:619369); ORPHA:363999
ANGPTL3	Hypobetalipoproteinemia, familial, 2 (OMIM:605019)
ANGPTL4	OMIM:615881
ANGPTL5	—
ANGPTL6	Familial cerebral saccular aneurysm (ORPHA:231160)
ANGPTL8	—
ANK1	Spherocytosis, type 1 (OMIM:182900); Hereditary spherocytosis (ORPHA:822); 8p11.2 deletion syndrome (ORPHA:251066)

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ANK2	Cardiac arrhythmia, ankyrin-B-related (OMIM:600919); Romano-Ward syndrome (ORPHA:101016)
ANK3	Intellectual developmental disorder, autosomal recessive 37 (OMIM:615493); ANK3-related intellectual disability-sleep disturbance syndrome (ORPHA:356996)
ANKH	Chondrocalcinosis 2 (OMIM:118600); Craniometaphyseal dysplasia (OMIM:123000); Familial calcium pyrophosphate deposition (ORPHA:1416); Craniometaphyseal dysplasia (ORPHA:1522)
ANKK1	—
ANKRD1	Familial isolated dilated cardiomyopathy (ORPHA:154)
ANKRD11	KBG syndrome (OMIM:148050); KBG syndrome (ORPHA:2332); 16q24.3 microdeletion syndrome (ORPHA:261250)
ANKRD26	Thrombocytopenia 2 (OMIM:188000); ORPHA:168629
ANKRD27	—
ANKS1A	—
ANKS1B	—
ANKS6	Nephronophthisis 16 (OMIM:615382); ORPHA:93591; ORPHA:93592
ANO10	Spinocerebellar ataxia, autosomal recessive 10 (OMIM:613728); Adult-onset autosomal recessive cerebellar ataxia (ORPHA:284289)
ANO3	Dystonia 24 (OMIM:615034); Cranio-cervical dystonia with laryngeal and upper-limb involvement (ORPHA:420485)
ANO5	Miyoshi muscular dystrophy 3 (OMIM:613319); Muscular dystrophy, limb-girdle, type 2L (OMIM:611307); Gnathodiaphyseal dysplasia (OMIM:166260); Gnathodiaphyseal dysplasia (ORPHA:53697); Distal anoctaminopathy (ORPHA:399096); ORPHA:689021; Isolated asymptomatic elevation of creatine phosphokinase (ORPHA:206599); Anoctamin-5-related limb-girdle muscular dystrophy R12 (ORPHA:206549)
ANO6	Scott syndrome (OMIM:262890); Scott syndrome (ORPHA:806)
ANO7	—
ANOS1	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Hypogonadotropic hypogonadism 1 with or without anosmia (OMIM:308700); Kallmann syndrome (ORPHA:478)
ANTXR1	Hemangioma, capillary infantile (OMIM:602089); GAPO syndrome (OMIM:230740); GAPO syndrome (ORPHA:2067)
ANTXR2	Hyaline fibromatosis syndrome (OMIM:228600); Juvenile hyaline fibromatosis (ORPHA:2028); Infantile systemic hyalinosis (ORPHA:2176)
ANXA5	Pregnancy loss, recurrent, susceptibility to, 3 (OMIM:614391)
AOAH	—
AOPEP	Dystonia 31 (OMIM:619565)
AP1S1	MEDNIK syndrome (OMIM:609313); MEDNIK syndrome (ORPHA:171851)
AP1S2	Pettigrew syndrome (OMIM:304340); Fried syndrome (ORPHA:85335); X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome (ORPHA:85329); X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome (ORPHA:1568)
AP2B1	—
AP3B1	Hermansky-Pudlak syndrome 2 (OMIM:608233); ORPHA:664500
AP3B2	Developmental and epileptic encephalopathy 48 (OMIM:617276); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
AP3D1	Hermansky-Pudlak syndrome 10 (OMIM:617050); X-linked recessive ocular albinism (ORPHA:54); ORPHA:664511; Ocular albinism with late-onset sensorineural deafness (ORPHA:1000)
AP4B1	Spastic paraplegia 47, autosomal recessive (OMIM:614066); Severe intellectual disability and progressive spastic paraplegia (ORPHA:280763)
AP4E1	Stuttering, familial persistent, 1 (OMIM:184450); Spastic paraplegia 51, autosomal recessive (OMIM:613744); Severe intellectual disability and progressive spastic paraplegia (ORPHA:280763)
AP4M1	Spastic paraplegia 50, autosomal recessive (OMIM:612936); Severe intellectual disability and progressive spastic paraplegia (ORPHA:280763)
AP4S1	Spastic paraplegia 52, autosomal recessive (OMIM:614067); Severe intellectual disability and progressive spastic paraplegia (ORPHA:280763)
AP5Z1	Spastic paraplegia 48, autosomal recessive (OMIM:613647); Autosomal recessive spastic paraplegia type 48 (ORPHA:306511)
APAF1	—
APBA2	—
APBB1IP	—
APBB2	—
APC	Desmoid disease, hereditary (OMIM:135290); Gastric adenocarcinoma and proximal polyposis of the stomach (OMIM:619182); Adenomatous polyposis coli (OMIM:175100); Colorectal cancer (OMIM:114500); Hepatocellular carcinoma (OMIM:114550); Gastric cancer, somatic (OMIM:613659); Attenuated familial adenomatous polyposis (ORPHA:220460); Cenani-Lenz syndrome (ORPHA:3258); ORPHA:314022; 5q22 microdeletion syndrome (ORPHA:261584); Desmoid tumor (ORPHA:873)
APCDD1	Hypotrichosis 1 (OMIM:605389); Hypotrichosis simplex (ORPHA:55654)
APEX1	—
APIP	—
APLF	—
APOA1	Amyloidosis, hereditary systemic 3 (OMIM:620657); Hypoalphalipoproteinemia, primary, 2, intermediate (OMIM:619836); Hypoalphalipoproteinemia, primary, 2 (OMIM:618463); Apolipoprotein A-I deficiency (ORPHA:425); ORPHA:93560

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APOA2	Hypercholesterolemia, familial, 1 (OMIM:143890); Apolipoprotein A-II deficiency] (OMIM:621417); ORPHA:238269
APOA4	Tubulointerstitial kidney disease, autosomal dominant 6 (OMIM:621106)
APOA5	Hypertriglyceridemia, familial (OMIM:145750); Hyperlipoproteinemia, type V (OMIM:144650); ORPHA:530849
APOB	Hypobetalipoproteinemia, familial, 1 (OMIM:615558); Hypercholesterolemia, familial, 2 (OMIM:144010); Homozygous familial hypercholesterolemia (ORPHA:391665)
APOBEC1	—
APOBEC3H	—
APOC2	Apolipoprotein C-II deficiency (OMIM:207750); ORPHA:309020
APOC4	—
APOD	—
APOE	Lipoprotein glomerulopathy (OMIM:611771); Sea-Blue histiocyte disease (OMIM:269600); Macular degeneration, age-related, 1 (OMIM:603075); Alzheimer disease 4 (OMIM:606889); Alzheimer disease 2 (OMIM:104310); OMIM:617347; Alzheimer disease 3 (OMIM:607822); ORPHA:329481; Dysbetalipoproteinemia (ORPHA:412)
APOH	—
APOL1	Focal segmental glomerulosclerosis 4, susceptibility to (OMIM:612551); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
APOL3	—
APOOL	—
APP	Cerebral amyloid angiopathy, APP-related (OMIM:605714); Alzheimer disease (OMIM:104300); Early-onset autosomal dominant Alzheimer disease (ORPHA:1020); ABeta amyloidosis, Arctic type (ORPHA:324723); ORPHA:324718; ABeta amyloidosis, Italian type (ORPHA:324713); ABetaL34V amyloidosis (ORPHA:324703); ABeta amyloidosis, Iowa type (ORPHA:324708); ABeta amyloidosis, Dutch type (ORPHA:100006)
APRT	Adenine phosphoribosyltransferase deficiency (OMIM:614723); Adenine phosphoribosyltransferase deficiency (ORPHA:976)
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia (OMIM:208920); Coenzyme Q10 deficiency, primary, 1 (OMIM:607426); Ataxia-oculomotor apraxia type 1 (ORPHA:1168)
AQP1	OMIM:110450
AQP2	Diabetes insipidus, nephrogenic, 2 (OMIM:125800); Arginine vasopressin resistance (ORPHA:223)
AQP3	OMIM:607457
AQP4	Megalencephalic leukoencephalopathy with subcortical cysts 4, remitting (OMIM:620448)
AQP5	Palmoplantar keratoderma, Bothnian type (OMIM:600231); Diffuse palmoplantar keratoderma, Bothnian type (ORPHA:2337)
AQP7	OMIM:614411
AR	Spinal and bulbar muscular atrophy, X-linked 1 (OMIM:313200); Androgen insensitivity syndrome (OMIM:300068); Prostate cancer, susceptibility to (OMIM:301120); Reifenstein syndrome (OMIM:312300); Hypospadias 1, X-linked (OMIM:300633); Partial androgen insensitivity syndrome (ORPHA:90797); Non-syndromic posterior hypospadias (ORPHA:95706); Complete androgen insensitivity syndrome (ORPHA:99429); Kennedy disease (ORPHA:481)
ARF4	—
ARFGEF1	Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures (OMIM:619964)
ARFGEF2	Periventricular heterotopia with microcephaly, autosomal recessive (OMIM:608097); Periventricular nodular heterotopia (ORPHA:98892)
ARG1	Argininemia (OMIM:207800); Argininemia (ORPHA:90)
ARHGAP18	—
ARHGAP19	Charcot-Marie-Tooth disease, axonal, type 2KK (OMIM:621466)
ARHGAP24	Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
ARHGAP26	Juvenile myelomonocytic leukemia (OMIM:607785)
ARHGAP31	Adams-Oliver syndrome 1 (OMIM:100300); Adams-Oliver syndrome (ORPHA:974)
ARHGAP4	—
ARHGAP44	—
ARHGAP45	—
ARHGAP6	—
ARHGAP9	—
ARHGDIA	Nephrotic syndrome, type 8 (OMIM:615244); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
ARHGEF10	Slowed nerve conduction velocity, autosomal dominant (OMIM:608236); ORPHA:140481
ARHGEF12	—
ARHGEF18	Retinitis pigmentosa 78 (OMIM:617433); Retinitis pigmentosa (ORPHA:791)
ARHGEF39	—
ARHGEF6	ORPHA:777
ARHGEF7	—
ARHGEF9	Developmental and epileptic encephalopathy 8 (OMIM:300607); Hyperekplexia-epilepsy syndrome (ORPHA:163985)
ARID1A	Coffin-Siris syndrome 2 (OMIM:614607); Coffin-Siris syndrome (ORPHA:1465)
ARID1B	Coffin-Siris syndrome 1 (OMIM:135900); Coffin-Siris syndrome (ORPHA:1465); 6q25.2q25.3 microdeletion syndrome (ORPHA:251056)
ARID4A	—
ARID4B	—

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ARL10	—
ARL13B	Joubert syndrome 8 (OMIM:612291); Isolated Joubert syndrome (ORPHA:475)
ARL14EP	—
ARL3	Joubert syndrome 35 (OMIM:618161); Retinitis pigmentosa 83 (OMIM:618173); Retinitis pigmentosa (ORPHA:791); Isolated Joubert syndrome (ORPHA:475)
ARL6	Bardet-Biedl syndrome 3 (OMIM:600151); Retinitis pigmentosa (OMIM:268000); Bardet-Biedl syndrome 1 (OMIM:209900); Retinitis pigmentosa 55 (OMIM:613575); Bardet-Biedl syndrome (ORPHA:110); Retinitis pigmentosa (ORPHA:791)
ARMS2	OMIM:613778
ARPC1B	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia (OMIM:617718); ORPHA:714496
ARSA	Metachromatic leukodystrophy (OMIM:250100); Metachromatic leukodystrophy, adult form (ORPHA:309271); Metachromatic leukodystrophy, juvenile form (ORPHA:309263); Metachromatic leukodystrophy, late infantile form (ORPHA:309256)
ARSB	Mucopolysaccharidosis, type VI (OMIM:253200); ORPHA:276212; ORPHA:276223
ARSL	Chondrodysplasia punctata 1, X-linked recessive (OMIM:302950); Brachytelephalangic chondrodysplasia punctata (ORPHA:79345)
ARX	Partington syndrome (OMIM:309510); Developmental and epileptic encephalopathy 1 (OMIM:308350); Corpus callosum, agenesis of, with abnormal genitalia (OMIM:300004); Intellectual developmental disorder, X-linked 29 (OMIM:300419); Lissencephaly, X-linked, 2 (OMIM:300215); X-linked spasticity-intellectual disability-epilepsy syndrome (ORPHA:3175); Corpus callosum agenesis-abnormal genitalia syndrome (ORPHA:2508); Infantile epileptic spasms syndrome (ORPHA:697160); ORPHA:777; Early infantile developmental and epileptic encephalopathy (ORPHA:1934); ORPHA:364063; Partington syndrome (ORPHA:94083); X-linked lissencephaly with abnormal genitalia (ORPHA:452)
AS	—
AS1	—
AS2	—
AS3	—
AS3MT	—
ASAH1	Spinal muscular atrophy with progressive myoclonic epilepsy (OMIM:159950); Farber lipogranulomatosis (OMIM:228000); Farber disease (ORPHA:333); Spinal muscular atrophy-progressive myoclonic epilepsy syndrome (ORPHA:2590)
ASB1	—
ASB10	Glaucoma 1, open angle, F (OMIM:603383)
ASCC1	Barrett esophagus (OMIM:614266); Spinal muscular atrophy with congenital bone fractures 2 (OMIM:616867); ORPHA:486811
ASCC3	Intellectual developmental disorder, autosomal recessive 81 (OMIM:620700)
ASCL1	Haddad syndrome (ORPHA:99803)
ASIC3	—
ASIP	OMIM:611742
ASL	Argininosuccinic aciduria (OMIM:207900); Argininosuccinic aciduria (ORPHA:23)
ASMT	—
ASNS	Asparagine synthetase deficiency (OMIM:615574); ORPHA:391376
ASPA	Canavan disease (OMIM:271900); Mild Canavan disease (ORPHA:314918); Severe Canavan disease (ORPHA:314911)
ASPHD1	—
ASPHD2	—
ASPM	Microcephaly 5, primary, autosomal recessive (OMIM:608716); Autosomal recessive primary microcephaly (ORPHA:2512)
ASPN	OMIM:603932; Hand osteoarthritis (OMIM:607850)
ASPRV1	Ichthyosis, lamellar, autosomal dominant (OMIM:146750); Lamellar ichthyosis (ORPHA:313)
ASS1	Citrullinemia, classic (OMIM:215700); ORPHA:247573; ORPHA:247546
ASTE1	—
ASTN2	—
ASXL1	Myelodysplastic syndrome (OMIM:614286); Bohring-Opitz syndrome (OMIM:605039); Bohring-Opitz syndrome (ORPHA:97297); Aggressive systemic mastocytosis (ORPHA:98850); Systemic mastocytosis with associated hematologic neoplasm (ORPHA:98849); ORPHA:86845; ORPHA:98823
ATAD1	Hyperekplexia 4 (OMIM:618011); Hereditary hyperekplexia (ORPHA:3197)
ATCAY	Cerebellar ataxia, Cayman type (OMIM:601238); Cerebellar ataxia, Cayman type (ORPHA:94122)
ATF3	—
ATF6	Achromatopsia 7 (OMIM:616517); Cone rod dystrophy (ORPHA:1872); Achromatopsia (ORPHA:49382)
ATF7IP	—
ATG16L1	OMIM:611081
ATG2B	—
ATG7	Spinocerebellar ataxia, autosomal recessive 31 (OMIM:619422)
ATG9B	—
ATIC	AICA-ribosiduria due to ATIC deficiency (OMIM:608688); AICA-ribosiduria (ORPHA:250977)
ATL1	Spastic paraplegia 3, autosomal dominant (OMIM:182600); Neuropathy, hereditary sensory, type ID (OMIM:613708); Hereditary sensory and autonomic neuropathy type 1 (ORPHA:36386); Autosomal dominant

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	spastic paraplegia type 3 (ORPHA:100984)
ATM	Ataxia-telangiectasia (OMIM:208900); Breast cancer (OMIM:114480); ORPHA:370109; Mantle cell lymphoma (ORPHA:52416); Hereditary breast and/or ovarian cancer syndrome (ORPHA:145); Ataxia-telangiectasia (ORPHA:100); ORPHA:227535; ORPHA:1331; ORPHA:67038; Familial colorectal cancer Type X (ORPHA:440437)
ATN1	Dentatorubral-pallidoluysian atrophy (OMIM:125370); Congenital hypotonia, epilepsy, developmental delay, and digital anomalies (OMIM:618494); Dentatorubral pallidoluysian atrophy (ORPHA:101)
ATOH7	Persistent hyperplastic primary vitreous, autosomal recessive (OMIM:221900); ORPHA:289499; Persistent hyperplastic primary vitreous (ORPHA:91495)
ATP10A	Angelman syndrome due to imprinting defect in 15q11-q13 (ORPHA:411515)
ATP10D	—
ATP13A2	Kufor-Rakeb syndrome (OMIM:606693); Spastic paraplegia 78, autosomal recessive (OMIM:617225); Autosomal recessive spastic paraplegia type 78 (ORPHA:513436); Kufor-Rakeb syndrome (ORPHA:306674); CLN12 disease (ORPHA:314632)
ATP13A4	—
ATP1A2	Alternating hemiplegia of childhood 1 (OMIM:104290); Developmental and epileptic encephalopathy 98 (OMIM:619605); Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies (OMIM:619602); Migraine, familial hemiplegic, 2 (OMIM:602481); Non-specific early-onset epileptic encephalopathy (ORPHA:442835); Familial or sporadic hemiplegic migraine (ORPHA:569); Alternating hemiplegia of childhood (ORPHA:2131)
ATP1A3	Dystonia 12 (OMIM:128235); Developmental and epileptic encephalopathy 99 (OMIM:619606); Cerebellar ataxia, areflexia, pes cavus, optic atrophy, and sensorineural hearing loss (OMIM:601338); Alternating hemiplegia of childhood 2 (OMIM:614820); Rapid-onset dystonia-parkinsonism (ORPHA:71517); Non-specific early-onset epileptic encephalopathy (ORPHA:442835); Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome (ORPHA:1171); Alternating hemiplegia of childhood (ORPHA:2131)
ATP1B1	Hypertension, essential (OMIM:145500)
ATP2A1	Brody myopathy (OMIM:601003); ORPHA:53347
ATP2A2	Acrokeratosis verruciformis (OMIM:101900); Rhabdomyolysis, susceptibility to, 2 (OMIM:621236); Darier-White disease (OMIM:124200); Darier disease (ORPHA:218); Acrokeratosis verruciformis of Hopf (ORPHA:79151)
ATP2A3	—
ATP2B2	Deafness, autosomal dominant 82 (OMIM:619804); ORPHA:90636
ATP2B3	Spinocerebellar ataxia, X-linked 1 (OMIM:302500); X-linked non progressive cerebellar ataxia (ORPHA:314978)
ATP2B4	—
ATP2C1	Benign chronic pemphigus (OMIM:169600); Hailey-Hailey disease (ORPHA:2841)
ATP5MF	—
ATP6AP2	Parkinsonism with spasticity, X-linked (OMIM:300911); Congenital disorder of glycosylation, type IIr (OMIM:301045); Intellectual developmental disorder, X-linked syndromic, Hedera type (OMIM:300423); X-linked intellectual disability, Hedera type (ORPHA:93952); X-linked parkinsonism-spasticity syndrome (ORPHA:363654)
ATP6V0A2	Wrinkly skin syndrome (OMIM:278250); Cutis laxa, autosomal recessive, type IIA (OMIM:219200); Wrinkly skin syndrome (ORPHA:2834); Autosomal recessive cutis laxa type 2, classic type (ORPHA:357074)
ATP6V0A4	Distal renal tubular acidosis 3, with or without sensorineural hearing loss (OMIM:602722); ORPHA:402041
ATP6V0D1	—
ATP6V1B1	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss (OMIM:267300); ORPHA:402041
ATP6V1H	—
ATP7A	Menkes disease (OMIM:309400); Spinal muscular atrophy, distal, X-linked 3 (OMIM:300489); Occipital horn syndrome (OMIM:304150); Hirschsprung disease (ORPHA:388); ORPHA:139557; Occipital horn syndrome (ORPHA:198); Menkes disease (ORPHA:565)
ATP7B	Wilson disease (OMIM:277900); Wilson disease (ORPHA:905)
ATP8A2	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4 (OMIM:615268); Dysequilibrium syndrome (ORPHA:1766)
ATP8A2P3	—
ATP8B1	Cholestasis, benign recurrent intrahepatic 1 (OMIM:243300); Cholestasis, intrahepatic, of pregnancy, 1 (OMIM:147480); Cholestasis, progressive familial intrahepatic, 3 (OMIM:602347); Cholestasis, progressive familial intrahepatic 1 (OMIM:211600); ORPHA:99960; Intrahepatic cholestasis of pregnancy (ORPHA:69665); ORPHA:79306
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1 (OMIM:604273); Isolated ATP synthase deficiency (ORPHA:254913)
ATR	Cutaneous telangiectasia and cancer syndrome, familial (OMIM:614564); Seckel syndrome 1 (OMIM:210600); ORPHA:313846; Seckel syndrome (ORPHA:808)
ATRIP	Seckel syndrome (ORPHA:808)
ATRNL1	—
ATRX	Intellectual disability-hypotonic facies syndrome, X-linked (OMIM:309580); Alpha-thalassemia/impaired intellectual development syndrome (OMIM:301040); Alpha-Thalassemia myelodysplasia syndrome (OMIM:300448); Cushing disease (ORPHA:96253); X-linked alpha-thalassemia-intellectual disability syndrome (ORPHA:847); Neuroendocrine tumor of stomach (ORPHA:100075); Alpha-thalassemia-myelodysplastic syndrome (ORPHA:231401)
ATXN1	Spinocerebellar ataxia 1 (OMIM:164400); Spinocerebellar ataxia type 1 (ORPHA:98755)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
ATXN10	Spinocerebellar ataxia 10 (OMIM:603516); Spinocerebellar ataxia type 10 (ORPHA:98761)
ATXN2	Spinocerebellar ataxia 2 (OMIM:183090); Parkinson disease, late-onset (OMIM:168600); Spinocerebellar ataxia type 2 (ORPHA:98756); Amyotrophic lateral sclerosis (ORPHA:803)
ATXN3	Machado-Joseph disease (OMIM:109150); Parkinson disease, late-onset (OMIM:168600); Machado-Joseph disease type 1 (ORPHA:276238); Machado-Joseph disease type 3 (ORPHA:276244); Machado-Joseph disease type 2 (ORPHA:276241)
ATXN3L	—
ATXN7	Spinocerebellar ataxia 7 (OMIM:164500); Spinocerebellar ataxia type 7 (ORPHA:94147)
ATXN7L3	Harel-Tora neurodevelopmental syndrome (OMIM:621377)
AUH	3-methylglutaconic aciduria, type I (OMIM:250950); 3-methylglutaconic aciduria type 1 (ORPHA:67046)
AURKA	Colorectal cancer (OMIM:114500)
AURKC	Male infertility with large-headed, multiflagellar, polyploid spermatozoa (OMIM:243060); ORPHA:137893
AUTS2	Intellectual developmental disorder, autosomal dominant 26 (OMIM:615834); Autism spectrum disorder due to AUTS2 deficiency (ORPHA:352490); ORPHA:641372
AVIL	Nephrotic syndrome, type 21 (OMIM:618594); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
AVP	Diabetes insipidus, Neurohypophyseal type (OMIM:125700); Hereditary arginine vasopressin deficiency (ORPHA:30925)
AVPR1A	—
AVPR2	Diabetes insipidus, nephrogenic, X-linked (OMIM:304800); Nephrogenic syndrome of inappropriate antidiuresis (OMIM:300539); Arginine vasopressin resistance (ORPHA:223); ORPHA:93606
AVPR3	—
AVPR4	—
AXDND1	—
AXIN1	Craniometadiaphyseal osteosclerosis with hip dysplasia (OMIM:620558); Hepatocellular carcinoma (OMIM:114550); Caudal duplication anomaly (OMIM:607864); ORPHA:210159
AXIN2	Colorectal cancer (OMIM:114500); Oligodontia-colorectal cancer syndrome (OMIM:608615); AXIN2-related polyposis (ORPHA:401911)
AXL	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110)
AZIN2	—
B	—
B2M	Amyloidosis, hereditary systemic 6 (OMIM:620659); Immunodeficiency 43 (OMIM:241600); ORPHA:34592; Variant ABeta2M amyloidosis (ORPHA:314652)
B3GALNT1	OMIM:615021; OMIM:111400
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11) (OMIM:615181); Walker-Warburg syndrome (ORPHA:899); ORPHA:88616; Muscle-eye-brain disease (ORPHA:588)
B3GALT1	—
B3GALT2	—
B3GALT5	—
B3GALT6	Al-Gazali syndrome (OMIM:609465); Ehlers-Danlos syndrome, spondylodysplastic type, 2 (OMIM:615349); Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures (OMIM:271640); ORPHA:642099; B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome (ORPHA:536467)
B3GAT1	—
B3GAT2	—
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects (OMIM:245600); ORPHA:284139
B3GLCT	Peters-plus syndrome (OMIM:261540); Peters plus syndrome (ORPHA:709)
B3GNT2	—
B3GNT3	—
B3GNT4	—
B3GNT5	—
B3GNT6	—
B3GNT7	—
B3GNT8	—
B4GALNT1	Spastic paraplegia 26, autosomal recessive (OMIM:609195); Autosomal recessive spastic paraplegia type 26 (ORPHA:101006)
B4GALNT2	OMIM:615018
B4GALNT3	—
B4GALNT4	—
B4GALT1	Congenital disorder of glycosylation, type IID (OMIM:607091); Combined low LDL and fibrinogen (OMIM:620364); B4GALT1-CDG (ORPHA:79332)
B4GALT2	—
B4GALT3	—
B4GALT4	—
B4GALT5	—
B4GALT6	—
B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic type, 1 (OMIM:130070); B4GALT7-related spondylodysplastic Ehlers-Danlos syndrome (ORPHA:75496)
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13 (OMIM:615287); Walker-Warburg syndrome (ORPHA:899)

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B9D1	Meckel syndrome, type 9 (OMIM:614209); Joubert syndrome 27 (OMIM:617120); Meckel syndrome (ORPHA:564); Isolated Joubert syndrome (ORPHA:475)
BAALC	—
BAAT	Bile acid conjugation defect 1 (OMIM:619232); ORPHA:238475
BACH1	—
BAG3	Charcot-Marie-Tooth disease, axonal, type 2JJ (OMIM:621095); Neuronopathy, distal hereditary motor, autosomal dominant 15 (OMIM:621094); Myopathy, myofibrillar, 6 (OMIM:612954); Cardiomyopathy, dilated, 1HH (OMIM:613881); ORPHA:199340; Familial isolated dilated cardiomyopathy (ORPHA:154)
BANF1	Nestor-Guillermo progeria syndrome (OMIM:614008); ORPHA:280576
BANK1	Systemic lupus erythematosus (ORPHA:536)
BAP1	Tumor predisposition syndrome (OMIM:614327); Melanoma, uveal, susceptibility to, 2 (OMIM:606661); Kury-Isidor syndrome (OMIM:619762); ORPHA:289539; Uveal melanoma (ORPHA:39044); Familial melanoma (ORPHA:618); Meningioma (ORPHA:2495); Pleural mesothelioma (ORPHA:50251); ORPHA:528084
BARD1	Breast cancer (OMIM:114480); Hereditary breast and/or ovarian cancer syndrome (ORPHA:145)
BARX2	—
BAX	Leukemia, acute lymphocytic, susceptibility to, 1 (OMIM:613065); Colorectal cancer (OMIM:114500)
BAZ1A	—
BAZ1B	Williams syndrome (ORPHA:904)
BBOF1	—
BBS1	Bardet-Biedl syndrome 1 (OMIM:209900); Bardet-Biedl syndrome (ORPHA:110); Retinitis pigmentosa (ORPHA:791)
BBS10	Bardet-Biedl syndrome 10 (OMIM:615987); Bardet-Biedl syndrome (ORPHA:110)
BBS12	Bardet-Biedl syndrome 12 (OMIM:615989); Bardet-Biedl syndrome (ORPHA:110)
BBS2	Retinitis pigmentosa 74 (OMIM:616562); Bardet-Biedl syndrome 2 (OMIM:615981); Bardet-Biedl syndrome (ORPHA:110); Retinitis pigmentosa (ORPHA:791)
BBS4	Bardet-Biedl syndrome 4 (OMIM:615982); Bardet-Biedl syndrome (ORPHA:110)
BBS5	Bardet-Biedl syndrome 5 (OMIM:615983); Bardet-Biedl syndrome (ORPHA:110)
BBS7	Bardet-Biedl syndrome 7 (OMIM:615984); Bardet-Biedl syndrome (ORPHA:110)
BBS9	Bardet-Biedl syndrome 9 (OMIM:615986); Bardet-Biedl syndrome (ORPHA:110)
BCAM	OMIM:111200; LUTHERAN NULL (OMIM:247420)
BCAS3	Hengel-Maroofian-Schols syndrome (OMIM:619641); ORPHA:697067
BCAT2	Hypervalinemia or hyperleucine-isoleucinemia (OMIM:618850)
BCHE	Butyrylcholinesterase deficiency (OMIM:617936); Hereditary butyrylcholinesterase deficiency (ORPHA:132)
BCKDHA	Maple syrup urine disease (OMIM:248600); ORPHA:268145; ORPHA:268162; ORPHA:268173
BCKDHB	Maple syrup urine disease, type Ib (OMIM:620698); ORPHA:268145; ORPHA:268162; ORPHA:268173
BCKDK	Branched-chain keto acid dehydrogenase kinase deficiency (OMIM:614923); ORPHA:308410
BCL10	Testicular tumor, somatic (OMIM:273300); Mesothelioma, malignant (OMIM:156240); Lymphoma, non-hodgkin, familial (OMIM:605027); Gastric lymphoma, primary (OMIM:137245); Immunodeficiency 37 (OMIM:616098); MALT lymphoma (ORPHA:52417)
BCL11A	Intellectual developmental disorder with persistence of fetal hemoglobin (OMIM:617101); Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome (ORPHA:251380); ORPHA:619233
BCL11B	Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities (OMIM:618092); Immunodeficiency 49 (OMIM:617237); ORPHA:662829
BCL2	ORPHA:480541; ORPHA:98839; Follicular lymphoma (ORPHA:545)
BCL2L1	—
BCL2L11	—
BCL2L14	—
BCL2L2	—
BCL6	ORPHA:480541; ORPHA:98838; ORPHA:98839; Follicular lymphoma (ORPHA:545)
BCL9	—
BCO1	Carotenemia, familial (OMIM:115300); ORPHA:199285
BCOR	Microphthalmia, syndromic 1 (OMIM:309800); Microphthalmia, syndromic 2 (OMIM:300166); Oculofaciocardiodental syndrome (ORPHA:2712); Microphthalmia, Lenz type (ORPHA:568); Acute promyelocytic leukemia (ORPHA:520); ORPHA:457246
BCORL1	Shukla-Vernon syndrome (OMIM:301029); ORPHA:528084
BCR	Leukemia, acute lymphocytic, susceptibility to, 1 (OMIM:613065); Leukemia, chronic myeloid (OMIM:608232); ORPHA:99861; ORPHA:585909; Distal 22q11.2 microdeletion syndrome (ORPHA:261330); Chronic myeloid leukemia (ORPHA:521)
BCS1L	Gracile syndrome (OMIM:603358); Mitochondrial complex III deficiency, nuclear type 1 (OMIM:124000); Bjornstad syndrome (OMIM:262000); GRACILE syndrome (ORPHA:53693); ORPHA:1460; Björnstad syndrome (ORPHA:123); ORPHA:254902
BDNF	WAGR syndrome (ORPHA:893); Congenital central hypoventilation syndrome (ORPHA:661)
BEAN1	Spinocerebellar ataxia 31 (OMIM:117210); Spinocerebellar ataxia type 31 (ORPHA:217012)
BEST1	Bestrophinopathy, autosomal recessive (OMIM:611809); Macular dystrophy, vitelliform, 2 (OMIM:153700); Vitreoretinochoroidopathy (OMIM:193220); Retinitis pigmentosa-50 (OMIM:613194); Adult-onset foveomacular vitelliform dystrophy (ORPHA:99000); ORPHA:263347; Best vitelliform macular dystrophy (ORPHA:1243); ORPHA:139455; ORPHA:3086; Retinitis pigmentosa (ORPHA:791); Nanophthalmos (ORPHA:35612)
BFSP1	Cataract 33, multiple types (OMIM:611391); ORPHA:98991
BFSP2	Cataract, autosomal dominant, multiple types 1 (OMIM:611597); ORPHA:98984; ORPHA:98985;

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	ORPHA:441452
BGN	Meester-Loeys syndrome (OMIM:300989); Spondyloepimetaphyseal dysplasia, X-linked (OMIM:300106); ORPHA:93349; ORPHA:622925
BHLHA9	Syndactyly, mesoaxial synostotic, with phalangeal reduction (OMIM:609432); Camptosynpolydactyly, complex (OMIM:607539); Tibial aplasia-ectrodactyly syndrome (ORPHA:3329); Gollop-Wolfgang complex (ORPHA:1986); Mesoaxial synostotic syndactyly with phalangeal reduction (ORPHA:157801)
BHLHE41	SHORT SLEEPER (OMIM:612975)
BICC1	Renal dysplasia, cystic, susceptibility to (OMIM:601331); Autosomal dominant polycystic kidney disease (ORPHA:730)
BIN1	Myopathy, centronuclear, 2 (OMIM:255200); Autosomal recessive centronuclear myopathy (ORPHA:169186); Autosomal dominant centronuclear myopathy (ORPHA:169189)
BIVM	—
BLK	Maturity-onset diabetes of the young, type 11 (OMIM:613375); MODY (ORPHA:552); Systemic lupus erythematosus (ORPHA:536)
BLM	Bloom syndrome (OMIM:210900); Bloom syndrome (ORPHA:125)
BLMH	—
BLNK	Agammaglobulinemia 4, autosomal recessive (OMIM:613502); Autosomal non-syndromic agammaglobulinemia (ORPHA:33110)
BLOC1S5	Hermansky-Pudlak syndrome 11 (OMIM:619172); ORPHA:231531
BLOC1S6	Hermansky-Pudlak syndrome 9 (OMIM:614171); ORPHA:231531
BLTP2	—
BLVRA	Hyperbiliverdinemia (OMIM:614156); ORPHA:276405
BMI1	—
BMP1	Osteogenesis imperfecta, type XIII (OMIM:614856); ORPHA:314029; ORPHA:216812
BMP15	Ovarian dysgenesis 2 (OMIM:300510); 46,XX gonadal dysgenesis (ORPHA:243); ORPHA:398987
BMP2	Brachydactyly, type A2 (OMIM:112600); Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1 (OMIM:617877); Hemochromatosis, type 1 (OMIM:235200); 20p12.3 microdeletion syndrome (ORPHA:261295); Brachydactyly type A2 (ORPHA:93396)
BMP4	Orofacial cleft 11 (OMIM:600625); Microphthalmia, syndromic 6 (OMIM:607932); Cleft lip/palate (ORPHA:199306); Microphthalmia with brain and digit anomalies (ORPHA:139471); ORPHA:93100; Stickler syndrome (ORPHA:828)
BMP5	—
BMP7	—
BMPER	Diaphanospondylodysostosis (OMIM:608022); Diaphanospondylodysostosis (ORPHA:66637)
BMPR1A	Juvenile polyposis syndrome (OMIM:174900); Polyposis syndrome, hereditary mixed, 2 (OMIM:610069); Generalized juvenile polyposis/juvenile polyposis coli (ORPHA:329971); Juvenile polyposis of infancy (ORPHA:79076); Hereditary mixed polyposis syndrome (ORPHA:157794); Familial colorectal cancer Type X (ORPHA:440437)
BMPR1B	Brachydactyly, type A2 (OMIM:112600); Acromesomelic dysplasia, Demirhan type (OMIM:609441); Brachydactyly, type A1, D (OMIM:616849); Fibular aplasia-complex brachydactyly syndrome (ORPHA:2639); Brachydactyly type A2 (ORPHA:93396); Brachydactyly type C (ORPHA:93384); Brachydactyly type A1 (ORPHA:93388); Acromesomelic dysplasia, Grebe type (ORPHA:2098)
BMPR2	Pulmonary hypertension, primary, 1 (OMIM:178600); Pulmonary venoocclusive disease 1 (OMIM:265450); ORPHA:275777; ORPHA:275786; ORPHA:31837
BNC2	Lower urinary tract obstruction, congenital (OMIM:618612); Posterior urethral valve (ORPHA:93110)
BOC	—
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia (OMIM:614299); ORPHA:401874
BORCS7	—
BORCS8	Neurodegeneration, infantile-onset, with optic atrophy and brain abnormalities (OMIM:620987)
BPGM	Erythrocytosis, familial, 8 (OMIM:222800); ORPHA:247378; ORPHA:714
BPNT2	Chondrodysplasia with joint dislocations, Gpapp type (OMIM:614078); ORPHA:280586
BRAF	Cardiofaciocutaneous syndrome 1 (OMIM:115150); Colorectal cancer (OMIM:114500); Lung cancer, susceptibility to (OMIM:211980); Leopard syndrome 3 (OMIM:613707); Noonan syndrome 7 (OMIM:613706); Melanoma, cutaneous malignant (OMIM:155600); Noonan syndrome 1 (OMIM:163950); Langerhans cell histiocytosis (ORPHA:389); ORPHA:146; Cushing disease (ORPHA:96253); Cardiofaciocutaneous syndrome (ORPHA:1340); Craniopharyngioma (ORPHA:54595); Syringocystadenoma papilliferum (ORPHA:840); ORPHA:58017; Large/giant congenital melanocytic nevus (ORPHA:626); Noonan syndrome (ORPHA:648); Noonan syndrome with multiple lentigines (ORPHA:500); ORPHA:251615
BRAT1	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures (OMIM:618056); Rigidity and multifocal seizure syndrome, lethal neonatal (OMIM:614498); ORPHA:435845
BRCA1	Fanconi anemia, complementation group S (OMIM:617883); Breast-Ovarian cancer, familial, susceptibility to, 1 (OMIM:604370); OMIM:614320; Breast cancer (OMIM:114480); Fanconi anemia (ORPHA:84); Hereditary breast and/or ovarian cancer syndrome (ORPHA:145); Primary peritoneal carcinoma (ORPHA:168829); ORPHA:694963; ORPHA:227535; Familial pancreatic carcinoma (ORPHA:1333); ORPHA:1331; ORPHA:667662; Cholangiocarcinoma (ORPHA:70567)
BRCA2	Fanconi anemia, complementation group D1 (OMIM:605724); Glioma susceptibility 3 (OMIM:613029); Wilms tumor 1 (OMIM:194070); Breast cancer (OMIM:114480); Pancreatic cancer, susceptibility to, 2 (OMIM:613347); Prostate cancer (OMIM:176807); Breast-Ovarian cancer, familial, susceptibility to, 2 (OMIM:612555); MEDULLOBLASTOMA (OMIM:155255); Fanconi anemia (ORPHA:84); Hereditary breast

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	and/or ovarian cancer syndrome (ORPHA:145); ORPHA:178; ORPHA:694963; ORPHA:227535; Familial pancreatic carcinoma (ORPHA:1333); ORPHA:1331; ORPHA:667662; Cholangiocarcinoma (ORPHA:70567); Nephroblastoma (ORPHA:654); ORPHA:319462; Familial colorectal cancer Type X (ORPHA:440437)
BRCC3	Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome (ORPHA:280679)
BRD1	—
BRD2	—
BRF1	Cerebellofaciodental syndrome (OMIM:616202); Cerebellar-facial-dental syndrome (ORPHA:444072)
BRINP3	—
BRIP1	Fanconi anemia, complementation group J (OMIM:609054); Breast cancer (OMIM:114480); Fanconi anemia (ORPHA:84); Hereditary breast and/or ovarian cancer syndrome (ORPHA:145)
BRME1	—
BRSK2	ORPHA:178469
BRWD1	Ciliary dyskinesia, primary, 51 (OMIM:620438); Primary ciliary dyskinesia (ORPHA:244)
BRWD3	Intellectual developmental disorder, X-linked 93 (OMIM:300659); ORPHA:528084
BSCL2	Lipodystrophy, congenital generalized, type 2 (OMIM:269700); Spastic paraplegia 17 (OMIM:270685); Neuropathy, distal hereditary motor, type VC (OMIM:619112); Encephalopathy, progressive, with or without lipodystrophy (OMIM:615924); Distal hereditary motor neuropathy type 5 (ORPHA:139536); Progressive encephalopathy-severe neurodegeneration-lipodystrophy syndrome (ORPHA:363400); Autosomal dominant spastic paraplegia type 17 (ORPHA:100998); ORPHA:696289
BSG	OMIM:111380
BSND	Bartter syndrome, type 4A, neonatal, with sensorineural deafness (OMIM:602522); ORPHA:90636; Bartter syndrome type 4 (ORPHA:89938)
BTBD18	—
BTBD9	—
BTD	Biotinidase deficiencymultiple carboxylase deficiency, late-onset (OMIM:253260); Biotinidase deficiency (ORPHA:79241)
BTK	Isolated growth hormone deficiency, type III, with agammaglobulinemia (OMIM:307200); Agammaglobulinemia, X-linked (OMIM:300755); X-linked agammaglobulinemia (ORPHA:47); ORPHA:632
BUB1	Microcephaly 30, primary, autosomal recessive (OMIM:620183); Colorectal cancer (OMIM:114500); Mosaic variegated aneuploidy syndrome (ORPHA:1052)
BUB1B	Premature chromatid separation trait (OMIM:176430); Colorectal cancer (OMIM:114500); Mosaic variegated aneuploidy syndrome 1 (OMIM:257300); Mosaic variegated aneuploidy syndrome (ORPHA:1052)
C10orf105	—
C10orf55	—
C11orf65	—
C12orf43	—
C12orf57	Temtamy syndrome (OMIM:218340)
C12orf60	—
C14orf39	Spermatogenic failure 52 (OMIM:619202); Premature ovarian failure 18 (OMIM:619203)
C15orf62	—
C17orf107	—
C17orf67	—
C1GALT1	—
C1GALT1C1	Hemolytic uremic syndrome, atypical, 8, with rhizomelic short stature (OMIM:301110); Tn polyagglutination syndrome (OMIM:300622)
C1orf141	—
C1QA	C1q deficiency (OMIM:613652); ORPHA:300345; ORPHA:169147
C1QB	C1q deficiency 2 (OMIM:620321); ORPHA:169147
C1QC	C1q deficiency 3 (OMIM:620322); ORPHA:169147
C1QTNF3	—
C1R	Ehlers-Danlos syndrome, periodontal type, 1 (OMIM:130080); ORPHA:300345; Periodontal Ehlers-Danlos syndrome (ORPHA:75392); ORPHA:169147
C1RL	—
C1S	Complement component c1s deficiency (OMIM:613783); Ehlers-Danlos syndrome, periodontal type, 2 (OMIM:617174); Periodontal Ehlers-Danlos syndrome (ORPHA:75392); ORPHA:169147
C21orf91	—
C2CD5	—
C2orf42	—
C2orf88	—
C3	Hemolytic uremic syndrome, atypical, susceptibility to, 5 (OMIM:612925); OMIM:611378; Complement component 3 deficiency, autosomal recessive (OMIM:613779); ORPHA:280133; ORPHA:544472
C3AR1	—
C4orf19	—
C5	C5 deficiency (OMIM:609536); OMIM:615749; ORPHA:169150
C5AR2	—
C5orf46	—
C6	C6 deficiency (OMIM:612446); ORPHA:169150
C7	C7 deficiency (OMIM:610102); ORPHA:169150

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C8A	Complement component 8 deficiency, type I (OMIM:613790); ORPHA:169150
C8B	Complement component 8 deficiency, type II (OMIM:613789); ORPHA:169150
C9	Macular degeneration, age-related, 15, susceptibility to (OMIM:615591); Complement component 9 deficiency (OMIM:613825); ORPHA:169150
C9orf72	Amyotrophic lateral sclerosis and/or frontotemporal dementia 1 (OMIM:105550)
CA1	—
CA12	Hyperchlorhidrosis, isolated (OMIM:143860); ORPHA:542657
CA2	Osteopetrosis, autosomal recessive 3 (OMIM:259730); Osteopetrosis with renal tubular acidosis (ORPHA:2785)
CA3	—
CA4	Retinitis pigmentosa (ORPHA:791)
CA8	Spinocerebellar ataxia, autosomal recessive 34 (OMIM:613227); Dysequilibrium syndrome (ORPHA:1766)
CABIN1	—
CABP2	Deafness, autosomal recessive 93 (OMIM:614899); ORPHA:90636
CABP4	Cone-Rod synaptic disorder, congenital nonprogressive (OMIM:610427); Sleep-related hypermotor epilepsy (ORPHA:98784); ORPHA:714070
CACNA1A	Spinocerebellar ataxia 6 (OMIM:183086); Migraine, familial hemiplegic, 1 (OMIM:141500); Episodic ataxia, type 2 (OMIM:108500); Epileptic encephalopathy, early infantile, 42 (OMIM:617106); Familial paroxysmal ataxia (ORPHA:97); Benign paroxysmal torticollis of infancy (ORPHA:71518); Spinocerebellar ataxia type 6 (ORPHA:98758); Lennox-Gastaut syndrome (ORPHA:2382); Non-specific early-onset epileptic encephalopathy (ORPHA:442835); Familial or sporadic hemiplegic migraine (ORPHA:569); Alternating hemiplegia of childhood (ORPHA:2131)
CACNA1C	Timothy syndrome (OMIM:601005); Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures (OMIM:620029); Brugada syndrome 3 (OMIM:611875); Long QT syndrome 8 (OMIM:618447); ORPHA:595098; Romano-Ward syndrome (ORPHA:101016); ORPHA:595105; ORPHA:595109; Brugada syndrome (ORPHA:130); ORPHA:528084
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities (OMIM:615474); Sinoatrial node dysfunction and deafness (OMIM:614896); ORPHA:324321; Primary hyperaldosteronism-seizures-neurological abnormalities syndrome (ORPHA:369929)
CACNA1E	Developmental and epileptic encephalopathy 69 (OMIM:618285); Early infantile developmental and epileptic encephalopathy (ORPHA:1934)
CACNA1F	Night blindness, congenital stationary, type 2A (OMIM:300071); Cone-Rod dystrophy, X-linked, 3 (OMIM:300476); Aland island eye disease (OMIM:300600); Åland Islands eye disease (ORPHA:178333); ORPHA:714070; Cone rod dystrophy (ORPHA:1872)
CACNA1G	Spinocerebellar ataxia 42 (OMIM:616795); Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits (OMIM:618087); Spinocerebellar ataxia type 42 (ORPHA:458803)
CACNA1H	OMIM:611942; Hyperaldosteronism, familial, type IV (OMIM:617027); ORPHA:642671; Childhood absence epilepsy (ORPHA:64280)
CACNA1S	Congenital myopathy 18 due to dihydropyridine receptor defect (OMIM:620246); Hypokalemic periodic paralysis, type 1 (OMIM:170400); Thyrotoxic periodic paralysis, susceptibility to, 1 (OMIM:188580); Malignant hyperthermia, susceptibility to, 5 (OMIM:601887); Thyrotoxic periodic paralysis (ORPHA:79102); Hypokalemic periodic paralysis (ORPHA:681); ORPHA:397755; Malignant hyperthermia of anesthesia (ORPHA:423)
CACNA2D1	Developmental and epileptic encephalopathy 110 (OMIM:620149); Congenital short QT syndrome (ORPHA:51083); Brugada syndrome (ORPHA:130); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
CACNA2D2	Cerebellar atrophy with seizures and variable developmental delay (OMIM:618501)
CACNA2D3	—
CACNA2D4	Retinal cone dystrophy 4 (OMIM:610478); ORPHA:714070; Cone rod dystrophy (ORPHA:1872)
CACNB2	Brugada syndrome 4 (OMIM:611876); Brugada syndrome (ORPHA:130)
CACNB4	Epilepsy, idiopathic generalized, susceptibility to, 9 (OMIM:607682); Episodic ataxia, type 5 (OMIM:613855); Juvenile myoclonic epilepsy (ORPHA:307); Episodic ataxia type 5 (ORPHA:211067)
CACNG2	Intellectual developmental disorder, autosomal dominant 10 (OMIM:614256); ORPHA:178469
CACNG3	—
CACNG4	—
CADM1	—
CADM2	—
CADM3	Charcot-Marie-Tooth disease, axonal, type 2FF (OMIM:619519)
CADPS2	—
CAHM	—
CALCA	—
CALCB	—
CALCR	OSTEOPOROSIS (OMIM:166710)
CALCRL	Lymphatic malformation 8 (OMIM:618773); ORPHA:363999
CALHM1	—
CALM1	Long QT syndrome 14 (OMIM:616247); Ventricular tachycardia, catecholaminergic polymorphic, 4 (OMIM:614916); Romano-Ward syndrome (ORPHA:101016); Catecholaminergic polymorphic ventricular tachycardia (ORPHA:3286)
CALM3	Long QT syndrome 16 (OMIM:618782); Romano-Ward syndrome (ORPHA:101016); Catecholaminergic polymorphic ventricular tachycardia (ORPHA:3286)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
CALR	Myelofibrosis with myeloid metaplasia, somatic (OMIM:254450); Thrombocythemia 1 (OMIM:187950); Essential thrombocythemia (ORPHA:3318); Budd-Chiari syndrome (ORPHA:131); Primary myelofibrosis (ORPHA:824)
CALR3	—
CALU	—
CAMK1	—
CAMK4	—
CAMKMT	ORPHA:238523; 2p21 microdeletion syndrome (ORPHA:163693); ORPHA:369881
CAMSAP2	—
CAMTA1	Cerebellar dysfunction with variable cognitive and behavioral abnormalities (OMIM:614756); Non-progressive cerebellar ataxia with intellectual disability (ORPHA:314647); ORPHA:157791
CANT1	Epiphyseal dysplasia, multiple, 7 (OMIM:617719); Desbuquois dysplasia 1 (OMIM:251450); ORPHA:647676; Desbuquois syndrome (ORPHA:1425)
CAPN10	OMIM:601283
CAPN3	Muscular dystrophy, limb-girdle, type 2A (OMIM:253600); Muscular dystrophy, limb-girdle, autosomal dominant 4 (OMIM:618129); Calpain-3-related limb-girdle muscular dystrophy R1 (ORPHA:267); ORPHA:565909
CARD11	Card11 immunodeficiency (OMIM:615206); B-cell expansion with NFKB and T-cell anergy (OMIM:616452); Immunodeficiency 11B with atopic dermatitis (OMIM:617638); ORPHA:357237; ORPHA:300324; ORPHA:619972; ORPHA:464336
CARD14	Psoriasis 2 (OMIM:602723); Pityriasis rubra pilaris (OMIM:173200); Pityriasis rubra pilaris (ORPHA:2897)
CARD9	Candidiasis, familial chronic mucocutaneous, autosomal recessive (OMIM:212050); ORPHA:457088
CARS2	Combined oxidative phosphorylation deficiency 27 (OMIM:616672); Combined oxidative phosphorylation defect type 27 (ORPHA:477774)
CARTPT	OBESITY (OMIM:601665)
CASC11	—
CASK	Hemolytic anemia, G6PD deficient (favism) (OMIM:300908); Fg syndrome 4 (OMIM:300422); Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia (OMIM:300749); X-linked intellectual disability, Najm type (ORPHA:163937); ORPHA:777; Early infantile developmental and epileptic encephalopathy (ORPHA:1934)
CASP1	—
CASP10	Lymphoma, non-hodgkin, familial (OMIM:605027); Autoimmune lymphoproliferative syndrome, type IIA (OMIM:603909); Gastric cancer, somatic (OMIM:613659); Autoimmune lymphoproliferative syndrome (ORPHA:3261)
CASP12	—
CASP2	Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly (OMIM:620653)
CASP8	Autoimmune lymphoproliferative syndrome, type IIB (OMIM:607271); Breast cancer (OMIM:114480); Hepatocellular carcinoma (OMIM:114550); Lung cancer, susceptibility to (OMIM:211980); ORPHA:210159; ORPHA:275517
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 1, with or without atrial dysfunction and/or dilated cardiomyopathy (OMIM:604772); Ventricular tachycardia, catecholaminergic polymorphic, 2 (OMIM:611938); Catecholaminergic polymorphic ventricular tachycardia (ORPHA:3286)
CASR	Epilepsy idiopathic generalized, susceptibility to, 8 (OMIM:612899); Hyperparathyroidism, neonatal severe (OMIM:239200); Hypocalcemia, autosomal dominant 1 (OMIM:601198); Hypocalciuric hypercalcemia, familial, type I (OMIM:145980); Autosomal dominant hereditary chronic pancreatitis (ORPHA:676); ORPHA:93372; Autosomal dominant hypocalcemia (ORPHA:428); Neonatal severe primary hyperparathyroidism (ORPHA:417)
CAST	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads (OMIM:616295); ORPHA:444138
CAT	Acatalasemia (OMIM:614097); Acatalasemia (ORPHA:926)
CATIP	Spermatogenic failure 54 (OMIM:619379); Male infertility with azoospermia or oligozoospermia due to single gene mutation (ORPHA:399805)
CATSPER1	Spermatogenic failure 7 (OMIM:612997); ORPHA:276234
CATSPER2	Spermatogenic failure 7 (OMIM:612997); Deafness, sensorineural, and male infertility (OMIM:611102); Deafness-infertility syndrome (ORPHA:94064)
CATSPER3	—
CATSPER4	—
CAV1	Pulmonary hypertension, primary, 3 (OMIM:615343); Lipodystrophy, familial partial, type 7 (OMIM:606721); Lipodystrophy, congenital generalized, type 3 (OMIM:612526); Limited cutaneous systemic sclerosis (ORPHA:220402); Diffuse cutaneous systemic sclerosis (ORPHA:220393); ORPHA:275777; ORPHA:696206
CAV3	Creatine phosphokinase, elevated serum (OMIM:123320); Cardiomyopathy, familial hypertrophic 1 (OMIM:192600); Myopathy, distal, Tateyama type (OMIM:614321); Long QT syndrome 9 (OMIM:611818); Rippling muscle disease (OMIM:606072); ORPHA:97238; Romano-Ward syndrome (ORPHA:101016); Distal myopathy, Tateyama type (ORPHA:488650); Isolated asymptomatic elevation of creatine phosphokinase (ORPHA:206599)
CAVIN1	Lipodystrophy, congenital generalized, type 4 (OMIM:613327); ORPHA:228429
CAVIN4	—
CBFA2T2	—
CBFA2T3	ORPHA:329469
CBFB	Cleidocranial dysplasia 2 (OMIM:620099); ORPHA:98829

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia (OMIM:613563); Juvenile myelomonocytic leukemia (OMIM:607785); ORPHA:363972; Aggressive systemic mastocytosis (ORPHA:98850); ORPHA:86834; Noonan syndrome (ORPHA:648)
CBLB	Autoimmune disease, multisystem, infantile-onset, 3 (OMIM:620430)
CBLIF	Intrinsic factor deficiency (OMIM:261000); Congenital intrinsic factor deficiency (ORPHA:332)
CBR1	—
CBR3	—
CBR4	—
CBS	Homocystinuria due to cystathionine beta-synthase deficiency (OMIM:236200); Homocystinuria due to cystathionine beta-synthase deficiency (ORPHA:394)
CBX2	46XY sex reversal 5 (OMIM:613080); 46,XY complete gonadal dysgenesis (ORPHA:242)
CBX4	—
CBY1	Isolated Joubert syndrome (ORPHA:475)
CC2D1A	Intellectual developmental disorder, autosomal recessive 3 (OMIM:608443); ORPHA:88616
CC2D2A	Coach syndrome 2 (OMIM:619111); Retinitis pigmentosa 93 (OMIM:619845); Joubert syndrome 9 (OMIM:612285); Meckel syndrome, type 6 (OMIM:612284); Joubert syndrome with hepatic defect (ORPHA:1454); Retinitis pigmentosa (ORPHA:791); Joubert syndrome with oculorenal defect (ORPHA:2318); Meckel syndrome (ORPHA:564)
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome (OMIM:235510); Hennekam syndrome (ORPHA:2136)
CCDC103	—
CCDC107	—
CCDC12	—
CCDC127	—
CCDC14	—
CCDC140	—
CCDC168	—
CCDC183	—
CCDC22	Ritscher-Schinzel syndrome 2 (OMIM:300963); 3C syndrome (ORPHA:7)
CCDC28B	Bardet-Biedl syndrome 1 (OMIM:209900)
CCDC33	—
CCDC39	Ciliary dyskinesia, primary, 14 (OMIM:613807); Primary ciliary dyskinesia (ORPHA:244)
CCDC40	Ciliary dyskinesia, primary, 15 (OMIM:613808); Primary ciliary dyskinesia (ORPHA:244)
CCDC50	Deafness, autosomal dominant 44 (OMIM:607453); ORPHA:90635
CCDC66	—
CCDC78	Myopathy, centronuclear, 4 (OMIM:614807); ORPHA:319160
CCDC8	3-M syndrome 3 (OMIM:614205); 3M syndrome (ORPHA:2616)
CCDC88C	Hydrocephalus, nonsyndromic, autosomal recessive 1 (OMIM:236600); Spinocerebellar ataxia 40 (OMIM:616053); Spinocerebellar ataxia type 40 (ORPHA:423275); ORPHA:269510
CCDST	—
CCKAR	—
CCKBR	—
CCL11	OMIM:609423; Asthma, susceptibility to (OMIM:600807)
CCL2	OMIM:609423
CCL3	OMIM:609423
CCL5	OMIM:609423
CCM2	Cerebral cavernous malformations 2 (OMIM:603284); Cerebral cavernous malformations 1 (OMIM:116860); Familial cerebral cavernous malformation (ORPHA:221061)
CCN1	—
CCN2	Spondyloepimetaphyseal dysplasia, Li-Shao-Li type (OMIM:621099); Kyphomelic dysplasia (OMIM:211350); Limited cutaneous systemic sclerosis (ORPHA:220402); Diffuse cutaneous systemic sclerosis (ORPHA:220393)
CCN6	Arthropathy, progressive pseudorheumatoid, of childhood (OMIM:208230); Progressive pseudorheumatoid dysplasia (ORPHA:1159)
CCND1	Multiple myeloma (OMIM:254500); Colorectal cancer (OMIM:114500); von Hippel-Lindau syndrome (OMIM:193300); Mantle cell lymphoma (ORPHA:52416); Von Hippel-Lindau disease (ORPHA:892); ORPHA:67038; Multiple myeloma (ORPHA:29073)
CCNH	—
CCNQ	STAR syndrome (OMIM:300707); Syndactyly-telecanthus-anogenital and renal malformations syndrome (ORPHA:140952)
CCPG1	—
CCR2	OMIM:609423; Cystic disease of lung (OMIM:219600)
CCR3	—
CCR4	—
CCR5	OMIM:610379; OMIM:609423; OMIM:609532; OMIM:612522
CCR5AS	—
CCR7	—
CCT5	Neuropathy, hereditary sensory, with spastic paraplegia, autosomal recessive (OMIM:256840); Mutilating hereditary sensory neuropathy with spastic paraplegia (ORPHA:139578)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
CD109	Fetal and neonatal alloimmune thrombocytopenia (ORPHA:853)
CD151	OMIM:179620; Nephropathy with pretibial epidermolysis bullosa and deafness (OMIM:609057); ORPHA:300333
CD177	—
CD19	Immunodeficiency, common variable, 3 (OMIM:613493); Immunodeficiency, common variable, 2 (OMIM:240500); ORPHA:696881
CD200	—
CD207	OMIM:613393
CD209	OMIM:609423; OMIM:614371; OMIM:607948
CD244	Rheumatoid arthritis (OMIM:180300)
CD247	Immunodeficiency 25 (OMIM:610163); Oligoarticular juvenile idiopathic arthritis (ORPHA:85410); Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis (ORPHA:85408); T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta (ORPHA:169160)
CD27	Lymphoproliferative syndrome 2 (OMIM:615122); ORPHA:238505
CD28	Immunodeficiency 123 with HPV-related verrucosis (OMIM:620901); Sézary syndrome (ORPHA:3162); Classic mycosis fungoides (ORPHA:2584)
CD2AP	Focal segmental glomerulosclerosis 3, susceptibility to (OMIM:607832); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect (OMIM:613646); ORPHA:280183
CD36	Platelet glycoprotein IV deficiency (OMIM:608404); OMIM:610938; OMIM:611162
CD38	—
CD3D	Immunodeficiency 19 (OMIM:615617); T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta (ORPHA:169160)
CD3E	Immunodeficiency 18 (OMIM:615615); T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta (ORPHA:169160)
CD3G	Immunodeficiency 17 (OMIM:615607); ORPHA:169082
CD4	Immunodeficiency 79 (OMIM:619238); Okt4 epitope deficiency (OMIM:613949)
CD40	Immunodeficiency with hyper-igm, type 3 (OMIM:606843); ORPHA:101090
CD40LG	Immunodeficiency, X-linked, with hyper-IgM (OMIM:308230); ORPHA:101088
CD44	OMIM:609027
CD46	Hemolytic uremic syndrome, atypical, susceptibility to, 2 (OMIM:612922); HELLP syndrome (ORPHA:244242); ORPHA:544472
CD5	—
CD55	Blood group, cromer system (OMIM:613793); Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy (OMIM:226300); ORPHA:566175
CD58	—
CD59	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy (OMIM:612300); ORPHA:169464
CD74	—
CD79A	Agammaglobulinemia 3, autosomal recessive (OMIM:613501); Autosomal non-syndromic agammaglobulinemia (ORPHA:33110)
CD79B	Agammaglobulinemia 6, autosomal recessive (OMIM:612692); Autosomal non-syndromic agammaglobulinemia (ORPHA:33110)
CD80	—
CD81	Immunodeficiency, common variable, 6 (OMIM:613496); ORPHA:696881
CD86	—
CD8A	Cd8 deficiency, familial (OMIM:608957); ORPHA:169085
CD96	C syndrome (OMIM:211750); C syndrome (ORPHA:1308)
CDA	—
CDAN1	Anemia, congenital dyserythropoietic, type Ia (OMIM:224120); ORPHA:98869
CDC20	Oocyte/zygote/embryo maturation arrest 14 (OMIM:620276)
CDC25C	—
CDC26	—
CDC42	Takenouchi-Kosaki syndrome (OMIM:616737); ORPHA:619363; Takenouchi-Kosaki syndrome (ORPHA:487796)
CDC42BPB	Chilton-Okur-Chung neurodevelopmental syndrome (OMIM:619841)
CDC42SE1	—
CDC6	Meier-Gorlin syndrome 5 (OMIM:613805); Ear-patella-short stature syndrome (ORPHA:2554)
CDC73	Hyperparathyroidism 1 (OMIM:145000); Hyperparathyroidism 2 (OMIM:145001); Parathyroid carcinoma (OMIM:608266); Hyperparathyroidism-jaw tumor syndrome (ORPHA:99880); Familial isolated hyperparathyroidism (ORPHA:99879); Parathyroid carcinoma (ORPHA:143)
CDCA3	—
CDCA7L	—
CDH1	Endometrial carcinoma, somatic (OMIM:608089); Breast cancer (OMIM:114480); Ovarian cancer (OMIM:167000); Gastric cancer, hereditary diffuse (OMIM:137215); Blepharocheilodontic syndrome 1 (OMIM:119580); Cleft lip/palate (ORPHA:199306); ORPHA:26106; ORPHA:227535; ORPHA:1331; Blepharo-cheilo-odontic syndrome (ORPHA:1997)
CDH13	—
CDH15	Intellectual developmental disorder, autosomal dominant 3 (OMIM:612580); ORPHA:178469

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
CDH23	Pituitary adenoma 5, multiple types (OMIM:617540); Usher syndrome, type ID (OMIM:601067); Deafness, autosomal recessive 12 (OMIM:601386); ORPHA:90636; Cushing disease (ORPHA:96253); TSH-secreting pituitary adenoma (ORPHA:91347); Prolactinoma (ORPHA:2965); ORPHA:314777; Usher syndrome type 1 (ORPHA:231169)
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy syndrome (OMIM:225280); Hypotrichosis, congenital, with juvenile macular dystrophy (OMIM:601553); Hypotrichosis with juvenile macular degeneration (ORPHA:1573); EEM syndrome (ORPHA:1897)
CDH5	—
CDHR1	Cone-rod dystrophy 15 (OMIM:613660); Cone rod dystrophy (ORPHA:1872); Retinitis pigmentosa (ORPHA:791)
CDK16	—
CDK4	Melanoma, cutaneous malignant, susceptibility to, 3 (OMIM:609048); Microcephaly 31, primary, autosomal recessive (OMIM:621507); ORPHA:99970; Well-differentiated liposarcoma (ORPHA:99971); Familial melanoma (ORPHA:618)
CDK5R1	—
CDK5RAP2	Microcephaly 3, primary, autosomal recessive (OMIM:604804); Autosomal recessive primary microcephaly (ORPHA:2512)
CDK6	Microcephaly 12, primary, autosomal recessive (OMIM:616080); Autosomal recessive primary microcephaly (ORPHA:2512)
CDK7	—
CDKAL1	—
CDKL3	—
CDKL5	Developmental and epileptic encephalopathy 2 (OMIM:300672); CDKL5-deficiency disorder (ORPHA:505652); Atypical Rett syndrome (ORPHA:3095); Infantile epileptic spasms syndrome (ORPHA:697160); Early infantile developmental and epileptic encephalopathy (ORPHA:1934)
CDKN1A	—
CDKN1B	Multiple endocrine neoplasia, type IV (OMIM:610755); Multiple endocrine neoplasia type 4 (ORPHA:276152); Multiple endocrine neoplasia type 1 (ORPHA:652)
CDKN1C	Beckwith-Wiedemann syndrome (OMIM:130650); Intrauterine growth retardation, metaphyseal dysplasia, adrenal hypoplasiacongenita, and genital anomalies (OMIM:614732); ORPHA:231120; Silver-Russell syndrome due to a point mutation (ORPHA:397590); Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome (ORPHA:436144); IMAGe syndrome (ORPHA:85173)
CDKN2A	Melanoma-Astrocytoma syndrome (OMIM:155755); Melanoma-Pancreatic cancer syndrome (OMIM:606719); Melanoma, cutaneous malignant, susceptibility to, 2 (OMIM:155601); ORPHA:99861; Adrenocortical carcinoma (ORPHA:1501); ORPHA:585909; Familial pancreatic carcinoma (ORPHA:1333); ORPHA:404560; Familial melanoma (ORPHA:618); ORPHA:252206; Li-Fraumeni syndrome (ORPHA:524)
CDKN2B	Familial melanoma (ORPHA:618); Multiple endocrine neoplasia type 1 (ORPHA:652)
CDKN2C	—
CDON	Holoprosencephaly 11 (OMIM:614226); Septopreoptic holoprosencephaly (ORPHA:280195); Pituitary stalk interruption syndrome (ORPHA:95496); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925)
CDT1	Meier-Gorlin syndrome 4 (OMIM:613804); Ear-patella-short stature syndrome (ORPHA:2554)
CDX2	—
CDYL	—
CEACAM16	Deafness, autosomal dominant 4B (OMIM:614614); Deafness, autosomal recessive 113 (OMIM:618410); ORPHA:90635; ORPHA:90636
CEBPA	Leukemia, acute myeloid (OMIM:601626); ORPHA:102724; ORPHA:319480; ORPHA:319465
CEBPE	Immunodeficiency 108 with autoinflammation (OMIM:260570); Specific granule deficiency (OMIM:245480); ORPHA:169142; ORPHA:566067
CECR2	—
CEL	Maturity-Onset diabetes of the young, type 8, with exocrine dysfunction (OMIM:609812); MODY (ORPHA:552)
CELA2A	Abdominal obesity-metabolic syndrome 4 (OMIM:618620)
CELA2B	—
CELSR1	Yellow nail syndrome (OMIM:153300); Lymphatic malformation 9 (OMIM:619319); ORPHA:569816
CELSR2	—
CEMIP	—
CENPJ	—
CENPO	—
CENPP	ORPHA:90635
CEP128	—
CEP135	Microcephaly 8, primary, autosomal recessive (OMIM:614673); Autosomal recessive primary microcephaly (ORPHA:2512)
CEP152	Seckel syndrome 5 (OMIM:613823); Microcephaly 9, primary, autosomal recessive (OMIM:614852); Autosomal recessive primary microcephaly (ORPHA:2512); Seckel syndrome (ORPHA:808)
CEP164	Nephronophthisis 15 (OMIM:614845); Senior-Loken syndrome (ORPHA:3156)
CEP290	Leber congenital amaurosis 10 (OMIM:611755); Joubert syndrome 5 (OMIM:610188); Senior-Loken syndrome 6 (OMIM:610189); Bardet-Biedl syndrome 14 (OMIM:615991); Meckel syndrome, type 4 (OMIM:611134); Senior-Loken syndrome (ORPHA:3156); Leber congenital amaurosis (ORPHA:65); Bardet-Biedl syndrome

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	(ORPHA:110); Joubert syndrome with oculorenal defect (ORPHA:2318); Meckel syndrome (ORPHA:564)
CEP41	Joubert syndrome 15 (OMIM:614464); Joubert syndrome with ocular defect (ORPHA:220493); Isolated Joubert syndrome (ORPHA:475)
CEP57	Mosaic variegated aneuploidy syndrome 2 (OMIM:614114); Mosaic variegated aneuploidy syndrome (ORPHA:1052)
CEP63	Seckel syndrome 6 (OMIM:614728); Seckel syndrome (ORPHA:808)
CEP72	—
CEP85L	Lissencephaly 10 (OMIM:618873); Posterior-predominant lissencephaly-broad flat pons and medulla-midline crossing defects syndrome (ORPHA:572013)
CERKL	Retinitis pigmentosa 26 (OMIM:608380); Retinitis pigmentosa (ORPHA:791)
CERNA1	—
CERS1	Epilepsy, progressive myoclonic, 8 (OMIM:616230); ORPHA:424027
CERS6	—
CERT1	Neurodevelopmental disorder with hypotonia, speech delay, and dysmorphic facies (OMIM:616351); ORPHA:528084
CES1	Drug metabolism, altered, CES1-related (OMIM:618057)
CES2	—
CETP	Hyperalphalipoproteinemia 1 (OMIM:143470); ORPHA:181428
CFAP119	—
CFAP20DC	—
CFAP418	Cone-Rod dystrophy 16 (OMIM:614500); Retinitis pigmentosa (OMIM:268000); Bardet-Biedl syndrome 21 (OMIM:617406); Bardet-Biedl syndrome (ORPHA:110); Cone rod dystrophy (ORPHA:1872); Retinitis pigmentosa (ORPHA:791)
CFAP53	Heterotaxy, visceral, 6, autosomal (OMIM:614779); ORPHA:101063; ORPHA:157769
CFAP57	Spermatogenic failure 95 (OMIM:620917)
CFAP92	—
CFD	Complement factor D deficiency (OMIM:613912); ORPHA:169467
CFH	Complement factor H deficiency (OMIM:609814); Macular degeneration, age-related, 4 (OMIM:610698); Hemolytic uremic syndrome, atypical, susceptibility to, 1 (OMIM:235400); Basal laminar drusen (OMIM:126700); ORPHA:329903; ORPHA:244275; HELLP syndrome (ORPHA:244242); Familial drusen (ORPHA:75376); ORPHA:544472; ORPHA:200421; ORPHA:93571
CFHR1	Macular degeneration, age-related, 1 (OMIM:603075); Hemolytic uremic syndrome, atypical, susceptibility to, 1 (OMIM:235400); ORPHA:329931; ORPHA:93571
CFHR5	Nephropathy due to CFHR5 deficiency (OMIM:614809); ORPHA:329931
CFI	Hemolytic uremic syndrome, atypical, susceptibility to, 3 (OMIM:612923); Macular degeneration, age-related, 13 (OMIM:615439); Complement factor I deficiency (OMIM:610984); ORPHA:244275; HELLP syndrome (ORPHA:244242); Familial drusen (ORPHA:75376); ORPHA:544472; ORPHA:200418
CFL1	—
CFL2	Nemaline myopathy 7 (OMIM:610687); Typical nemaline myopathy (ORPHA:171436)
CFP	Properdin deficiency, X-linked (OMIM:312060); ORPHA:2966
CFTR	Pancreatitis, hereditary (OMIM:167800); Cystic fibrosis (OMIM:219700); Vas deferens, congenital bilateral aplasia of (OMIM:277180); Bronchiectasis with or without elevated sweat chloride 1 (OMIM:211400); Congenital bilateral absence of vas deferens (ORPHA:48); Male infertility with azoospermia or oligozoospermia due to single gene mutation (ORPHA:399805); Idiopathic bronchiectasis (ORPHA:60033); ORPHA:700124; Aquagenic palmoplantar keratoderma (ORPHA:498359); Cystic fibrosis (ORPHA:586)
CGB3	—
CGB5	—
CGREF1	—
CHAT	Myasthenic syndrome, congenital, 6, presynaptic (OMIM:254210); Presynaptic congenital myasthenic syndromes (ORPHA:98914)
CHD1L	—
CHD2	Develomental and epileptic encephalopathy 94 (OMIM:615369); Lennox-Gastaut syndrome (ORPHA:2382); Epilepsy with myoclonic-atonic seizures (ORPHA:1942)
CHD3	Snijders Blok-Campeau syndrome (OMIM:618205); ORPHA:599082
CHD6	Hallermann-Streiff syndrome (ORPHA:2108)
CHD7	Charge syndrome (OMIM:214800); Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Hypogonadotropic hypogonadism 5 with or without anosmia (OMIM:612370); CHARGE syndrome (ORPHA:138); Omenn syndrome (ORPHA:39041); Kallmann syndrome (ORPHA:478); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432)
CHD8	Intellectual developmental disorder with autism and macrocephaly (OMIM:615032); ORPHA:642675; 14q11.2 microduplication syndrome (ORPHA:261229)
CHEK2	Li-Fraumeni syndrome 2 (OMIM:609265); Osteosarcoma (OMIM:259500); Prostate cancer (OMIM:176807); Hereditary breast and/or ovarian cancer syndrome (ORPHA:145); ORPHA:1331; Osteosarcoma (ORPHA:668); Li-Fraumeni syndrome (ORPHA:524); Familial colorectal cancer Type X (ORPHA:440437)
CHGA	—
CHI3L1	OMIM:611960; Schizophrenia (OMIM:181500)
CHIA	—
CHIC2	Leukemia, acute myeloid (OMIM:601626)
CHIT1	Chitotriosidase deficiency (OMIM:614122)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
CHKB	Muscular dystrophy, congenital, megaconial type (OMIM:602541); ORPHA:280671; ORPHA:521305
CHL1	—
CHM	Choroideremia (OMIM:303100); Choroideremia (ORPHA:180)
CHMP1A	Pontocerebellar hypoplasia, type 8 (OMIM:614961); ORPHA:324569
CHMP2B	Frontotemporal dementia and/or amytrophic lateral sclerosis 7 (OMIM:600795); Behavioral variant of frontotemporal dementia (ORPHA:275864); Amyotrophic lateral sclerosis (ORPHA:803); Progressive non-fluent aphasia (ORPHA:100070); Semantic dementia (ORPHA:100069)
CHMP4B	Cataract 31, multiple types (OMIM:605387); ORPHA:441447; ORPHA:98993
CHN1	Duane retraction syndrome 2 (OMIM:604356); Duane retraction syndrome (ORPHA:233)
CHPF	—
CHPF2	—
CHPT1	—
CHRD	—
CHRDL1	Megalocornea 1, X-linked (OMIM:309300); ORPHA:91489
CHRM1	—
CHRM2	—
CHRM3	Prune belly syndrome (OMIM:100100); Prune belly syndrome (ORPHA:2970)
CHRNA1	Myasthenic syndrome, congenital, 1B, fast-channel (OMIM:608930); Myasthenic syndrome, congenital, 1A, slow-channel (OMIM:601462); Multiple pterygium syndrome, Lethal type (OMIM:253290); Postsynaptic congenital myasthenic syndrome (ORPHA:98913); ORPHA:33108
CHRNA2	Epilepsy, nocturnal frontal lobe, 4 (OMIM:610353); Sleep-related hypermotor epilepsy (ORPHA:98784)
CHRNA3	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT (OMIM:191800); OMIM:612052
CHRNA4	OMIM:188890; Epilepsy, nocturnal frontal lobe, type 1 (OMIM:600513); Sleep-related hypermotor epilepsy (ORPHA:98784)
CHRNA5	OMIM:612052
CHRNA7	Chromosome 15q13.3 microdeletion syndrome (OMIM:612001); 15q13.3 microdeletion syndrome (ORPHA:199318)
CHRNA9	—
CHRNB1	Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency (OMIM:616314); Myasthenic syndrome, congenital, 2A, slow-channel (OMIM:616313); Postsynaptic congenital myasthenic syndrome (ORPHA:98913)
CHRNB2	Epilepsy, nocturnal frontal lobe, 3 (OMIM:605375); Sleep-related hypermotor epilepsy (ORPHA:98784)
CHRNB4	—
CHRND	Myasthenic syndrome, congenital, 3B, fast-channel (OMIM:616322); Myasthenic syndrome, congenital, 3A, slow-channel (OMIM:616321); Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency (OMIM:616323); Multiple pterygium syndrome, Lethal type (OMIM:253290); Postsynaptic congenital myasthenic syndrome (ORPHA:98913); ORPHA:33108
CHRNE	Myasthenic syndrome, congenital, 4A, slow-channel (OMIM:605809); Myasthenic syndrome, congenital, 4B, fast-channel (OMIM:616324); Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency (OMIM:608931); Postsynaptic congenital myasthenic syndrome (ORPHA:98913)
CHRNG	Escobar syndrome (OMIM:265000); Multiple pterygium syndrome, Lethal type (OMIM:253290); Autosomal recessive multiple pterygium syndrome (ORPHA:2990); ORPHA:33108
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1 (OMIM:601776); Musculocontractural Ehlers-Danlos syndrome (ORPHA:2953)
CHST3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects (OMIM:245600); Spondyloepiphyseal dysplasia with congenital joint dislocations (OMIM:143095); CHST3-related skeletal dysplasia (ORPHA:263463)
CHST6	Macular dystrophy, corneal, 1 (OMIM:217800); Macular corneal dystrophy (ORPHA:98969)
CHST7	—
CHST8	ORPHA:263548
CHSY1	Temtamy preaxial brachydactyly syndrome (OMIM:605282); Temtamy preaxial brachydactyly syndrome (ORPHA:363417)
CHSY3	—
CHUK	Popliteal pterygium syndrome, Bartsocas-Papas type 2 (OMIM:619339); Fetal encasement syndrome (OMIM:613630); ORPHA:465824; ORPHA:697403
CHURC1	—
CIB2	Deafness, autosomal recessive 48 (OMIM:609439); ORPHA:90636; Usher syndrome type 1 (ORPHA:231169)
CIC	Intellectual developmental disorder, autosomal dominant 45 (OMIM:617600); ORPHA:178469
CIDEC	Lipodystrophy, familial partial, type 5 (OMIM:615238); CIDEC-related familial partial lipodystrophy (ORPHA:435651)
CIITA	Rheumatoid arthritis (OMIM:180300); Bare lymphocyte syndrome, type II (OMIM:209920); Immunodeficiency by defective expression of MHC class II (ORPHA:572)
CILK1	ENDOCRINE-CEREBROOSTEODYSPLASIA (OMIM:612651); Cranioectodermal dysplasia 6 (OMIM:621337); Epilepsy, juvenile myoclonic, susceptibility to, 10 (OMIM:617924); ORPHA:199332; Juvenile myoclonic epilepsy (ORPHA:307)
CILP	OMIM:603932
CIMAP1B	—
CISD2	Wolfram syndrome 2 (OMIM:604928); Wolfram syndrome (ORPHA:3463)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
CISH	OMIM:614383; OMIM:607948; OMIM:611162
CITED2	Atrial septal defect 8 (OMIM:614433); Ventricular septal defect 2 (OMIM:614431); ORPHA:101063; Atrial septal defect, sinus venosus type (ORPHA:99105); Atrial septal defect, ostium secundum type (ORPHA:99103); Tetralogy of Fallot (ORPHA:3303)
CIZ1	Adult-onset cervical dystonia, DYT23 type (ORPHA:420492)
CKM	—
CKMT1B	—
CLCA1	—
CLCC1	Retinitis pigmentosa 32 (OMIM:609913)
CLCF1	Cold-Induced sweating syndrome 2 (OMIM:610313); Crisponi syndrome (ORPHA:1545); ORPHA:157820
CLCN1	Myopathy, congenital (OMIM:255300); Myotonia congenita, autosomal recessive (OMIM:255700); Myotonia congenita, autosomal dominant (OMIM:160800); Thomsen and Becker disease (ORPHA:614)
CLCN2	Hyperaldosteronism, familial, type II (OMIM:605635); Leukoencephalopathy with ataxia (OMIM:615651); Epilepsy with grand mal seizures on awakening (OMIM:607628); Juvenile myoclonic epilepsy (ORPHA:307); ORPHA:363540; Familial hyperaldosteronism type II (ORPHA:404)
CLCN3	Neurodevelopmental disorder with hypotonia and brain abnormalities (OMIM:619512); Neurodevelopmental disorder with seizures and brain abnormalities (OMIM:619517); ORPHA:528084
CLCN4	Raynaud-Claes syndrome (OMIM:300114); ORPHA:777; CLCN4-related X-linked intellectual disability syndrome (ORPHA:485350)
CLCN5	Nephrolithiasis, type I (OMIM:310468); Dent disease 1 (OMIM:300009); Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis (OMIM:308990); Hypophosphatemic rickets, X-linked recessive (OMIM:300554); ORPHA:93622
CLCN6	Neurodegeneration, childhood-onset, hypotonia, respiratory insufficiency and brain imaging abnormalities (OMIM:619173); ORPHA:610573
CLCN7	Osteopetrosis, autosomal recessive 4 (OMIM:611490); Osteopetrosis, autosomal dominant 2 (OMIM:166600); Hypopigmentation, organomegaly, and delayed myelination and development (OMIM:618541); Albers-Schönberg osteopetrosis (ORPHA:53); Intermediate osteopetrosis (ORPHA:210110); Autosomal recessive malignant osteopetrosis (ORPHA:667)
CLCNKA	Bartter syndrome, type 4B, neonatal, with sensorineural deafness (OMIM:613090); Bartter syndrome type 4 (ORPHA:89938)
CLCNKB	Bartter syndrome, type 4B, neonatal, with sensorineural deafness (OMIM:613090); Bartter syndrome, type 3 (OMIM:607364); Gitelman syndrome (ORPHA:358); Bartter syndrome type 4 (ORPHA:89938); ORPHA:93605
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis (OMIM:607626); Neonatal ichthyosis-sclerosing cholangitis syndrome (ORPHA:59303)
CLDN14	Deafness, autosomal recessive 29 (OMIM:614035); ORPHA:90636
CLDN16	Hypomagnesemia 3, renal (OMIM:248250); ORPHA:31043
CLDN19	Hypomagnesemia 5, renal, with or without ocular involvement (OMIM:248190); Primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement (ORPHA:2196)
CLEC10A	—
CLEC3B	Macular dystrophy, retinal, 4 (OMIM:619977)
CLEC7A	OMIM:614079; Candidiasis, familial, 4 (OMIM:613108); Chronic mucocutaneous candidiasis (ORPHA:1334)
CLIC2	OMIM:300815; X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome (ORPHA:324410)
CLIP2	Williams syndrome (ORPHA:904)
CLIP4	—
CLK2	—
CLMP	Congenital short bowel syndrome (OMIM:615237); Congenital short bowel syndrome (ORPHA:2301)
CLN3	Retinitis pigmentosa 101 (OMIM:621548); Ceroid lipofuscinosis, neuronal, 3 (OMIM:204200); ORPHA:699796; ORPHA:699780
CLN5	Ceroid lipofuscinosis, neuronal, 5 (OMIM:256731); ORPHA:699812; ORPHA:699807; ORPHA:699802
CLN6	Ceroid lipofuscinosis, neuronal, 6B (Kufs type) (OMIM:204300); Ceroid lipofuscinosis, neuronal, 6 (OMIM:601780); ORPHA:700467; ORPHA:700477; ORPHA:700472
CLN8	Ceroid lipofuscinosis, neuronal, 8, northern epilepsy variant (OMIM:610003); Ceroid lipofuscinosis, neuronal, 8 (OMIM:600143); ORPHA:700484; Northern epilepsy (ORPHA:1947)
CLOCK	—
CLPTM1	—
CLPTM1L	—
CLRN1	Usher syndrome, type IIIA (OMIM:276902); Retinitis pigmentosa 61 (OMIM:614180); Retinitis pigmentosa (OMIM:268000); Retinitis pigmentosa (ORPHA:791); Usher syndrome type 3 (ORPHA:231183)
CLTA	—
CLTCL1	Congenital insensitivity to pain with severe intellectual disability (ORPHA:453510)
CLU	—
CMKLR2	—
CMPK1	—
CNBP	Dystrophia myotonica 2 (OMIM:602668); Proximal myotonic myopathy (ORPHA:606)
CNGA1	Retinitis pigmentosa (OMIM:268000); Retinitis pigmentosa 49 (OMIM:613756); Retinitis pigmentosa (ORPHA:791)
CNGA3	Achromatopsia 2 (OMIM:216900); Cone rod dystrophy (ORPHA:1872); Achromatopsia (ORPHA:49382)
CNGB1	Retinitis pigmentosa 45 (OMIM:613767); Retinitis pigmentosa (ORPHA:791)

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CNGB3	Achromatopsia 3 (OMIM:262300); Progressive cone dystrophy (ORPHA:1871); Stargardt disease (ORPHA:827); Achromatopsia (ORPHA:49382)
CNKSR1	—
CNKSR2	Intellectual developmental disorder, X-linked syndromic, Houge type (OMIM:301008); ORPHA:777; Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
CNN3	—
CNNM2	Hypomagnesemia, seizures, and impaired intellectual development 1 (OMIM:616418); Hypomagnesemia 6, renal (OMIM:613882); ORPHA:620363
CNNM4	Jalili syndrome (OMIM:217080); Jalili syndrome (ORPHA:1873)
CNOT3	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies (OMIM:618672); ORPHA:99861; ORPHA:528084
CNOT4	—
CNPY3	Infantile epileptic spasms syndrome (ORPHA:697160)
CNR1	—
CNR2	—
CNTF	—
CNTN1	Myopathy, congenital, compton-north (OMIM:612540); ORPHA:210163
CNTN4	—
CNTNAP2	Pitt-Hopkins like syndrome 1 (OMIM:610042); OMIM:612100; CNTNAP2-related developmental and epileptic encephalopathy (ORPHA:163681)
CNTNAP4	—
CNTRL	—
COA3	Mitochondrial complex IV deficiency, nuclear type 14 (OMIM:619058); ORPHA:254905
COA5	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3 (OMIM:616500); ORPHA:1561
COCH	Deafness, autosomal dominant 9 (OMIM:601369); Deafness, autosomal recessive 110 (OMIM:618094); ORPHA:90635
COG1	Congenital disorder of glycosylation, type IIg (OMIM:611209); COG1-CDG (ORPHA:263508)
COG2	Congenital disorder of glycosylation, type IIq (OMIM:617395); COG2-CDG (ORPHA:435934)
COG3	Congenital disorder of glycosylation, type IIbb (OMIM:620546)
COG4	Congenital disorder of glycosylation, type IIj (OMIM:613489); Saul-Wilson syndrome (OMIM:618150); COG4-CDG (ORPHA:263501); Microcephalic osteodysplastic dysplasia, Saul-Wilson type (ORPHA:85172)
COG5	Congenital disorder of glycosylation, type IIi (OMIM:613612); COG5-CDG (ORPHA:263487)
COG6	Shaheen syndrome (OMIM:615328); Congenital disorder of glycosylation, type III (OMIM:614576); ORPHA:464443; Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome (ORPHA:363523)
COG7	Congenital disorder of glycosylation, type IIe (OMIM:608779); COG7-CDG (ORPHA:79333)
COG8	Congenital disorder of glycosylation, type IIh (OMIM:611182); COG8-CDG (ORPHA:95428)
COL10A1	Metaphyseal chondrodysplasia, Schmid type (OMIM:156500); Metaphyseal chondrodysplasia, Schmid type (ORPHA:174)
COL11A1	Marshall syndrome (OMIM:154780); Stickler syndrome, type II (OMIM:604841); OMIM:603932; Fibrochondrogenesis 1 (OMIM:228520); Deafness, autosomal dominant 37 (OMIM:618533); Stickler syndrome type 2 (ORPHA:90654); ORPHA:90635; Fibrochondrogenesis (ORPHA:2021); Autosomal dominant myopia-midfacial retrusion-sensorineural hearing loss-rhizomelic dysplasia syndrome (ORPHA:440354); Marshall syndrome (ORPHA:560)
COL11A2	Deafness, autosomal recessive 53 (OMIM:609706); Stickler syndrome, type III (OMIM:184840); Fibrochondrogenesis 2 (OMIM:614524); Otospondylomegaepiphyseal dysplasia (OMIM:215150); Deafness, autosomal dominant nonsyndromic sensorineural 13 (OMIM:601868); ORPHA:90635; ORPHA:90636; Autosomal recessive otospondylomegaepiphyseal dysplasia (ORPHA:1427); Fibrochondrogenesis (ORPHA:2021); Autosomal dominant otospondylomegaepiphyseal dysplasia (ORPHA:166100)
COL12A1	Bethlem myopathy 2 (OMIM:616471); Ullrich congenital muscular dystrophy 2 (OMIM:616470); Ullrich congenital muscular dystrophy (ORPHA:75840); Bethlem muscular dystrophy (ORPHA:610); Myopathic Ehlers-Danlos syndrome (ORPHA:536516)
COL17A1	Epidermolysis bullosa, junctional, Non-Herlitz type (OMIM:226650); Epithelial recurrent erosion dystrophy (OMIM:122400); Epidermolysis bullosa, junctional 4, intermediate (OMIM:619787); Late-onset junctional epidermolysis bullosa (ORPHA:79406); Intermediate generalized junctional epidermolysis bullosa (ORPHA:79402); Localized junctional epidermolysis bullosa (ORPHA:251393); Epithelial recurrent erosion dystrophy (ORPHA:293381)
COL18A1	Glaucoma, primary closed-angle (OMIM:618880); Knobloch syndrome, type 1 (OMIM:267750); Knobloch syndrome (ORPHA:1571)
COL1A1	OSTEOPOROSIS (OMIM:166710); Caffey disease (OMIM:114000); Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1 (OMIM:619115); Osteogenesis imperfecta, type III (OMIM:259420); Ehlers-Danlos syndrome, arthrochalasia type, 1 (OMIM:130060); Osteogenesis imperfecta, type II (OMIM:166210); Osteogenesis imperfecta, type IV (OMIM:166220); Osteogenesis imperfecta, type I (OMIM:166200); ORPHA:314029; Classical Ehlers-Danlos syndrome (ORPHA:287); ORPHA:216812; ORPHA:216804; ORPHA:216820; Caffey disease (ORPHA:1310); ORPHA:216796; Arthrochalasia Ehlers-Danlos syndrome (ORPHA:1899); Dermatofibrosarcoma protuberans (ORPHA:31112); ORPHA:230857
COL1A2	Ehlers-Danlos syndrome, autosomal recessive, cardiac valvular form (OMIM:225320); Ehlers-Danlos syndrome, arthrochalasia type, 2 (OMIM:617821); OSTEOPOROSIS (OMIM:166710); Combined osteogenesis

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	imperfecta and Ehlers-Danlos syndrome 2 (OMIM:619120); Osteogenesis imperfecta, type III (OMIM:259420); Osteogenesis imperfecta, type II (OMIM:166210); Osteogenesis imperfecta, type IV (OMIM:166220); ORPHA:314029; ORPHA:216812; ORPHA:216804; ORPHA:216820; ORPHA:216796; Arthrochalasia Ehlers-Danlos syndrome (ORPHA:1899); Cardiac-valvular Ehlers-Danlos syndrome (ORPHA:230851); ORPHA:230857
COL21A1	—
COL2A1	Spondyloepiphyseal dysplasia, Stanescu type (OMIM:616583); Spondylometaphyseal dysplasia, corner Fracture type (OMIM:184255); Spondylometaphyseal dysplasia, Algerian type (OMIM:184253); Spondyloepimetaphyseal dysplasia, Strudwick type (OMIM:184250); Czech dysplasia, Metatarsal type (OMIM:609162); Vitreoretinopathy with phalangeal epiphyseal dysplasia (OMIM:619248); Osteoarthritis with mild chondrodysplasia (OMIM:604864); Platyspondylic lethal skeletal dysplasia, Torrance type (OMIM:151210); Achondrogenesis, type II (OMIM:200610); Stickler syndrome, type I (OMIM:108300); Stickler sydrome, type I, nonsyndromic ocular (OMIM:609508); Avascular necrosis of femoral head, primary, 1 (OMIM:608805); Epiphyseal dysplasia, multiple, with myopia and conductive deafness (OMIM:132450); Spondyloepiphyseal dysplasia congenita (OMIM:183900); Kniest dysplasia (OMIM:156550); Legg-Calve-Perthes disease (OMIM:150600); Spondyloperipheral dysplasia (OMIM:271700); Stickler syndrome type 1 (ORPHA:90653); ORPHA:209867; ORPHA:137678; Autosomal dominant otospondylomegaepiphyseal dysplasia (ORPHA:166100); Familial avascular necrosis of femoral head (ORPHA:86820); Spondyloperipheral dysplasia-short ulna syndrome (ORPHA:1856); ORPHA:93279; Legg-Calvé-Perthes disease (ORPHA:2380); Achondrogenesis type 2 (ORPHA:93296); ORPHA:93297; Spondylometaphyseal dysplasia, 'corner fracture' type (ORPHA:93315); Spondylometaphyseal dysplasia, Schmidt type (ORPHA:93316); Spondyloepimetaphyseal dysplasia congenita, Strudwick type (ORPHA:93346); Dysspondyloenchondromatosis (ORPHA:85198); ORPHA:459051; Kniest dysplasia (ORPHA:485); Spondyloepiphyseal dysplasia congenita (ORPHA:94068); Platyspondylic dysplasia, Torrance type (ORPHA:85166)
COL3A1	Polymicrogyria with or without vascular-type EDS (OMIM:618343); Ehlers-Danlos syndrome, Vascular type (OMIM:130050); ORPHA:636941; ORPHA:86; Vascular Ehlers-Danlos syndrome (ORPHA:286); Acrogeria (ORPHA:2500); Familial cerebral saccular aneurysm (ORPHA:231160)
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps (OMIM:611773); OMIM:614519; Retinal arteries, tortuosity of (OMIM:180000); Brain small vessel disease 1 with or without ocular anomalies (OMIM:175780); Microangiopathy and leukoencephalopathy, pontine, autosomal dominant (OMIM:618564); ORPHA:99810; HANAC syndrome (ORPHA:73229); ORPHA:36383; ORPHA:481986; ORPHA:75326; Walker-Warburg syndrome (ORPHA:899); ORPHA:477749
COL4A2	Brain small vessel disease 2B, autosomal recessive (OMIM:621414); OMIM:614519; Brain small vessel disease 2 (OMIM:614483); ORPHA:99810; ORPHA:36383
COL4A3	Alport syndrome 3, autosomal dominant (OMIM:104200); Alport syndrome, X-linked (OMIM:301050); Alport syndrome 3B, autosomal recessive (OMIM:620536); Hematuria, benign familial, 2 (OMIM:620320); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656); ORPHA:653722; ORPHA:88919; ORPHA:88918
COL4A4	Hematuria, benign familial (OMIM:141200); Alport syndrome, X-linked (OMIM:301050); Alport syndrome 2, autosomal recessive (OMIM:203780); ORPHA:653722; ORPHA:88919; ORPHA:88918
COL4A5	Alport syndrome, X-linked (OMIM:301050); X-linked Alport syndrome-diffuse leiomyomatosis (ORPHA:1018); ORPHA:653722; ORPHA:88917
COL4A6	Deafness, X-linked 6 (OMIM:300914); ORPHA:90625; X-linked Alport syndrome-diffuse leiomyomatosis (ORPHA:1018)
COL5A1	Fibromuscular dysplasia, multifocal (OMIM:619329); Ehlers-danlos syndrome, type I (OMIM:130000); Classical Ehlers-Danlos syndrome (ORPHA:287)
COL5A2	Ehlers-Danlos syndrome, classic type, 2 (OMIM:130010); Classical Ehlers-Danlos syndrome (ORPHA:287)
COL6A1	Ullrich congenital muscular dystrophy 1 (OMIM:254090); Bethlem myopathy 1A (OMIM:158810); Ullrich congenital muscular dystrophy (ORPHA:75840); Bethlem muscular dystrophy (ORPHA:610); ORPHA:646113
COL6A2	Myosclerosis, autosomal recessive (OMIM:255600); Ullrich congenital muscular dystrophy 1B (OMIM:620727); Bethlem myopathy 1B (OMIM:620725); ORPHA:289380; Ullrich congenital muscular dystrophy (ORPHA:75840); Bethlem muscular dystrophy (ORPHA:610); ORPHA:646113
COL6A3	Dystonia 27 (OMIM:616411); Bethlem myopathy 1C (OMIM:620726); Ullrich congenital muscular dystrophy 1C (OMIM:620728); Ullrich congenital muscular dystrophy (ORPHA:75840); Primary dystonia, DYT27 type (ORPHA:464440); Bethlem muscular dystrophy (ORPHA:610); ORPHA:646113
COL7A1	Epidermolysis bullosa dystrophica, autosomal recessive (OMIM:226600); Epidermolysis bullosa dystrophica, pretibial (OMIM:131850); Epidermolysis bullosa dystrophica, autosomal dominant (OMIM:131750); Transient bullous dermolysis of the newborn (OMIM:131705); Epidermolysis bullosa with congenital localized absence of skin anddeformity of nails (OMIM:132000); Nail disorder, nonsyndromic congenital, 8 (OMIM:607523); Epidermolysis bullosa pruriginosa (OMIM:604129); Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form (ORPHA:79408); Recessive dystrophic epidermolysis bullosa inversa (ORPHA:79409); Localized dystrophic epidermolysis bullosa, pretibial form (ORPHA:79410); Self-improving dystrophic epidermolysis bullosa (ORPHA:79411); Localized dystrophic epidermolysis bullosa, nails only (ORPHA:158676); Localized dystrophic epidermolysis bullosa, acral form (ORPHA:158673); Autosomal recessive generalized dystrophic epidermolysis bullosa, intermediate form (ORPHA:89842); Dystrophic epidermolysis bullosa pruriginosa (ORPHA:89843); Autosomal dominant generalized dystrophic epidermolysis bullosa (ORPHA:231568)
COL8A2	Corneal dystrophy, posterior polymorphous, 2 (OMIM:609140); Corneal dystrophy, fuchs endothelial, 1 (OMIM:136800); Posterior polymorphous corneal dystrophy (ORPHA:98973); Fuchs endothelial corneal dystrophy (ORPHA:98974)

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COL9A1	Stickler syndrome, type IV (OMIM:614134); Epiphyseal dysplasia, multiple, 6 (OMIM:614135); Autosomal recessive Stickler syndrome (ORPHA:250984); Multiple epiphyseal dysplasia due to collagen 9 anomaly (ORPHA:166002)
COL9A2	Stickler syndrome, type V (OMIM:614284); Epiphyseal dysplasia, multiple, 2 (OMIM:600204); Autosomal recessive Stickler syndrome (ORPHA:250984); Multiple epiphyseal dysplasia due to collagen 9 anomaly (ORPHA:166002)
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy (OMIM:600969); Stickler syndrome, type VI (OMIM:620022); OMIM:603932; Autosomal recessive Stickler syndrome (ORPHA:250984); Multiple epiphyseal dysplasia due to collagen 9 anomaly (ORPHA:166002)
COLEC11	3MC syndrome 2 (OMIM:265050); 3MC syndrome (ORPHA:293843)
COLQ	Myasthenic syndrome, congenital, 5 (OMIM:603034); Synaptic congenital myasthenic syndrome (ORPHA:98915)
COMETT	—
COMMD1	—
COMMD3	—
COMP	Pseudoachondroplasia (OMIM:177170); Carpal tunnel syndrome 2 (OMIM:619161); Epiphyseal dysplasia, multiple, 1 (OMIM:132400); Pseudoachondroplasia (ORPHA:750); Multiple epiphyseal dysplasia type 1 (ORPHA:93308)
COMT	OMIM:621296; 22q11.2 deletion syndrome (ORPHA:567)
COPA	Autoimmune interstitial lung, joint, and kidney disease (OMIM:616414); ORPHA:444092
COPB2	Microcephaly 19, primary, autosomal recessive (OMIM:617800); Osteoporosis, childhood- or juvenile-onset, with developmental delay (OMIM:619884); Autosomal recessive primary microcephaly (ORPHA:2512)
COQ2	Multiple system atrophy 1, susceptibility to (OMIM:146500); Coenzyme Q10 deficiency, primary, 1 (OMIM:607426); Multiple system atrophy, cerebellar type (ORPHA:227510); Multiple system atrophy, parkinsonian type (ORPHA:98933)
COQ5	Coenzyme Q10 deficiency, primary, 9 (OMIM:619028)
COQ6	Coenzyme Q10 deficiency, primary, 6 (OMIM:614650); Full schwannomatosis (ORPHA:93921); ORPHA:280406
COQ8A	Coenzyme Q10 deficiency, primary, 4 (OMIM:612016); Autosomal recessive ataxia due to ubiquinone deficiency (ORPHA:139485)
COQ8B	Nephrotic syndrome, type 9 (OMIM:615573); Retinitis pigmentosa (ORPHA:791); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
COQ9	Coenzyme Q10 deficiency, primary, 5 (OMIM:614654); ORPHA:319678
CORIN	Preeclampsia/eclampsia 5 (OMIM:614595); Cardiomyopathy, familial hypertrophic, 30, atrial (OMIM:620734); Preeclampsia (ORPHA:275555)
CORO1A	Immunodeficiency 8 (OMIM:615401); ORPHA:228003
COX10	Mitochondrial complex IV deficiency, nuclear type 3 (OMIM:619046); Mitochondrial complex IV deficiency, nuclear type 1 (OMIM:220110); ORPHA:254905
COX14	Mitochondrial complex IV deficiency, nuclear type 10 (OMIM:619053); ORPHA:254905
COX4I1	Mitochondrial complex IV deficiency, nuclear type 16 (OMIM:619060); ORPHA:254905
COX4I2	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarialhyperostosis (OMIM:612714); ORPHA:199337
COX6B1	Mitochondrial complex IV deficiency, nuclear type 7 (OMIM:619051); Mitochondrial complex IV deficiency, nuclear type 1 (OMIM:220110); ORPHA:254905
COX6C	—
COX7B	Linear skin defects with multiple congenital anomalies 1 (OMIM:309801); Linear skin defects with multiple congenital anomalies 2 (OMIM:300887); Microphthalmia with linear skin defects syndrome (ORPHA:2556)
CP	ACERULOPLASMINEMIA (OMIM:604290); Aceruloplasminemia (ORPHA:48818)
CPA2	—
CPA6	Epilepsy, familial temporal lobe, 5 (OMIM:614417); Febrile seizures, familial, 11 (OMIM:614418); Genetic epilepsy with febrile seizure plus (ORPHA:36387); ORPHA:163717
CPB2	—
CPE	Intellectual developmental disorder and hypogonadotropic hypogonadism (OMIM:619326); ORPHA:633028
CPEB1	—
CPLANE1	Orofaciodigital syndrome VI (OMIM:277170); Joubert syndrome 17 (OMIM:614615); Monomelic amyotrophy (ORPHA:65684); Orofaciodigital syndrome type 6 (ORPHA:2754); Isolated Joubert syndrome (ORPHA:475)
CPLX2	—
CPN1	Carboxypeptidase N deficiency (OMIM:212070)
CPOX	Harderoporphyria (OMIM:618892); Coproporphyria (OMIM:121300); ORPHA:659672; Hereditary coproporphyria (ORPHA:79273)
CPS1	Carbamoyl phosphate synthetase I deficiency, hyperammonemia due to (OMIM:237300); Carbamoyl-phosphate synthetase 1 deficiency (ORPHA:147)
CPT1A	Carnitine palmitoyltransferase I deficiency (OMIM:255120); Carnitine palmitoyl transferase 1A deficiency (ORPHA:156)
CPT1B	—
CPT2	Carnitine palmitoyltransferase II deficiency, myopathic, stress-induced (OMIM:255110); Encephalopathy, acute, infection-induced, susceptibility to, 4 (OMIM:614212); Carnitine palmitoyltransferase II deficiency, lethal neonatal (OMIM:608836); Carnitine palmitoyltransferase II deficiency, infantile (OMIM:600649); Carnitine palmitoyl transferase II deficiency, myopathic form (ORPHA:228302); Carnitine palmitoyl transferase II

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	deficiency, severe infantile form (ORPHA:228305); Carnitine palmitoyl transferase II deficiency, neonatal form (ORPHA:228308); ORPHA:263524
CPZ	—
CR1	OMIM:607486; OMIM:611162
CR2	Immunodeficiency, common variable, 7 (OMIM:614699); OMIM:610927; Immunodeficiency, common variable, 2 (OMIM:240500); ORPHA:696894; Systemic lupus erythematosus (ORPHA:536)
CRADD	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly (OMIM:614499); ORPHA:88616
CRB1	Pigmented paravenous chorioretinal atrophy (OMIM:172870); Retinitis pigmentosa 12 (OMIM:600105); Leber congenital amaurosis 8 (OMIM:613835); Leber congenital amaurosis (ORPHA:65); ORPHA:251295; Retinitis pigmentosa (ORPHA:791); Nanophthalmos (ORPHA:35612)
CRBN	Intellectual developmental disorder, autosomal recessive 2 (OMIM:607417); ORPHA:88616
CREB1	Histiocytoma, angiomatoid fibrous, somatic (OMIM:612160); ORPHA:97338; ORPHA:569164
CREB3L3	Hypertriglyceridemia 2 (OMIM:619324); ORPHA:300293
CREBBP	Rubinstein-Taybi syndrome 1 (OMIM:180849); Menke-Hennekam syndrome 1 (OMIM:618332); Rubinstein-Taybi syndrome due to 16p13.3 microdeletion (ORPHA:353281); Rubinstein-Taybi syndrome due to CREBBP mutations (ORPHA:353277); Menke-Hennekam syndrome (ORPHA:592574); ORPHA:370026
CRELD1	Atrioventricular septal defect, partial, with heterotaxy syndrome (OMIM:606217); Jeffries-Lakhani neurodevelopmental syndrome (OMIM:620771); ORPHA:99067; ORPHA:99068; ORPHA:576235
CRIM1	ORPHA:468672
CRISP2	—
CRKL	Distal 22q11.2 microdeletion syndrome (ORPHA:261330)
CRLF1	Crisponi/cold-induced sweating syndrome 1 (OMIM:272430); Crisponi syndrome (ORPHA:1545); Idiopathic achalasia (ORPHA:930); ORPHA:157820
CRMP1	—
CRNDE	—
CRP	—
CRPPA	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7 (OMIM:614643); Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7 (OMIM:616052); Congenital muscular dystrophy without intellectual disability (ORPHA:370980); ISPD-related limb-girdle muscular dystrophy R20 (ORPHA:352479); Walker-Warburg syndrome (ORPHA:899); Muscle-eye-brain disease (ORPHA:588)
CRTAP	Osteogenesis imperfecta, type VII (OMIM:610682); Cole-Carpenter syndrome (ORPHA:2050); ORPHA:216812; ORPHA:216804; ORPHA:216820
CRX	Retinitis pigmentosa (OMIM:268000); Leber congenital amaurosis 7 (OMIM:613829); Cone-Rod dystrophy 2 (OMIM:120970); Leber congenital amaurosis (ORPHA:65); Cone rod dystrophy (ORPHA:1872); Retinitis pigmentosa (ORPHA:791)
CRYAA	Cataract 9, multiple types (OMIM:604219); Cataract-microcornea syndrome (ORPHA:1377); ORPHA:98994; ORPHA:98991; ORPHA:98988; ORPHA:441452
CRYAB	Cardiomyopathy, dilated, 1II (OMIM:615184); Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related (OMIM:613869); Cataract 16, multiple types (OMIM:613763); Alpha-B crystallin-related late-onset myopathy (ORPHA:399058); Familial isolated dilated cardiomyopathy (ORPHA:154); ORPHA:280553; ORPHA:98991; ORPHA:98993; ORPHA:441452
CRYBA1	Cataract, congenital zonular, with sutural opacities (OMIM:600881); ORPHA:98991; ORPHA:98993; ORPHA:98985; ORPHA:441452
CRYBA4	Cataract 23 (OMIM:610425); Cataract-microcornea syndrome (ORPHA:1377); ORPHA:441452
CRYBB1	Cataract 17, multiple types (OMIM:611544); Cataract-microcornea syndrome (ORPHA:1377); ORPHA:98991; ORPHA:98984
CRYBB2	Cataract 3, multiple types (OMIM:601547); Cataract-microcornea syndrome (ORPHA:1377); ORPHA:441447; ORPHA:98994; ORPHA:98991; ORPHA:98989; ORPHA:98984; ORPHA:98985
CRYBB3	Cataract 22, multiple types (OMIM:609741); ORPHA:98991; ORPHA:98988
CRYGA	—
CRYGB	Cataract, multiple types (OMIM:615188); ORPHA:98994; ORPHA:98988; ORPHA:441452
CRYGC	Cataract 2, multiple types (OMIM:604307); Cataract-microcornea syndrome (ORPHA:1377); ORPHA:98991; ORPHA:98984; ORPHA:441452
CRYGD	Cataract, crystalline aculeiform (OMIM:115700); Cataract-microcornea syndrome (ORPHA:1377); ORPHA:98990; ORPHA:98991; ORPHA:98989; ORPHA:98984; ORPHA:441452
CRYGS	Cataract 20, multiple types (OMIM:116100); ORPHA:98985; ORPHA:441452
CRYM	Deafness, autosomal dominant 40 (OMIM:616357); ORPHA:90635
CSAG1	—
CSDE1	—
CSE1L	—
CSF1	—
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids (OMIM:221820); Brain abnormalities, neurodegeneration, and dysosteosclerosis (OMIM:618476); ORPHA:313808; ORPHA:556985
CSF2	—
CSF2RA	Surfactant metabolism dysfunction, pulmonary, 4 (OMIM:300770); Hereditary pulmonary alveolar proteinosis (ORPHA:264675)
CSF2RB	Surfactant metabolism dysfunction, pulmonary, 5 (OMIM:614370); Hereditary pulmonary alveolar proteinosis (ORPHA:264675)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
CSF3R	Neutrophilia, hereditary (OMIM:162830); Neutropenia, severe congenital, 7, autosomal recessive (OMIM:617014); ORPHA:98824; ORPHA:86829; ORPHA:420702; ORPHA:279943
CSGALNACT1	Skeletal dysplasia, mild, with joint laxity and advanced bone age (OMIM:618870); Desbuquois syndrome (ORPHA:1425)
CSH1	—
CSMD1	—
CSMD2	—
CSMD3	—
CSNK1A1	—
CSNK1D	Advanced sleep phase syndrome, familial, 2 (OMIM:615224); ORPHA:164736
CSNK1G1	—
CSNK2A2	—
CSRP3	Cardiomyopathy, familial hypertrophic, 12 (OMIM:612124); Cardiomyopathy, dilated, 1M (OMIM:607482); Familial isolated dilated cardiomyopathy (ORPHA:154)
CST3	Macular degeneration, age-related, 11 (OMIM:611953); Leukodystrophy, adult-onset, autosomal dominant, without amyloid angiopathy (OMIM:621214); Amyloidosis VI (OMIM:105150); ACys amyloidosis (ORPHA:100008)
CST6	Ectodermal dysplasia 15, Hypohidrotic/hair type (OMIM:618535)
CSTA	Peeling skin syndrome 4 (OMIM:607936); Acral peeling skin syndrome (ORPHA:263534); ORPHA:289586
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg) (OMIM:254800); Progressive myoclonic epilepsy type 1 (ORPHA:308); Autosomal recessive hypohidrotic ectodermal dysplasia (ORPHA:248)
CSTF2T	—
CTC1	Cerebroretinal microangiopathy with calcifications and cysts 1 (OMIM:612199); ORPHA:313838; Dyskeratosis congenita (ORPHA:1775)
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy (OMIM:604168); Congenital cataracts-facial dysmorphism-neuropathy syndrome (ORPHA:48431)
CTF1	—
CTH	CYSTATHIONINURIA (OMIM:219500); Cystathioninuria (ORPHA:212)
CTHRC1	Barrett esophagus (OMIM:614266)
CTLA4	Autoimmune lymphoproliferative syndrome, type V (OMIM:616100); Sézary syndrome (ORPHA:3162); Classic mycosis fungoides (ORPHA:2584); Granulomatosis with polyangiitis (ORPHA:900); ORPHA:391490; Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency (ORPHA:436159); Systemic lupus erythematosus (ORPHA:536)
CTNNA2	Cortical dysplasia, complex, with other brain malformations 9 (OMIM:618174)
CTNNA3	Arrhythmogenic right ventricular dysplasia, familial, 13 (OMIM:615616); ORPHA:293888; ORPHA:293899; ORPHA:293910
CTNNB1	Neurodevelopmental disorder with spastic diplegia and visual defects (OMIM:615075); Exudative vitreoretinopathy 7 (OMIM:617572); Colorectal cancer (OMIM:114500); Ovarian cancer (OMIM:167000); Hepatocellular carcinoma (OMIM:114550); MEDULLOBLASTOMA (OMIM:155255); Pilomatrixoma, somatic (OMIM:132600); ORPHA:689430; ORPHA:210159; Osteopathia striata-cranial sclerosis syndrome (ORPHA:2780); Adrenocortical carcinoma (ORPHA:1501); Pediatric hepatocellular carcinoma (ORPHA:33402); Pilomatrixoma (ORPHA:91414); Desmoid tumor (ORPHA:873); Familial exudative vitreoretinopathy (ORPHA:891); Acrofacial dysostosis, Weyers type (ORPHA:952); Craniopharyngioma (ORPHA:54595); Intellectual disability-eye abnormalities-microcephaly-peripheral spasticity syndrome (ORPHA:404473); ORPHA:569248
CTNND2	ORPHA:178469; Monosomy 5p syndrome (ORPHA:281); Familial adult myoclonic epilepsy (ORPHA:86814)
CTNS	Cystinosis, late-onset juvenile or adolescent nephropathic (OMIM:219900); Cystinosis, nephropathic (OMIM:219800); Cystinosis, adult nonnephropathic (OMIM:219750); Infantile nephropathic cystinosis (ORPHA:411629); Ocular cystinosis (ORPHA:411641); Juvenile nephropathic cystinosis (ORPHA:411634)
CTRC	Pancreatitis, hereditary (OMIM:167800); Autosomal dominant hereditary chronic pancreatitis (ORPHA:676); Tropical pancreatitis (ORPHA:103918)
CTSA	Galactosialidosis (OMIM:256540); Brain small vessel disease 6 with leukoencephalopathy (OMIM:621394); Galactosialidosis (ORPHA:351); ORPHA:575553
CTSB	Keratolytic winter erythema (OMIM:148370); Keratolytic winter erythema (ORPHA:50943)
CTSC	Periodontitis, aggressive, 1 (OMIM:170650); Haim-Munk syndrome (OMIM:245010); Papillon-Lefevre syndrome (OMIM:245000); Haim-Munk syndrome (ORPHA:2342); Papillon-Lefèvre syndrome (ORPHA:678)
CTSD	Ceroid lipofuscinosis, neuronal, 10 (OMIM:610127); ORPHA:700487; ORPHA:700492; ORPHA:700497
CTSG	—
CTSK	Pycnodysostosis (OMIM:265800); Pycnodysostosis (ORPHA:763)
CTTNBP2	—
CUBN	Megaloblastic anemia 1 (OMIM:261100); Proteinuria, chronic benign (OMIM:618884); Imerslund-Gräsbeck syndrome (ORPHA:35858)
CUEDC1	—
CUL2	—
CUL3	Neurodevelopmental disorder with or without autism or seizures (OMIM:619239); Pseudohypoaldosteronism, type IIE (OMIM:614496); ORPHA:300530; ORPHA:528084
CUL4B	Intellectual developmental disorder, X-linked syndromic, Cabezas type (OMIM:300354); X-linked intellectual disability, Cabezas type (ORPHA:85293)
CUL5	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
CUL7	3-M syndrome 1 (OMIM:273750); 3M syndrome (ORPHA:2616)
CUTC	—
CUX1	Global developmental delay with or without impaired intellectual development (OMIM:618330); ORPHA:178469
CUX2	Epileptic encephalopathy, early infantile, 67 (OMIM:618141); Lennox-Gastaut syndrome (ORPHA:2382)
CX3CR1	OMIM:609423; OMIM:607339; OMIM:613784
CXCL12	OMIM:609423
CXCL5	—
CXCR1	OMIM:609423
CXCR4	Whim syndrome (OMIM:193670); WHIM syndrome (ORPHA:51636)
CYB5A	Methemoglobinemia and ambiguous genitalia (OMIM:250790); 46,XY difference of sex development due to isolated 17,20-lyase deficiency (ORPHA:90796); Autosomal recessive methemoglobinemia (ORPHA:621)
CYB5R3	Methemoglobinemia due to deficiency of methemoglobin reductase (OMIM:250800); Autosomal recessive methemoglobinemia (ORPHA:621)
CYB5R4	—
CYBA	GRANULOMATOUS DISEASE, CHRONIC, AUTOSOMAL RECESSIVE, CYTOCHROME b-NEGATIVE (OMIM:233690); Chronic granulomatous disease (ORPHA:379)
CYBB	Chronic granulomatous disease, X-linked (OMIM:306400); Atypical mycobacteriosis, familial, X-linked 2 (OMIM:300645); Chronic granulomatous disease (ORPHA:379); ORPHA:319605
CYBRD1	—
CYCSP38	—
CYCTP	—
CYFIP1	—
CYGB	—
CYLD	Frontotemporal dementia and/or amytrophic lateral sclerosis 8 (OMIM:619132); Brooke-Spiegler syndrome (OMIM:605041); Cylindromatosis, familial (OMIM:132700); Trichoepithelioma, multiple familial, 1 (OMIM:601606); Familial cylindromatosis (ORPHA:211); Familial multiple trichoepithelioma (ORPHA:867)
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete (OMIM:613743); Inherited isolated adrenal insufficiency due to partial CYP11A1 deficiency (ORPHA:289548); 46,XY difference of sex development-adrenal insufficiency due to CYP11A1 deficiency (ORPHA:168558)
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency (OMIM:202010); Aldosteronism, glucocorticoid-remediable (OMIM:103900); Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency (ORPHA:90795); Familial hyperaldosteronism type I (ORPHA:403)
CYP11B2	Hypoaldosteronism, congenital, due to CMO II deficiency (OMIM:610600); Corticosterone methyloxidase type I deficiency (OMIM:203400); Familial hyperaldosteronism type I (ORPHA:403); Early-onset familial hypoaldosteronism (ORPHA:556030)
CYP19A1	Aromatase deficiency (OMIM:613546); Aromatase deficiency (ORPHA:91); Aromatase excess syndrome (ORPHA:178345)
CYP1A2	—
CYP1B1	Glaucoma 3, primary infantile, B (OMIM:600975); Glaucoma 3, primary congenital, A (OMIM:231300); Anterior segment dysgenesis 6, multiple subtypes (OMIM:617315); Congenital glaucoma (ORPHA:98976); Juvenile glaucoma (ORPHA:98977); Peters anomaly (ORPHA:708)
CYP21A1P	—
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency (OMIM:201910); ORPHA:315311; ORPHA:315306
CYP24A1	Hypercalcemia, infantile, 1 (OMIM:143880); ORPHA:300547
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies (OMIM:614416); ORPHA:293925
CYP26C1	Focal facial dermal dysplasia 4 (OMIM:614974); Focal facial dermal dysplasia type IV (ORPHA:398189)
CYP27A1	Cerebrotendinous xanthomatosis (OMIM:213700); Cerebrotendinous xanthomatosis (ORPHA:909)
CYP27B1	Vitamin D hydroxylation-deficient rickets, type 1A (OMIM:264700); Hypocalcemic vitamin D-dependent rickets (ORPHA:289157)
CYP2A13	—
CYP2A6	OMIM:621426; Coumarin resistance (OMIM:122700); OMIM:188890; Lung cancer, susceptibility to (OMIM:211980)
CYP2B6	OMIM:614546
CYP2C18	—
CYP2C19	OMIM:609535
CYP2C8	OMIM:618018
CYP2C9	Coumarin resistance (OMIM:122700)
CYP2D6	Drug metabolism, poor, cyp2d6-relateddrug metabolism, ultrarapid, cyp2d6-related, included (OMIM:608902)
CYP2E1	—
CYP2F1	—
CYP2J2	—
CYP2R1	Vitamin D hydroxylation-deficient rickets, type 1B (OMIM:600081); Hypocalcemic vitamin D-dependent rickets (ORPHA:289157)
CYP2U1	Spastic paraplegia 56, autosomal recessive (OMIM:615030); Autosomal recessive spastic paraplegia type 56 (ORPHA:320411)
CYP3A4	Vitamin d-dependent rickets, type 3 (OMIM:619073)
CYP3A5	Hypertension, essential (OMIM:145500)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
CYP3A51P	—
CYP3A7	—
CYP4A22	—
CYP4F12	—
CYP4F2	—
CYP4F22	Ichthyosis, congenital, autosomal recessive 5 (OMIM:604777); Lamellar ichthyosis (ORPHA:313)
CYP4F3	—
CYP4V2	Bietti crystalline corneoretinal dystrophy (OMIM:210370); Bietti crystalline dystrophy (ORPHA:41751)
CYP51A1	ORPHA:521432
CYP7A1	Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency (ORPHA:209902)
CYP7B1	Spastic paraplegia 5A, autosomal recessive (OMIM:270800); Bile acid synthesis defect, congenital, 3 (OMIM:613812); Autosomal recessive spastic paraplegia type 5A (ORPHA:100986); Congenital bile acid synthesis defect type 3 (ORPHA:79302)
CYS1	—
D2HGDH	D-2-Hydroxyglutaric aciduria 1 (OMIM:600721); D-2-hydroxyglutaric aciduria (ORPHA:79315)
DACH1	—
DAD1	—
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9 (OMIM:616538); Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9 (OMIM:613818); Muscle-eye-brain disease with bilateral multicystic leucodystrophy (ORPHA:370997); Alpha-dystroglycan-related limb-girdle muscular dystrophy R16 (ORPHA:280333); Walker-Warburg syndrome (ORPHA:899); Isolated asymptomatic elevation of creatine phosphokinase (ORPHA:206599)
DALRD3	Epileptic encephalopathy, early infantile, 86 (OMIM:618910); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
DAO	Amyotrophic lateral sclerosis (ORPHA:803)
DARS1	Hypomyelination with brainstem and spinal cord involvement and legspasticity (OMIM:615281); ORPHA:363412
DARS2	Charcot-Marie-Tooth disease, axonal, type 2LL (OMIM:621485); Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation (OMIM:611105); Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome (ORPHA:137898)
DAZ4	Chromosome Y microdeletion syndrome (ORPHA:1646)
DAZL	—
DBH	Orthostatic hypotension 1, due to DBH deficiency (OMIM:223360); Dopamine beta-hydroxylase deficiency (ORPHA:230)
DBNL	—
DBT	Maple syrup urine disease, type II (OMIM:620699); ORPHA:268145; ORPHA:268162; ORPHA:268173; ORPHA:268184
DCAF17	Woodhouse-Sakati syndrome (OMIM:241080)
DCAF8	Giant axonal neuropathy, autosomal dominant (OMIM:610100); ORPHA:401964
DCBLD1	—
DCC	Gaze palsy, familial horizontal, with progressive scoliosis, 2 (OMIM:617542); Esophageal cancer, somatic (OMIM:133239); Colorectal cancer (OMIM:114500); Mirror movements 1 and/or agenesis of the corpus callosum (OMIM:157600); Horizontal gaze palsy with progressive scoliosis (ORPHA:2744); Familial congenital mirror movements (ORPHA:238722); Kallmann syndrome (ORPHA:478)
DCK	—
DCLRE1B	Dyskeratosis congenita, autosomal recessive 8 (OMIM:620133)
DCLRE1C	Omenn syndrome (OMIM:603554); Severe combined immunodeficiency with sensitivity to ionizing radiation (OMIM:602450); Severe combined immunodeficiency due to DCLRE1C deficiency (ORPHA:275); Omenn syndrome (ORPHA:39041)
DCN	Corneal dystrophy, congenital stromal (OMIM:610048); ORPHA:101068
DCTD	—
DCTN1	Amyotrophic lateral sclerosis 1 (OMIM:105400); Neuronopathy, distal hereditary motor, type VIIB (OMIM:607641); Perry syndrome (OMIM:168605); ORPHA:139589; Amyotrophic lateral sclerosis (ORPHA:803); Perry syndrome (ORPHA:178509)
DCX	Lissencephaly, X-linked, 1 (OMIM:300067); Lissencephaly type 1 due to doublecortin gene mutation (ORPHA:2148); ORPHA:99796
DCXR	Pentosuria (OMIM:260800); Pentosuria (ORPHA:2843)
DDB2	Xeroderma pigmentosum, complementation group E (OMIM:278740); Xeroderma pigmentosum (ORPHA:910)
DDC	Aromatic L-amino acid decarboxylase deficiency (OMIM:608643); Aromatic L-amino acid decarboxylase deficiency (ORPHA:35708)
DDHD1	Spastic paraplegia 28, autosomal recessive (OMIM:609340); Autosomal recessive spastic paraplegia type 28 (ORPHA:101008)
DDHD2	Spastic paraplegia 54, autosomal recessive (OMIM:615033); Autosomal recessive spastic paraplegia type 54 (ORPHA:320380)
DDI2	—
DDIAS	—
DDOST	Congenital disorder of glycosylation, type IR (OMIM:614507); DDOST-CDG (ORPHA:300536)
DDR2	Warburg-Cinotti syndrome (OMIM:618175); Spondylometaepiphyseal dysplasia, short Limb-Hand type (OMIM:271665); ORPHA:93358

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
DDRGK1	Spondyloepimetaphyseal dysplasia, Shohat type (OMIM:602557); Spondyloepimetaphyseal dysplasia, Shohat type (ORPHA:93352)
DDX25	—
DDX3X	Intellectual developmental disorder, X-linked, syndrome, snijders Blok type (OMIM:300958); ORPHA:99861; Toriello-Carey syndrome (ORPHA:3338); X-linked intellectual disability-hypotonia-movement disorder syndrome (ORPHA:457260)
DDX3Y	Spermatogenic failure, Y-linked, 2 (OMIM:415000); Chromosome Y microdeletion syndrome (ORPHA:1646)
DDX41	Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to (OMIM:616871); ORPHA:488647
DDX5	—
DDX6	Intellectual developmental disorder with impaired language and dysmorphic facies (OMIM:618653); ORPHA:528084
DDX60L	—
DEAF1	Vulto-van Silfout-de Vries syndrome (OMIM:615828); Dyskinesia, seizures, and intellectual developmental disorder (OMIM:617171); ORPHA:714385; Intellectual disability-epilepsy-extrapyramidal syndrome (ORPHA:468620)
DEF6	Immunodeficiency 87 and autoimmunity (OMIM:619573)
DEFB126	—
DENND2B	—
DENND4A	—
DES	Cardiomyopathy, dilated, 1I (OMIM:604765); Myopathy, myofibrillar, 1 (OMIM:601419); Scapuloperoneal syndrome, neurogenic, Kaeser type (OMIM:181400); Familial isolated dilated cardiomyopathy (ORPHA:154); Desminopathy (ORPHA:98909); ORPHA:85146
DGCR2	Velocardiofacial syndrome (OMIM:192430)
DGCR6	Velocardiofacial syndrome (OMIM:192430)
DGKD	—
DGKE	Nephrotic syndrome, type 7 (OMIM:615008); ORPHA:329903; ORPHA:357008
DGKQ	—
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type) (OMIM:251880); Portal hypertension, noncirrhotic (OMIM:617068); Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4 (OMIM:617070); Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency (ORPHA:329314); ORPHA:279934
DHCR24	Desmosterolosis (OMIM:602398); Desmosterolosis (ORPHA:35107)
DHCR7	Smith-Lemli-Opitz syndrome (OMIM:270400); Smith-Lemli-Opitz syndrome (ORPHA:818)
DHDDS	Developmental delay and seizures with or without movement abnormalities (OMIM:617836); Congenital disorder of glycosylation, type 1bb (OMIM:621567); Retinitis pigmentosa 59 (OMIM:613861); Retinitis pigmentosa (ORPHA:791); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency (OMIM:613839); ORPHA:319651
DHFR2	—
DHH	46XY gonadal dysgenesis with minifascicular neuropathy (OMIM:607080); 46,xy sex reversal 7 (OMIM:233420); 46,XY complete gonadal dysgenesis (ORPHA:242); 46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome (ORPHA:168563)
DHODH	Postaxial acrofacial dysostosis (OMIM:263750); Postaxial acrofacial dysostosis (ORPHA:246)
DHRS4	—
DHRS4L1	—
DHTKD1	Charcot-Marie-Tooth disease, axonal, type 2Q (OMIM:615025); Alpha-aminoadipic and alpha-ketoadipic aciduria (OMIM:204750); ORPHA:329258; 2-aminoadipic 2-oxoadipic aciduria (ORPHA:79154)
DHX15	—
DHX30	Neurodevelopmental disorder with severe motor impairment and absent language (OMIM:617804); ORPHA:647788
DHX37	46, XY sex reversal 11 (OMIM:273250); Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies (OMIM:618731); 46,XY partial gonadal dysgenesis (ORPHA:251510); 46,XY complete gonadal dysgenesis (ORPHA:242); Testicular regression syndrome (ORPHA:983)
DHX8	—
DIABLO	Deafness, autosomal dominant 64 (OMIM:614152); ORPHA:90635
DIAPH1	Deafness, autosomal dominant 1 (OMIM:124900); Seizures, cortical blindness, and microcephaly syndrome (OMIM:616632); Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome (ORPHA:477814); DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome (ORPHA:494444); Moyamoya disease (ORPHA:2573)
DIAPH2	Premature ovarian failure 2A (OMIM:300511)
DIAPH3	Auditory neuropathy, autosomal dominant, 1 (OMIM:609129); ORPHA:90635
DICER1	Global developmental delay, lung cysts, overgrowth, and wilms tumor (OMIM:618272); Pleuropulmonary blastoma (OMIM:601200); Goiter, multinodular 1 (OMIM:138800); Rhabdomyosarcoma, embryonal, 2 (OMIM:180295); ORPHA:99915; ORPHA:99916; ORPHA:99914; Familial multinodular goiter (ORPHA:276399); ORPHA:404476; ORPHA:284343; ORPHA:99757
DINOL	—
DIP2A	—
DIP2B	Intellectual developmental disorder, autosomal dominant, FRA12A type (OMIM:136630)
DIP2C	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
DIPK1A	—
DIS3L2	Perlman syndrome (OMIM:267000); Perlman syndrome (ORPHA:2849); Nephroblastoma (ORPHA:654)
DISC1	OMIM:604906
DISP1	OMIM:621143; Septopreoptic holoprosencephaly (ORPHA:280195); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925)
DKC1	Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1 (OMIM:301108); Dyskeratosis congenita, X-linked (OMIM:305000); Hoyeraal-Hreidarsson syndrome (ORPHA:3322); Dyskeratosis congenita (ORPHA:1775)
DKK3	—
DLAT	Pyruvate dehydrogenase E2 deficiency (OMIM:245348); Pyruvate dehydrogenase E2 deficiency (ORPHA:79244)
DLC1	Colorectal cancer (OMIM:114500)
DLD	Dihydrolipoamide dehydrogenase deficiency (OMIM:246900); Pyruvate dehydrogenase E3 deficiency (ORPHA:2394)
DLEC1	Squamous cell carcinoma of the esophagus (ORPHA:99977)
DLEU1	—
DLEU2	—
DLEU7	—
DLG2	—
DLG3	Intellectual developmental disorder, X-linked 90 (OMIM:300850); ORPHA:777
DLG4	Intellectual developmental disorder 62 (OMIM:618793); ORPHA:528084
DLGAP3	—
DLL1	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures (OMIM:618709); ORPHA:178469; Septopreoptic holoprosencephaly (ORPHA:280195); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925)
DLL3	Spondylocostal dysostosis, autosomal recessive 1 (OMIM:277300); Autosomal recessive spondylocostal dysostosis (ORPHA:2311)
DLX3	Trichodentoosseous syndrome (OMIM:190320); Amelogenesis imperfecta, type IV (OMIM:104510); Tricho-dento-osseous syndrome (ORPHA:3352); ORPHA:100034
DLX5	Split-Hand/foot malformation 1 with sensorineural hearing loss (OMIM:220600); Split-Hand/foot malformation 1 (OMIM:183600); Isolated split hand-split foot malformation (ORPHA:2440); ORPHA:71271
DLX6	Isolated split hand-split foot malformation (ORPHA:2440)
DM1	—
DMAC2	—
DMC1	—
DMD	Cardiomyopathy, dilated, 3B (OMIM:302045); Muscular dystrophy, Becker type (OMIM:300376); Duchenne muscular dystrophy (OMIM:310200); Familial isolated dilated cardiomyopathy (ORPHA:154); Becker muscular dystrophy (ORPHA:98895); Duchenne muscular dystrophy (ORPHA:98896); ORPHA:777; Symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers (ORPHA:206546)
DMGDH	Dimethylglycine dehydrogenase deficiency (OMIM:605850); Dimethylglycine dehydrogenase deficiency (ORPHA:243343)
DMP1	Hypophosphatemic rickets, autosomal recessive (OMIM:241520); Autosomal recessive hypophosphatemic rickets (ORPHA:289176)
DMPK	Myotonic dystrophy 1 (OMIM:160900); ORPHA:589833; ORPHA:589830; ORPHA:589827; Congenital-onset Steinert myotonic dystrophy (ORPHA:589821); ORPHA:589824
DMRT1	46,XY complete gonadal dysgenesis (ORPHA:242)
DMXL1	—
DNAAF1	Ciliary dyskinesia, primary, 13 (OMIM:613193); Primary ciliary dyskinesia (ORPHA:244)
DNAAF11	Ciliary dyskinesia, primary, 19 (OMIM:614935); Primary ciliary dyskinesia (ORPHA:244)
DNAAF2	Ciliary dyskinesia, primary, 10 (OMIM:612518); Primary ciliary dyskinesia (ORPHA:244)
DNAAF3	Ciliary dyskinesia, primary, 2 (OMIM:606763); Primary ciliary dyskinesia (ORPHA:244)
DNAAF4	Ciliary dyskinesia, primary, 25 (OMIM:615482); Dyslexia, susceptibility to, 1 (OMIM:127700); Primary ciliary dyskinesia (ORPHA:244)
DNAAF5	Ciliary dyskinesia, primary, 18 (OMIM:614874); Primary ciliary dyskinesia (ORPHA:244)
DNAH11	Ciliary dyskinesia, primary, 7 (OMIM:611884); Primary ciliary dyskinesia (ORPHA:244)
DNAH2	Spermatogenic failure 45 (OMIM:619094)
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus (OMIM:608644); Primary ciliary dyskinesia (ORPHA:244)
DNAH9	Ciliary dyskinesia, primary, 40 (OMIM:618300); ORPHA:101063; Primary ciliary dyskinesia (ORPHA:244); ORPHA:157769
DNAI1	Ciliary dyskinesia, primary, 1 (OMIM:244400); Primary ciliary dyskinesia (ORPHA:244)
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus (OMIM:612444); Primary ciliary dyskinesia (ORPHA:244)
DNAJB1	ORPHA:401920
DNAJB2	Spinal muscular atrophy, distal, autosomal recessive, 5 (OMIM:614881); ORPHA:443950; ORPHA:314485

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
DNAJB6	Muscular dystrophy, limb-girdle, type 1E (OMIM:603511); DNAJB6-related limb-girdle muscular dystrophy D1 (ORPHA:34516); ORPHA:708126
DNAJC17	—
DNAJC19	3-methylglutaconic aciduria, type V (OMIM:610198); Dilated cardiomyopathy with ataxia (ORPHA:66634)
DNAJC30	Leber hereditary optic neuropathy (ORPHA:104); Williams syndrome (ORPHA:904)
DNAJC5	Ceroid lipofuscinosis, neuronal, 4B, autosomal dominant (OMIM:162350); ORPHA:228343
DNAJC6	Parkinson disease 19a, juvenile-onset (OMIM:615528); Young-onset Parkinson disease (ORPHA:2828); Atypical juvenile parkinsonism (ORPHA:391411)
DNAJC9	—
DNAL1	Ciliary dyskinesia, primary, 16 (OMIM:614017); Primary ciliary dyskinesia (ORPHA:244)
DNASE1	Systemic lupus erythematosus (OMIM:152700); ORPHA:300345; Systemic lupus erythematosus (ORPHA:536)
DNASE1L1	—
DNASE1L3	Systemic lupus erythematosus 16 (OMIM:614420); ORPHA:300345; Hypocomplementemic urticarial vasculitis (ORPHA:36412)
DNASE2	Autoinflammatory-pancytopenia syndrome (OMIM:619858)
DNM1	Epileptic encephalopathy, early infantile, 31 (OMIM:616346); Developmental and epileptic encephalopathy 31B, autosomal recessive (OMIM:620352); Lennox-Gastaut syndrome (ORPHA:2382); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
DNM1L	Encephalopathy due to defective mitochondrial and peroxisomal fission 1 (OMIM:614388); Optic atrophy 5 (OMIM:610708); DNM1L-related encephalopathy due to mitochondrial and peroxisomal fission defect (ORPHA:330050); Autosomal dominant optic atrophy, classic form (ORPHA:98673)
DNM2	Lethal congenital contracture syndrome 5 (OMIM:615368); Centronuclear myopathy 1 (OMIM:160150); Charcot-Marie-Tooth disease, dominant intermediate B (OMIM:606482); ORPHA:228179; Autosomal dominant centronuclear myopathy (ORPHA:169189); ORPHA:363409; ORPHA:100044
DNMT1	Neuropathy, hereditary sensory, type IE (OMIM:614116); Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant (OMIM:604121); ORPHA:456318; Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome (ORPHA:314404)
DNMT3A	Tatton-Brown-Rahman syndrome (OMIM:615879); Heyn-Sproul-Jackson syndrome (OMIM:618724); Leukemia, acute myeloid (OMIM:601626); ORPHA:658595; ORPHA:86845; Tatton-Brown-Rahman syndrome (ORPHA:404443); Sporadic pheochromocytoma/secreting paraganglioma (ORPHA:276621)
DNMT3B	Facioscapulohumeral muscular dystrophy 4, digenic (OMIM:619478); Immunodeficiency-Centromeric instability-facial anomalies syndrome (OMIM:242860); Facioscapulohumeral dystrophy (ORPHA:269); ICF syndrome (ORPHA:2268)
DNMT3L	—
DOC2A	—
DOCK3	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia (OMIM:618292); ORPHA:528084
DOCK4	—
DOCK6	Adams-Oliver syndrome 2 (OMIM:614219); Adams-Oliver syndrome (ORPHA:974)
DOCK7	Developmental and epileptic encephalopathy 23 (OMIM:615859); Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome (ORPHA:411986)
DOCK8	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections (OMIM:243700); ORPHA:178469; Combined immunodeficiency due to DOCK8 deficiency (ORPHA:217390)
DOCK9	—
DOK1	—
DOK2	—
DOK5	—
DOK7	Myasthenia, limb-girdle, familial (OMIM:254300); Fetal akinesia deformation sequence 3 (OMIM:618389); Fetal akinesia deformation sequence (ORPHA:994); Postsynaptic congenital myasthenic syndrome (ORPHA:98913)
DOLK	Congenital disorder of glycosylation, type Im (OMIM:610768); DK1-CDG (ORPHA:91131); Familial isolated dilated cardiomyopathy (ORPHA:154)
DOLPP1	—
DOT1L	Nil-Deshwar neurodevelopmental syndrome (OMIM:621265)
DPAGT1	Congenital disorder of glycosylation, type Ij (OMIM:608093); Myasthenic syndrome, congenital, 13, with tubular aggregates (OMIM:614750); Congenital myasthenic syndrome with glycosylation defect (ORPHA:353327); DPAGT1-CDG (ORPHA:86309)
DPCD	—
DPM1	Congenital disorder of glycosylation, type IE (OMIM:608799); DPM1-CDG (ORPHA:79322)
DPM2	Congenital disorder of glycosylation, type Iu (OMIM:615042); Congenital muscular dystrophy with intellectual disability and severe epilepsy (ORPHA:329178)
DPM3	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15 (OMIM:612937); Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15 (OMIM:618992); DPM3-CDG (ORPHA:263494)
DPP6	Ventricular fibrillation, paroxysmal familial, 2 (OMIM:612956); Intellectual developmental disorder, autosomal dominant 33 (OMIM:616311); ORPHA:228140; Autosomal dominant primary microcephaly (ORPHA:2514)
DPY19L2	Spermatogenic failure 9 (OMIM:613958); ORPHA:171709
DPYD	Dihydropyrimidine dehydrogenase deficiency (OMIM:274270); 1p21.3 microdeletion syndrome (ORPHA:293948); Dihydropyrimidine dehydrogenase deficiency (ORPHA:1675)
DPYS	Dihydropyrimidinuria (OMIM:222748); Dihydropyrimidinuria (ORPHA:38874)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
DQB1	—
DRD1	—
DRD2	Myoclonus-dystonia syndrome (ORPHA:36899)
DRD3	Tremor, hereditary essential, 1 (OMIM:190300); Schizophrenia (OMIM:181500)
DRD4	Attention deficit-hyperactivity disorder (OMIM:143465)
DRD5	Blepharospasm, benign essential, susceptibility to (OMIM:606798); Attention deficit-hyperactivity disorder (OMIM:143465)
DROSHA	—
DSC2	Arrhythmogenic right ventricular dysplasia, familial, 11 (OMIM:610476); ORPHA:293888; ORPHA:293899; ORPHA:293910
DSC3	Hypotrichosis and recurrent skin vesicles (OMIM:613102); ORPHA:217407
DSCAM	—
DSCAS	—
DSE	Ehlers-Danlos syndrome, musculocontractural type, 2 (OMIM:615539); Musculocontractural Ehlers-Danlos syndrome (ORPHA:2953)
DSG1	Keratosis palmoplantaris striata I (OMIM:148700); Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper-ige (OMIM:615508); ORPHA:369999; ORPHA:369992; Striate palmoplantar keratoderma (ORPHA:50942); ORPHA:370002
DSG2	Cardiomyopathy, dilated, 1BB (OMIM:612877); Arrhythmogenic right ventricular dysplasia, familial, 10 (OMIM:610193); Familial isolated dilated cardiomyopathy (ORPHA:154); ORPHA:293888; ORPHA:293899; ORPHA:293910
DSG3	Blistering, acantholytic, of oral and laryngeal mucosa (OMIM:619226)
DSG4	Hypotrichosis 6 (OMIM:607903); Hypotrichosis simplex (ORPHA:55654); Monilethrix (ORPHA:573)
DSP	Keratosis palmoplantaris striata II (OMIM:612908); Cardiomyopathy, dilated, with woolly hair and keratoderma (OMIM:605676); Epidermolysis bullosa, lethal acantholytic (OMIM:609638); Cardiomyopathy, dilated, with woolly hair, keratoderma, and tooth agenesis (OMIM:615821); Arrhythmogenic right ventricular dysplasia, familial, 8 (OMIM:607450); ORPHA:369992; Lethal acantholytic erosive disorder (ORPHA:158687); Idiopathic pulmonary fibrosis (ORPHA:2032); Familial isolated dilated cardiomyopathy (ORPHA:154); ORPHA:293888; ORPHA:293899; Carvajal syndrome (ORPHA:65282); ORPHA:476096; Striate palmoplantar keratoderma (ORPHA:50942); ORPHA:293910; ORPHA:293165
DSPP	Deafness, autosomal dominant 39, with dentinogenesis imperfecta 1 (OMIM:605594); Dentinogenesis imperfecta, shields type III (OMIM:125500); Dentin dysplasia, type II (OMIM:125420); Dentinogenesis imperfecta 1 (OMIM:125490); ORPHA:166260; ORPHA:166265; ORPHA:99791; ORPHA:99789
DST	OMIM:621511; OMIM:621510; Epidermolysis bullosa simplex, autosomal recessive 2 (OMIM:615425); Neuropathy, hereditary sensory and autonomic, type VI (OMIM:614653); ORPHA:314381; ORPHA:412181
DT	—
DTNA	Left ventricular noncompaction 1 (OMIM:604169); Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis 2 (OMIM:620971); ORPHA:54260
DTNBP1	Hermansky-Pudlak syndrome 7 (OMIM:614076); ORPHA:231531
DUOX2	Thyroid dyshormonogenesis 6 (OMIM:607200); Genetic transient congenital hypothyroidism (ORPHA:226316); Familial thyroid dyshormonogenesis (ORPHA:95716)
DUOXA1	—
DUOXA2	Thyroid hormonogenesis, genetic defect in, 5 (OMIM:274900); Familial thyroid dyshormonogenesis (ORPHA:95716)
DUS4L	—
DUX4	Facioscapulohumeral dystrophy (ORPHA:269)
DVL2	—
DXO	—
DYM	Smith-Mccort dysplasia 1 (OMIM:607326); Dyggve-Melchior-Clausen disease (OMIM:223800); Dyggve-Melchior-Clausen disease (ORPHA:239); ORPHA:178355
DYNAP	—
DYNC1H1	Charcot-marie-tooth disease, axonal, type 2O (OMIM:614228); Spinal muscular atrophy, lower extremity-predominant, 1, autosomal dominant (OMIM:158600); Cortical dysplasia, complex, with other brain malformations 13 (OMIM:614563); ORPHA:178469; ORPHA:284232; ORPHA:209341
DYNC2H1	Asphyxiating thoracic dystrophy 3 (OMIM:613091); ORPHA:93270; Short rib-polydactyly syndrome, Verma-Naumoff type (ORPHA:93271); ORPHA:93269; Jeune syndrome (ORPHA:474)
DYNLT2	—
DYRK1A	Intellectual developmental disorder, autosomal dominant 7 (OMIM:614104); Intellectual disability syndrome due to a DYRK1A point mutation (ORPHA:464311); DYRK1A-related intellectual disability syndrome due to 21q22.13q22.2 microdeletion (ORPHA:268261)
DYSF	Miyoshi muscular dystrophy 1 (OMIM:254130); Muscular dystrophy, limb-girdle, type 2B (OMIM:253601); Myopathy, distal, with anterior tibial onset (OMIM:606768); ORPHA:199329; Distal myopathy with anterior tibial onset (ORPHA:178400); Dysferlin-related limb-girdle muscular dystrophy R2 (ORPHA:268); Miyoshi myopathy (ORPHA:45448)
E2F1	—
E2F4	—
E2F5	—
EARS2	Combined oxidative phosphorylation deficiency 12 (OMIM:614924); ORPHA:314051
EBAG9	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
EBF1	—
EBF4	—
EBLN3P	—
EBNA1BP2	—
EBP	Mend syndrome (OMIM:300960); Chondrodysplasia punctata 2, X-linked dominant (OMIM:302960); MEND syndrome (ORPHA:401973); X-linked dominant chondrodysplasia punctata (ORPHA:35173)
ECE1	Hypertension, essential (OMIM:145500); Hirschsprung disease, cardiac defects, and autonomic dysfunction (OMIM:613870); Hirschsprung disease (ORPHA:388)
ECE2	—
ECI1	—
ECM1	Urbach-Wiethe disease (OMIM:247100); Lipoid proteinosis (ORPHA:530)
ECM2	—
ECSIT	—
ECT2L	—
EDA	Ectodermal dysplasia 1, hypohidrotic, X-linked (OMIM:305100); Tooth agenesis, selective, X-linked, 1 (OMIM:313500); X-linked hypohidrotic ectodermal dysplasia (ORPHA:181); Oligodontia (ORPHA:99798)
EDA2R	X-linked hypohidrotic ectodermal dysplasia (ORPHA:181)
EDAR	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant (OMIM:129490); Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive (OMIM:224900); OMIM:612630; Autosomal recessive hypohidrotic ectodermal dysplasia (ORPHA:248); Autosomal dominant hypohidrotic ectodermal dysplasia (ORPHA:1810)
EDARADD	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant (OMIM:129490); Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive (OMIM:224900); Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant (OMIM:614940); Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive (OMIM:614941); Autosomal recessive hypohidrotic ectodermal dysplasia (ORPHA:248); Autosomal dominant hypohidrotic ectodermal dysplasia (ORPHA:1810); Oligodontia (ORPHA:99798)
EDN1	Question mark ears, isolated (OMIM:612798); Auriculocondylar syndrome 3 (OMIM:615706); Auriculocondylar syndrome (ORPHA:137888)
EDN3	Waardenburg syndrome, type 4B (OMIM:613265); Hirschsprung disease, susceptibility to, 4 (OMIM:613712); Hirschsprung disease (ORPHA:388); Waardenburg-Shah syndrome (ORPHA:897); Congenital central hypoventilation syndrome (ORPHA:661)
EDNRA	Mandibulofacial dysostosis with alopecia (OMIM:616367); Migraine with or without aura, susceptibility to, 1 (OMIM:157300); ORPHA:443995; Cystic fibrosis (ORPHA:586)
EDNRB	Hirschsprung disease, susceptibility to, 2 (OMIM:600155); Waardenburg-Shah syndrome (OMIM:277580); ABCD syndrome (OMIM:600501); Hirschsprung disease (ORPHA:388); Waardenburg-Shah syndrome (ORPHA:897); Waardenburg syndrome type 2 (ORPHA:895)
EEF1A1	—
EEF1AKMT4	—
EEF1E1	—
EEF2KMT	—
EFCAB5	—
EFEMP1	Doyne honeycomb retinal dystrophy (OMIM:126600); Glaucoma 1, open angle, H (OMIM:611276); Cutis laxa, autosomal recessive, type ID (OMIM:620780); Familial drusen (ORPHA:75376); Juvenile glaucoma (ORPHA:98977)
EFEMP2	Cutis laxa, autosomal recessive, type IB (OMIM:614437); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); Autosomal recessive cutis laxa type 1 (ORPHA:90349); ORPHA:314718
EFHC1	Epilepsy, juvenile absence (OMIM:607631); Myoclonic epilepsy, juvenile, susceptibility to, 1 (OMIM:254770); Juvenile myoclonic epilepsy (ORPHA:307); Juvenile absence epilepsy (ORPHA:1941)
EFHC2	—
EFNA5	—
EFNB1	Craniofrontonasal syndrome (OMIM:304110); Craniofrontonasal dysplasia (ORPHA:1520)
EFR3A	—
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type (OMIM:610536); Mandibulofacial dysostosis-microcephaly syndrome (ORPHA:79113)
EGF	Hypomagnesemia 4, renal (OMIM:611718); ORPHA:210159; ORPHA:620368
EGFR	Lung cancer, susceptibility to (OMIM:211980); Inflammatory skin and bowel disease, neonatal, 2 (OMIM:616069); ORPHA:251579; ORPHA:251576
EGLN1	OMIM:609070; Erythrocytosis, familial, 3 (OMIM:609820); ORPHA:247511
EGR1	—
EGR2	Charcot-Marie-Tooth disease, demyelinating, type 1D (OMIM:607678); Hypertrophic neuropathy of dejerine-sottas (OMIM:145900); Neuropathy, congenital hypomyelinating, 1, autosomal recessive (OMIM:605253); ORPHA:101084; ORPHA:99951; ORPHA:64748
EHBP1	OMIM:611868
EHD2	—
EHHADH	Fanconi renotubular syndrome 3 (OMIM:615605); ORPHA:300; Primary Fanconi renotubular syndrome (ORPHA:3337)
EHMT1	Kleefstra syndrome 1 (OMIM:610253); Kleefstra syndrome due to a point mutation (ORPHA:261652); Kleefstra syndrome due to 9q34 microdeletion (ORPHA:96147)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
EIF2AK3	Epiphyseal dysplasia, multiple, with early-onset diabetes mellitus (OMIM:226980); Wolcott-Rallison syndrome (ORPHA:1667)
EIF2B1	Leukoencephalopathy with vanishing white matter (OMIM:603896); ORPHA:99853; ORPHA:99854; ORPHA:157716; ORPHA:157719; ORPHA:157713
EIF2B2	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure (OMIM:620312); ORPHA:99853; ORPHA:99854; ORPHA:157716; ORPHA:157719; ORPHA:157713
EIF2B3	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure (OMIM:620313); ORPHA:99853; ORPHA:99854; ORPHA:157716; ORPHA:157719; ORPHA:157713
EIF2B4	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure (OMIM:620314); ORPHA:99853; ORPHA:99854; ORPHA:157716; ORPHA:157719; ORPHA:157713
EIF2B5	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure (OMIM:620315); ORPHA:99853; ORPHA:99854; ORPHA:157716; ORPHA:157719; ORPHA:157713
EIF2D	—
EIF2S3	Mehmo syndrome (OMIM:300148); MEHMO syndrome (ORPHA:85282)
EIF4E	OMIM:615091
EIF4G1	Parkinson disease 18 (OMIM:614251); Hereditary late-onset Parkinson disease (ORPHA:411602)
EIF4H	Williams syndrome (ORPHA:904)
ELAC2	Combined oxidative phosphorylation deficiency 17 (OMIM:615440); OMIM:614731; ORPHA:1331; ORPHA:369913
ELANE	Cyclic neutropenia (OMIM:162800); Neutropenia, severe congenital, 1, autosomal dominant (OMIM:202700); Cyclic neutropenia (ORPHA:2686); Autosomal dominant severe congenital neutropenia (ORPHA:486)
ELF1	—
ELF4	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2 (OMIM:301074); ORPHA:676125; ORPHA:632
ELK1	—
ELK3	—
ELL	—
ELMOD2	—
ELN	Supravalvular aortic stenosis (OMIM:185500); Cutis laxa, autosomal dominant 1 (OMIM:123700); Williams-Beuren syndrome (OMIM:194050); Supravalvular aortic stenosis (ORPHA:3193); Williams syndrome (ORPHA:904); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); Autosomal dominant cutis laxa (ORPHA:90348)
ELOVL4	Spinocerebellar ataxia 34 (OMIM:133190); Stargardt disease 3 (OMIM:600110); Ichthyosis, spastic quadriplegia, and impaired intellectual development (OMIM:614457); ORPHA:352333; Stargardt disease (ORPHA:827); Spinocerebellar ataxia type 34 (ORPHA:1955)
ELP1	Neuropathy, hereditary sensory and autonomic, type III (OMIM:223900); MEDULLOBLASTOMA (OMIM:155255); Familial dysautonomia (ORPHA:1764)
ELP2	Intellectual developmental disorder, autosomal recessive 58 (OMIM:617270)
ELP4	Aniridia 2 (OMIM:617141)
EMC4	—
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked (OMIM:310300); Cardiomyopathy, dilated, 3C (OMIM:301163); X-linked Emery-Dreifuss muscular dystrophy (ORPHA:98863)
EME1	—
EMG1	Bowen-Conradi syndrome (OMIM:211180); Bowen-Conradi syndrome (ORPHA:1270)
EMX1	—
EMX2	SCHIZENCEPHALY (OMIM:269160); ORPHA:485275
EMX2OS	—
EN2	—
ENAM	Amelogenesis imperfecta, type IB (OMIM:104500); Amelogenesis imperfecta, type IC (OMIM:204650); ORPHA:100031
ENG	Telangiectasia, hereditary hemorrhagic, type 1 (OMIM:187300); Generalized juvenile polyposis/juvenile polyposis coli (ORPHA:329971); ORPHA:275777; Hereditary hemorrhagic telangiectasia (ORPHA:774); Familial cerebral saccular aneurysm (ORPHA:231160)
ENO3	Glycogen storage disease XIII (OMIM:612932); ORPHA:99849
ENOX1	—
ENPP1	Diabetes mellitus, noninsulin-dependent (OMIM:125853); Cole disease (OMIM:615522); Hypophosphatemic rickets, autosomal recessive, 2 (OMIM:613312); Arterial calcification, generalized, of infancy, 1 (OMIM:208000); OBESITY (OMIM:601665); ORPHA:324561; Pseudoxanthoma elasticum (ORPHA:758); Autosomal recessive hypophosphatemic rickets (ORPHA:289176); Generalized arterial calcification of infancy (ORPHA:51608)
ENTPD1	Spastic paraplegia 64, autosomal recessive (OMIM:615683); Autosomal recessive spastic paraplegia type 64 (ORPHA:401810)
ENTPD5	—
ENTPD7	—
EOMES	Microcephaly-polymicrogyria-corpus callosum agenesis syndrome (ORPHA:171703)
EP300	Rubinstein-Taybi syndrome 1 (OMIM:180849); Menke-Hennekam syndrome 2 (OMIM:618333); Colorectal cancer (OMIM:114500); Rubinstein-Taybi syndrome 2 (OMIM:613684); Rubinstein-Taybi syndrome due to EP300 haploinsufficiency (ORPHA:353284)
EPAS1	Erythrocytosis, familial, 4 (OMIM:611783); ORPHA:324299; Sporadic pheochromocytoma/secreting paraganglioma (ORPHA:276621); ORPHA:247511

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
EPB41	Elliptocytosis 1 (OMIM:611804); Hereditary elliptocytosis (ORPHA:288)
EPB41L1	Chromosome 20q11-q12 deletion syndrome (OMIM:614257); ORPHA:178469
EPB41L2	—
EPB42	Spherocytosis, type 5 (OMIM:612690); Hereditary spherocytosis (ORPHA:822)
EPC2	—
EPCAM	Diarrhea 5, with tufting enteropathy, congenital (OMIM:613217); Colorectal cancer, hereditary nonpolyposis, type 8 (OMIM:613244); Lynch syndrome (ORPHA:144); Congenital tufting enteropathy (ORPHA:92050)
EPHA2	Cataract, posterior polar, 1 ctpa cataract, congenital total, included (OMIM:116600); ORPHA:441447; ORPHA:98994; ORPHA:98991; ORPHA:98993
EPHA3	—
EPHA5	—
EPHA7	—
EPHB2	Prostate cancer/brain cancer susceptibility (OMIM:603688); Bleeding disorder, platelet-type, 22 (OMIM:618462); ORPHA:1331
EPHB6	—
EPHX1	ORPHA:238475
EPHX2	Hypercholesterolemia, familial, 1 (OMIM:143890)
EPM2A	Myoclonic epilepsy of Lafora (OMIM:254780); Lafora disease (ORPHA:501)
EPN2	—
EPO	Erythrocytosis, familial, 5 (OMIM:617907); Diamond-Blackfan anemia-like (OMIM:617911); OMIM:612623; ORPHA:247511
EPOR	Erythrocytosis, familial, 1 (OMIM:133100); Primary familial polycythemia (ORPHA:90042)
EPS8L2	Deafness, autosomal recessive 106 (OMIM:617637); ORPHA:90636
EPX	Eosinophil peroxidase deficiency (OMIM:261500)
ERAP1	Behçet disease (ORPHA:117)
ERAP2	—
ERBB2	Glioma susceptibility 1 (OMIM:137800); Visceral neuropathy, familial, 2, autosomal recessive (OMIM:619465); Ovarian cancer (OMIM:167000); Lung cancer, susceptibility to (OMIM:211980); Gastric cancer, somatic (OMIM:613659); Hirschsprung disease (ORPHA:388); ORPHA:2800; Adenocarcinoma of the oesophagus and oesophagogastric junction (ORPHA:99976); ORPHA:213726
ERBB3	Visceral neuropathy, familial, autosomal recessive (OMIM:243180); Erythroleukemia, familial, susceptibility to (OMIM:133180); Lethal congenital contracture syndrome 2 (OMIM:607598); Hirschsprung disease (ORPHA:388); ORPHA:137776
ERBB4	Amyotrophic lateral sclerosis 19 (OMIM:615515); ORPHA:178469; Amyotrophic lateral sclerosis (ORPHA:803)
ERCC1	Cerebrooculofacioskeletal syndrome 4 (OMIM:610758); COFS syndrome (ORPHA:1466); Cockayne syndrome type 2 (ORPHA:90322)
ERCC2	Cerebrooculofacioskeletal syndrome 2 (OMIM:610756); Xeroderma pigmentosum, complementation group D (OMIM:278730); Trichothiodystrophy 1, photosensitive (OMIM:601675); COFS syndrome (ORPHA:1466); Trichothiodystrophy (ORPHA:33364); Xeroderma pigmentosum-Cockayne syndrome complex (ORPHA:220295); Xeroderma pigmentosum (ORPHA:910)
ERCC3	Trichothiodystrophy 2, photosensitive (OMIM:616390); Xeroderma pigmentosum, complementation group B (OMIM:610651); Trichothiodystrophy (ORPHA:33364); Xeroderma pigmentosum-Cockayne syndrome complex (ORPHA:220295); Xeroderma pigmentosum (ORPHA:910)
ERCC4	Fanconi anemia, complementation group Q (OMIM:615272); XFE progeroid syndrome (OMIM:610965); Xeroderma pigmentosum, complementation group F (OMIM:278760); Fanconi anemia (ORPHA:84); Xeroderma pigmentosum-Cockayne syndrome complex (ORPHA:220295); Xeroderma pigmentosum (ORPHA:910); Cockayne syndrome type 1 (ORPHA:90321)
ERCC5	Cerebrooculofacioskeletal syndrome 3 (OMIM:616570); Xeroderma pigmentosum, complementation group G (OMIM:278780); COFS syndrome (ORPHA:1466); Xeroderma pigmentosum-Cockayne syndrome complex (ORPHA:220295); Xeroderma pigmentosum (ORPHA:910)
ERCC6	Premature ovarian failure 11 (OMIM:616946); Cockayne syndrome, type B (OMIM:133540); De Sanctis-Cacchione syndrome (OMIM:278800); UV-sensitive syndrome 1 (OMIM:600630); Lung cancer, susceptibility to (OMIM:211980); Cerebrooculofacioskeletal syndrome 1 (OMIM:214150); OMIM:613761; COFS syndrome (ORPHA:1466); ORPHA:178338; Cockayne syndrome type 3 (ORPHA:90324); Cockayne syndrome type 1 (ORPHA:90321); Cockayne syndrome type 2 (ORPHA:90322)
ERCC8	Cockayne syndrome, type A (OMIM:216400); UV-sensitive syndrome 2 (OMIM:614621); ORPHA:178338; Cockayne syndrome type 3 (ORPHA:90324); Cockayne syndrome type 1 (ORPHA:90321); Cockayne syndrome type 2 (ORPHA:90322)
ERI2	—
ERICH6B	—
ERLIN2	Spastic paraplegia 18A, autosomal dominant (OMIM:620512); Spastic paraplegia 18, autosomal recessive (OMIM:611225); Autosomal spastic paraplegia type 18 (ORPHA:209951); Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome (ORPHA:280384); Juvenile primary lateral sclerosis (ORPHA:247604)
ERMAP	OMIM:111750; OMIM:111620
ERMP1	—
ERRFI1	—
ESCO2	Roberts-SC phocomelia syndrome (OMIM:268300); Cleft lip/palate with abnormal thumbs and microcephaly (OMIM:216100); Roberts syndrome (ORPHA:3103); Juberg-Hayward syndrome (ORPHA:2319)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
ESPN	Deafness, autosomal recessive 36, with or without vestibular involvement (OMIM:609006); OMIM:621556; Usher syndrome, type 1M (OMIM:618632); ORPHA:90636; Usher syndrome type 1 (ORPHA:231169)
ESR1	OMIM:608446; Migraine with or without aura, susceptibility to, 1 (OMIM:157300); Estrogen resistance (OMIM:615363); Breast cancer (OMIM:114480); Estrogen resistance syndrome (ORPHA:785)
ESR2	Ovarian dysgenesis 8 (OMIM:618187); ORPHA:99361
ESRG	—
ESRRB	Deafness, autosomal recessive 35 (OMIM:608565); ORPHA:90636
ESS2	Velocardiofacial syndrome (OMIM:192430)
ETFA	Multiple acyl-CoA dehydrogenation deficiency (OMIM:231680); ORPHA:394532; ORPHA:394529
ETFB	Multiple acyl-CoA dehydrogenation deficiency (OMIM:231680); ORPHA:394532; ORPHA:394529
ETFDH	Multiple acyl-CoA dehydrogenation deficiency (OMIM:231680); ORPHA:394532; ORPHA:394529
ETHE1	Encephalopathy, ethylmalonic (OMIM:602473); Ethylmalonic encephalopathy (ORPHA:51188)
ETNK1	—
ETV4	ORPHA:319
ETV6	Thrombocytopenia 5 (OMIM:616216); Leukemia, acute myeloid (OMIM:601626); ORPHA:314950; ORPHA:2030; ORPHA:146; ORPHA:585929; ORPHA:98823; ORPHA:2665; ORPHA:168629
EVC	Ellis-Van creveld syndrome (OMIM:225500); Weyers acrofacial dysostosis (OMIM:193530); Ellis Van Creveld syndrome (ORPHA:289); Acrofacial dysostosis, Weyers type (ORPHA:952)
EVC2	Ellis-Van creveld syndrome (OMIM:225500); Weyers acrofacial dysostosis (OMIM:193530); Ellis Van Creveld syndrome (ORPHA:289); Acrofacial dysostosis, Weyers type (ORPHA:952)
EVI2A	—
EVI5	—
EWSR1	Ewing sarcoma (OMIM:612219); ORPHA:319; Desmoplastic small round cell tumor (ORPHA:83469); ORPHA:97338; ORPHA:209916; ORPHA:370334; ORPHA:569164
EXO1	—
EXO5	—
EXOC4	—
EXOSC3	Pontocerebellar hypoplasia, type 1B (OMIM:614678); Pontocerebellar hypoplasia type 1 (ORPHA:2254)
EXOSC7	—
EXOSC8	Pontocerebellar hypoplasia, type 1C (OMIM:616081); Pontocerebellar hypoplasia type 1 (ORPHA:2254)
EXPH5	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive (OMIM:615028); ORPHA:412189
EXT1	Chondrosarcoma (OMIM:215300); Exostoses, multiple, type I (OMIM:133700); Multiple osteochondromas (ORPHA:321); ORPHA:55880; Trichorhinophalangeal syndrome type 2 (ORPHA:502)
EXT2	Seizures, scoliosis, and macrocephaly/microcephaly syndrome (OMIM:616682); Exostoses, multiple, type II (OMIM:133701); Multiple osteochondromas (ORPHA:321); Potocki-Shaffer syndrome (ORPHA:52022); Seizures-scoliosis-macrocephaly syndrome (ORPHA:466926)
EXTL1	—
EXTL2	—
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalitie (OMIM:617425); Skeletal dysplasia-T-cell immunodeficiency-developmental delay syndrome (ORPHA:508533)
EYA1	Otofaciocervical syndrome (OMIM:166780); Branchiootic syndrome 1 (OMIM:602588); Branchiootorenal syndrome 1 (OMIM:113650); Otofaciocervical syndrome (ORPHA:2792); Branchiootic syndrome (ORPHA:52429); BOR syndrome (ORPHA:107)
EYA4	Deafness, autosomal dominant nonsyndromic sensorineural 10 (OMIM:601316); Cardiomyopathy, dilated, 1J (OMIM:605362); ORPHA:90635; Sensorineural deafness with dilated cardiomyopathy (ORPHA:217622)
EYS	Retinitis pigmentosa 25 (OMIM:602772); Retinitis pigmentosa (ORPHA:791)
EZH2	Weaver syndrome (OMIM:277590); Weaver syndrome (ORPHA:3447)
F10	Factor X deficiency (OMIM:227600); Congenital factor X deficiency (ORPHA:328)
F11	Factor XI deficiency (OMIM:612416); Congenital factor XI deficiency (ORPHA:329)
F12	Angioedema, hereditary, 3 (OMIM:610618); Factor XII deficiency (OMIM:234000); Congenital factor XII deficiency (ORPHA:330); ORPHA:617919; ORPHA:100054
F13A1	Thrombophiliavenous thromboembolism, included (OMIM:188050); OMIM:608446; Factor XIII, A subunit, deficiency of (OMIM:613225); Congenital factor XIII deficiency (ORPHA:331)
F13B	Factor XIIIB deficiency (OMIM:613235); Congenital factor XIII deficiency (ORPHA:331)
F2	Thrombophiliavenous thromboembolism, included (OMIM:188050); Pregnancy loss, recurrent, susceptibility to, 2 (OMIM:614390); Ischemic stroke, susceptibility to (OMIM:601367); Congenital prothrombin deficiency (OMIM:613679); Congenital factor II deficiency (ORPHA:325); ORPHA:329217
F2RL1	—
F5	Thrombophilia due to deficiency of activated protein C cofactor (OMIM:188055); Budd-Chiari syndrome (OMIM:600880); Pregnancy loss, recurrent, susceptibility to, 1 (OMIM:614389); Ischemic stroke, susceptibility to (OMIM:601367); Factor V deficiency (OMIM:227400); Congenital factor V deficiency (ORPHA:326); ORPHA:329217; Budd-Chiari syndrome (ORPHA:131); ORPHA:599579; ORPHA:391320; ORPHA:600194
F7	OMIM:608446; Factor VII deficiency (OMIM:227500); Congenital factor VII deficiency (ORPHA:327)
F8	Thrombophilia 13, X-linked, due to factor VIII defect (OMIM:301071); Hemophilia A (OMIM:306700); Bleeding disorder in hemophilia A carriers (ORPHA:177926); Moderate hemophilia A (ORPHA:169805); ORPHA:169808; Severe hemophilia A (ORPHA:169802)
F9	OMIM:301052; Hemophilia B (OMIM:306900); Thrombophilia, X-linked, due to factor IX defect (OMIM:300807); ORPHA:177929; ORPHA:169799; ORPHA:169793; ORPHA:169796

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
FA2H	Spastic paraplegia 35, autosomal recessive (OMIM:612319); Autosomal recessive spastic paraplegia type 35 (ORPHA:171629); Fatty acid hydroxylase-associated neurodegeneration (ORPHA:329308)
FAAH	OMIM:606581
FAAH2	—
FABP1	—
FABP2	—
FADD	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations (OMIM:613759); ORPHA:99806; FADD-related immunodeficiency (ORPHA:306550)
FAH	Tyrosinemia, type I (OMIM:276700); Tyrosinemia type 1 (ORPHA:882)
FAM120A	—
FAM120AOS	—
FAM131A	—
FAM161A	Retinitis pigmentosa 28 (OMIM:606068); Retinitis pigmentosa (ORPHA:791)
FAM161B	—
FAM20A	Amelogenesis imperfecta, type IG (enamel-renal syndrome) (OMIM:204690); Enamel-renal syndrome (ORPHA:1031)
FAM20C	Raine syndrome (OMIM:259775); Osteosclerotic bone dysplasia (ORPHA:1832)
FAM47B	—
FAM53C	—
FAM83H	—
FAM8A1	—
FAM91A1	—
FAN1	Interstitial nephritis, karyomegalic (OMIM:614817); ORPHA:401996
FANCA	Fanconi anemia (OMIM:227650); Fanconi anemia (ORPHA:84)
FANCB	Fanconi anemia, complementation group B (OMIM:300514); Fanconi anemia (ORPHA:84); VACTERL with hydrocephalus (ORPHA:3412)
FANCC	Fanconi anemia, complementation group C (OMIM:227645); Fanconi anemia (ORPHA:84)
FANCD2	Fanconi anemia, complementation group D2 (OMIM:227646); Fanconi anemia (ORPHA:84)
FANCD2OS	—
FANCE	Fanconi anemia, complementation group E (OMIM:600901); Fanconi anemia (ORPHA:84)
FANCF	Fanconi anemia, complementation group F (OMIM:603467); Fanconi anemia (ORPHA:84)
FANCG	Fanconi anemia, complementation group G (OMIM:614082); Fanconi anemia (ORPHA:84)
FANCI	Fanconi anemia, complementation group I (OMIM:609053); Fanconi anemia (ORPHA:84)
FANCL	Fanconi anemia, complementation group L (OMIM:614083); Fanconi anemia (ORPHA:84)
FANCM	Spermatogenic failure 28 (OMIM:618086); Premature ovarian failure 15 (OMIM:618096); Fanconi anemia (ORPHA:84); Male infertility with azoospermia or oligozoospermia due to single gene mutation (ORPHA:399805)
FARS2	Spastic paraplegia 77, autosomal recessive (OMIM:617046); Combined oxidative phosphorylation deficiency 14 (OMIM:614946); Autosomal recessive spastic paraplegia type 77 (ORPHA:466722); ORPHA:319519
FAS	Autoimmune lymphoproliferative syndrome (OMIM:601859); Autoimmune lymphoproliferative syndrome (ORPHA:3261); Behçet disease (ORPHA:117); Vogt-Koyanagi-Harada disease (ORPHA:3437)
FASLG	Lung cancer, susceptibility to (OMIM:211980); Autoimmune lymphoproliferative syndrome (OMIM:601859); Autoimmune lymphoproliferative syndrome (ORPHA:3261)
FASN	—
FASTKD2	Combined oxidative phosphorylation deficiency 44 (OMIM:618855); Mitochondrial complex IV deficiency, nuclear type 1 (OMIM:220110); ORPHA:166105
FAT3	—
FBH1	—
FBLN1	FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome (ORPHA:404451); ORPHA:295197
FBLN5	Macular degeneration, age-related, 3 (OMIM:608895); Cutis laxa, autosomal dominant 2 (OMIM:614434); Charcot-Marie-Tooth disease, demyelinating, type 1H (OMIM:619764); Cutis laxa, autosomal recessive, type IA (OMIM:219100); ORPHA:280598; Autosomal dominant cutis laxa (ORPHA:90348); Autosomal recessive cutis laxa type 1 (ORPHA:90349)
FBN1	Mass syndrome (OMIM:604308); Marfan syndrome (OMIM:154700); Ectopia lentis, familial (OMIM:129600); Marfan lipodystrophy syndrome (OMIM:616914); Geleophysic dysplasia 2 (OMIM:614185); Stiff skin syndrome (OMIM:184900); Weill-Marchesani syndrome 2, dominant (OMIM:608328); Acromicric dysplasia (OMIM:102370); Stiff skin syndrome (ORPHA:2833); ORPHA:300382; Acromicric dysplasia (ORPHA:969); Geleophysic dysplasia (ORPHA:2623); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); Isolated ectopia lentis (ORPHA:1885); ORPHA:284963; Neonatal Marfan syndrome (ORPHA:284979); Shprintzen-Goldberg syndrome (ORPHA:2462); Glaucoma-ectopia lentis-microspherophakia-stiff joints-short stature syndrome (ORPHA:2084); Weill-Marchesani syndrome (ORPHA:3449)
FBN2	Contractural arachnodactyly, congenital (OMIM:121050); Macular degeneration, early-onset (OMIM:616118); Congenital contractural arachnodactyly (ORPHA:115)
FBP1	Fructose-1,6-bisphosphatase deficiency (OMIM:229700); Fructose-1,6-bisphosphatase deficiency (ORPHA:348)
FBXL3	Intellectual developmental disorder with short stature, facial anomalies, and speech defects (OMIM:606220)
FBXL6	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
FBXO10	—
FBXO11	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities (OMIM:618089); ORPHA:528084
FBXO38	Neuronopathy, distal hereditary motor, type IID (OMIM:615575); ORPHA:139525
FBXO7	Parkinson disease 15, autosomal recessive (OMIM:260300); Parkinsonian-pyramidal syndrome (ORPHA:171695)
FBXW11	Neurodevelopmental, jaw, eye, and digital syndrome (OMIM:618914); ORPHA:528084
FBXW4	Isolated split hand-split foot malformation (ORPHA:2440)
FBXW7	Developmental delay, hypotonia, and impaired language (OMIM:620012); ORPHA:528084
FCER1G	—
FCGBP	—
FCGR2A	Cystic fibrosis (OMIM:219700); Systemic lupus erythematosus (OMIM:152700); OMIM:611162
FCGR2B	Systemic lupus erythematosus (OMIM:152700); OMIM:611162; Systemic lupus erythematosus (ORPHA:536)
FCN2	—
FCN3	Immunodeficiency due to ficolin 3 deficiency (OMIM:613860); ORPHA:331190
FDFT1	Squalene synthase deficiency (OMIM:618156)
FECH	Protoporphyria, erythropoietic, 1 (OMIM:177000); Autosomal erythropoietic protoporphyria (ORPHA:79278)
FEM1A	—
FEM1B	Neurodevelopmental disorder with behavioral, ear, and skeletal abnormalities (OMIM:621263)
FER1L5	—
FERMT1	Kindler syndrome (OMIM:173650); Kindler epidermolysis bullosa (ORPHA:2908)
FERMT3	Leukocyte adhesion deficiency, type III (OMIM:612840); ORPHA:99844
FEZF2	—
FFAR1	—
FFAR4	OMIM:607514
FGA	Congenital dysfibrinogenemia (OMIM:616004); Afibrinogenemia, congenitalhypofibrinogenemia, congenital, included (OMIM:202400); Amyloidosis, familial visceral (OMIM:105200); Familial hypofibrinogenemia (ORPHA:101041); Familial afibrinogenemia (ORPHA:98880); Familial dysfibrinogenemia (ORPHA:98881); ORPHA:248408; ORPHA:93562
FGB	Congenital dysfibrinogenemia (OMIM:616004); Afibrinogenemia, congenitalhypofibrinogenemia, congenital, included (OMIM:202400); Familial hypofibrinogenemia (ORPHA:101041); Familial afibrinogenemia (ORPHA:98880); Familial dysfibrinogenemia (ORPHA:98881); ORPHA:248408
FGD1	Aarskog-Scott syndrome (OMIM:305400); Aarskog-Scott syndrome (ORPHA:915)
FGD3	—
FGD4	Charcot-marie-tooth disease, type 4H (OMIM:609311); ORPHA:99954
FGF10	Aplasia of lacrimal and salivary glands (OMIM:180920); Lacrimoauriculodentodigital syndrome-3 (OMIM:620193); ORPHA:86815; Lacrimoauriculodentodigital syndrome (ORPHA:2363)
FGF14	Spinocerebellar ataxia 27B, late-onset (OMIM:620174); Nystagmus 4, congenital, autosomal dominant (OMIM:193003); Spinocerebellar ataxia type 27A (ORPHA:98764); ORPHA:675216
FGF2	—
FGF23	Tumoral calcinosis, hyperphosphatemic, familial, 2 (OMIM:617993); Hypophosphatemic rickets, autosomal dominant (OMIM:193100); ORPHA:306661; Autosomal dominant hypophosphatemic rickets (ORPHA:89937)
FGF3	Deafness, congenital, with inner ear agenesis, microtia, and microdontia (OMIM:610706); ORPHA:99806; Otodental syndrome (ORPHA:2791); Deafness with labyrinthine aplasia, microtia, and microdontia (ORPHA:90024)
FGF8	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Hypogonadotropic hypogonadism 6 with or without anosmia (OMIM:612702); Septopreoptic holoprosencephaly (ORPHA:280195); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925); Kallmann syndrome (ORPHA:478); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432)
FGF9	Multiple synostoses syndrome 3 (OMIM:612961); Multiple synostoses syndrome (ORPHA:3237)
FGFR1	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Encephalocraniocutaneous lipomatosis (OMIM:613001); Trigonocephaly 1 (OMIM:190440); Hypogonadotropic hypogonadism 2 with or without anosmia (OMIM:147950); Hartsfield syndrome (OMIM:615465); Osteoglophonic dysplasia (OMIM:166250); Jackson-Weiss syndrome (OMIM:123150); Pfeiffer syndrome (OMIM:101600); ORPHA:251579; ORPHA:251576; Septo-optic dysplasia spectrum (ORPHA:3157); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); ORPHA:168953; ORPHA:314950; Non-syndromic metopic craniosynostosis (ORPHA:3366); Lobar holoprosencephaly (ORPHA:93924); Osteoglosphonic dysplasia (ORPHA:2645); Encephalocraniocutaneous lipomatosis (ORPHA:2396); Pfeiffer syndrome type 1 (ORPHA:93258); Hartsfield syndrome (ORPHA:2117); Kallmann syndrome (ORPHA:478); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432); Oligodontia (ORPHA:99798); ORPHA:251615
FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis (OMIM:207410); Crouzon syndrome (OMIM:123500); Beare-Stevenson cutis gyrata syndrome (OMIM:123790); Familial scaphocephaly syndrome, Mcgillivray type (OMIM:609579); Bent bone dysplasia syndrome (OMIM:614592); Apert syndrome (OMIM:101200); Saethre-Chotzen syndrome (OMIM:101400); Lacrimoauriculodentodigital syndrome-1 (OMIM:149730); Gastric cancer, somatic (OMIM:613659); Jackson-Weiss syndrome (OMIM:123150); Pfeiffer syndrome (OMIM:101600); Apert syndrome (ORPHA:87); Crouzon syndrome (ORPHA:207); Cutis gyrata-

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	acanthosis nigricans-craniosynostosis syndrome (ORPHA:1555); Jackson-Weiss syndrome (ORPHA:1540); FGFR2-related bent bone dysplasia (ORPHA:313855); Familial scaphocephaly syndrome, McGillivray type (ORPHA:168624); Saethre-Chotzen syndrome (ORPHA:794); Pfeiffer syndrome type 3 (ORPHA:93260); Pfeiffer syndrome type 1 (ORPHA:93258); Pfeiffer syndrome type 2 (ORPHA:93259); Lacrimoauriculodentodigital syndrome (ORPHA:2363); ORPHA:596008
FGFR3	Hypochondroplasia (OMIM:146000); Epidermal nevus, somatic (OMIM:162900); Achondroplasia, severe, with developmental delay and acanthosis nigricans (OMIM:616482); Achondroplasia (OMIM:100800); Lacrimoauriculodentodigital syndrome-2 (OMIM:620192); Testicular tumor, somatic (OMIM:273300); Camptodactyly, tall stature, and hearing loss syndrome (OMIM:610474); Cervical cancer (OMIM:603956); Bladder cancer (OMIM:109800); Thanatophoric dysplasia, type II (OMIM:187601); Thanatophoric dysplasia, type I (OMIM:187600); Colorectal cancer (OMIM:114500); Muenke syndrome (OMIM:602849); Crouzon syndrome with acanthosis nigricans (OMIM:612247); Achondroplasia (ORPHA:15); ORPHA:251579; ORPHA:251576; Non-syndromic bicoronal craniosynostosis (ORPHA:35099); Muenke syndrome (ORPHA:53271); Thanatophoric dysplasia type 1 (ORPHA:1860); Saethre-Chotzen syndrome (ORPHA:794); Thanatophoric dysplasia type 2 (ORPHA:93274); Crouzon syndrome-acanthosis nigricans syndrome (ORPHA:93262); Lacrimoauriculodentodigital syndrome (ORPHA:2363); Hypochondroplasia (ORPHA:429); Camptodactyly-tall stature-scoliosis-hearing loss syndrome (ORPHA:85164); Severe achondroplasia-developmental delay-acanthosis nigricans syndrome (ORPHA:85165)
FGFR4	—
FGFRL1	Wolf-Hirschhorn syndrome (OMIM:194190)
FGG	Congenital dysfibrinogenemia (OMIM:616004); Afibrinogenemia, congenitalhypofibrinogenemia, congenital, included (OMIM:202400); Familial hypofibrinogenemia (ORPHA:101041); Familial afibrinogenemia (ORPHA:98880); Familial dysfibrinogenemia (ORPHA:98881); ORPHA:248408
FH	Hereditary leiomyomatosis and renal cell cancer (OMIM:150800); Fumarase deficiency (OMIM:606812); Fumaric aciduria (ORPHA:24); Hereditary leiomyomatosis and renal cell cancer (ORPHA:523); Hereditary pheochromocytoma-paraganglioma (ORPHA:29072)
FHIT	ORPHA:422526
FHL1	Uruguay faciocardiomusculoskeletal syndrome (OMIM:300280); Myopathy, reducing body, X-linked, childhood-onset (OMIM:300718); Myopathy, reducing body, X-linked, early-onset, severe (OMIM:300717); Myopathy, X-linked, with postural muscle atrophy (OMIM:300696); Scapuloperoneal myopathy, X-linked dominant (OMIM:300695); ORPHA:97239; ORPHA:178461; X-linked Emery-Dreifuss muscular dystrophy (ORPHA:98863); ORPHA:431272
FHL2	Familial isolated dilated cardiomyopathy (ORPHA:154)
FICD	—
FIG4	Polymicrogyria, bilateral temporooccipital (OMIM:612691); Yunis-Varon syndrome (OMIM:216340); Charcot-Marie-Tooth disease, type 4J (OMIM:611228); Amyotrophic lateral sclerosis 11 (OMIM:612577); ORPHA:139515; Bilateral parasagittal parieto-occipital polymicrogyria (ORPHA:208441); Amyotrophic lateral sclerosis (ORPHA:803); Yunis-Varon syndrome (ORPHA:3472)
FIGLA	Premature ovarian failure 6 (OMIM:612310); 46,XX gonadal dysgenesis (ORPHA:243)
FIP1L1	ORPHA:168940; ORPHA:314950; Acute promyelocytic leukemia (ORPHA:520)
FIRRM	—
FKBP10	Bruck syndrome 1 (OMIM:259450); Osteogenesis imperfecta, type XI (OMIM:610968); Bruck syndrome (ORPHA:2771); ORPHA:216812; ORPHA:216820; Kuskokwim syndrome (ORPHA:1149)
FKBP14	Ehlers-Danlos syndrome, kyphoscoliotic type, 2 (OMIM:614557); Kyphoscoliotic Ehlers-Danlos syndrome due to FKBP22 deficiency (ORPHA:300179)
FKBP1A	—
FKBP4	—
FKBP5	Major depressive disorder (OMIM:608516)
FKBP6	Spermatogenic failure 77 (OMIM:620103); Williams syndrome (ORPHA:904)
FKBP8	—
FKRP	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5 (OMIM:607155); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1 (OMIM:236670); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5 (OMIM:613153); Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5 (OMIM:606612); FKRP-related limb-girdle muscular dystrophy R9 (ORPHA:34515); Congenital muscular dystrophy with intellectual disability (ORPHA:370968); Congenital muscular dystrophy without intellectual disability (ORPHA:370980); Congenital muscular dystrophy with cerebellar involvement (ORPHA:370959); Walker-Warburg syndrome (ORPHA:899); Muscle-eye-brain disease (ORPHA:588)
FKTN	Cardiomyopathy, dilated, 1X (OMIM:611615); Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4 (OMIM:611588); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1 (OMIM:236670); Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4 (OMIM:613152); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 (OMIM:253800); Congenital muscular dystrophy without intellectual disability (ORPHA:370980); Congenital muscular dystrophy, Fukuyama type (ORPHA:272); Familial isolated dilated cardiomyopathy (ORPHA:154); Walker-Warburg syndrome (ORPHA:899); ORPHA:206554; Muscle-eye-brain disease (ORPHA:588)
FLCN	Pneumothorax, primary spontaneous (OMIM:173600); Potocki-Lupski syndrome (OMIM:610883); Colorectal cancer (OMIM:114500); Renal cell carcinoma, nonpapillary (OMIM:144700); Birt-Hogg-Dube syndrome (OMIM:135150); Birt-Hogg-Dubé syndrome (ORPHA:122); Familial spontaneous pneumothorax (ORPHA:2903); ORPHA:422526

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
FLG	Dermatitis, atopic, susceptibility to, 2 (OMIM:605803); Ichthyosis vulgaris (OMIM:146700); Recessive X-linked ichthyosis (ORPHA:461)
FLI1	Bleeding disorder, platelet-type, 21 (OMIM:617443); ORPHA:319; ORPHA:370334; Peripheral primitive neuroectodermal tumor (ORPHA:370348); Paris-Trousseau thrombocytopenia (ORPHA:851); ORPHA:248340; Jacobsen syndrome (ORPHA:2308)
FLJ12825	—
FLNA	Melnick-Needles syndrome (OMIM:309350); Heterotopia, periventricular, X-linked dominant (OMIM:300049); Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (OMIM:300048); Cardiac valvular dysplasia, X-linked (OMIM:314400); Fg syndrome 2 (OMIM:300321); Terminal osseous dysplasia (OMIM:300244); Frontometaphyseal dysplasia (OMIM:305620); Otopalatodigital syndrome, type II (OMIM:304120); Otopalatodigital syndrome, type I (OMIM:311300); Otopalatodigital syndrome type 1 (ORPHA:90650); Otopalatodigital syndrome type 2 (ORPHA:90652); FLNA-related X-linked myxomatous valvular dysplasia (ORPHA:555877); Neuronal intestinal pseudoobstruction (ORPHA:99811); Terminal osseous dysplasia-pigmentary defects syndrome (ORPHA:88630); Periventricular nodular heterotopia (ORPHA:98892); ORPHA:482606; Frontometaphyseal dysplasia (ORPHA:1826); X-linked Ehlers-Danlos syndrome (ORPHA:75497); Congenital short bowel syndrome (ORPHA:2301); Melnick-Needles syndrome (ORPHA:2484)
FLNB	Spondylocarpotarsal synostosis syndrome (OMIM:272460); Atelosteogenesis, type III (OMIM:108721); Atelosteogenesis, type I (OMIM:108720); Larsen syndrome (OMIM:150250); Boomerang dysplasia (OMIM:112310); Spondylocarpotarsal synostosis (ORPHA:3275); Boomerang dysplasia (ORPHA:1263); Atelosteogenesis type III (ORPHA:56305); Atelosteogenesis type I (ORPHA:1190); Larsen syndrome (ORPHA:503)
FLNC	Myopathy, distal, 4 (OMIM:614065); Filaminopathy, autosomal dominant (OMIM:609524); Cardiomyopathy, familial hypertrophic, 26 (OMIM:617047); FLNC-related handgrip and calf weakness-distal myopathy (ORPHA:63273); Familial isolated restrictive cardiomyopathy (ORPHA:75249); Muscle filaminopathy (ORPHA:171445)
FLOT1	—
FLT1	Preeclampsia (ORPHA:275555)
FLT3	Leukemia, acute lymphocytic, susceptibility to, 1 (OMIM:613065); Leukemia, acute myeloid (OMIM:601626); ORPHA:99861; ORPHA:585909; ORPHA:98833; ORPHA:98834; ORPHA:98832; ORPHA:98829; ORPHA:589534; ORPHA:589595; ORPHA:102724
FLT4	Lymphatic malformation 1 (OMIM:153100); Hemangioma, capillary infantile (OMIM:602089); Congenital heart defects, multiple types, 7 (OMIM:618780); Milroy disease (ORPHA:79452); Tetralogy of Fallot (ORPHA:3303)
FLVCR1	Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia (OMIM:621060); Ataxia, posterior column, with retinitis pigmentosa (OMIM:609033); Posterior column ataxia-retinitis pigmentosa syndrome (ORPHA:88628)
FLVCR2	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome (OMIM:225790); ORPHA:221126
FMN1	—
FMN2	Intellectual developmental disorder, autosomal recessive 47 (OMIM:616193); ORPHA:88616
FMO1	—
FMO2	—
FMO3	Trimethylaminuria (OMIM:602079); Severe primary trimethylaminuria (ORPHA:468726)
FMO4	—
FMO5	—
FMOD	—
FMR1	Fragile X tremor/ataxia syndrome (OMIM:300623); Fragile X syndrome (OMIM:300624); Premature ovarian failure 1 (OMIM:311360); ORPHA:642691; Xq27.3q28 duplication syndrome (ORPHA:261483); Fragile X syndrome (ORPHA:908); Fragile X-associated tremor/ataxia syndrome (ORPHA:93256)
FN1	Spondylometaphyseal dysplasia, corner Fracture type (OMIM:184255); Glomerulopathy with fibronectin deposits 2 (OMIM:601894); Fibronectin glomerulopathy (ORPHA:84090); Spondylometaphyseal dysplasia, 'corner fracture' type (ORPHA:93315)
FN3K	—
FOLH1	—
FOLR1	Neurodegeneration due to cerebral folate transport deficiency (OMIM:613068); ORPHA:217382
FOXA1	—
FOXA2	Combined pituitary hormone deficiencies, genetic forms (ORPHA:95494)
FOXA3	—
FOXC1	Axenfeld-Rieger syndrome, type 3 (OMIM:602482); Anterior segment dysgenesis 3 (OMIM:601631); Isolated aniridia (ORPHA:250923); Axenfeld-Rieger syndrome (ORPHA:782); ORPHA:98978; ORPHA:91483; Peters anomaly (ORPHA:708)
FOXC2	Lymphedema-Distichiasis syndrome (OMIM:153400); Lymphedema-distichiasis syndrome (ORPHA:33001)
FOXD3	Autoimmune disease, susceptibility to, 1 (OMIM:607836)
FOXE1	Thyroid cancer, nonmedullary, 4 (OMIM:616534); Bamforth-Lazarus syndrome (OMIM:241850); ORPHA:146; Bamforth-Lazarus syndrome (ORPHA:1226); Athyreosis (ORPHA:95713); Familial papillary or follicular thyroid carcinoma (ORPHA:319487)
FOXE3	Cataract 34, multiple types (OMIM:612968); Anterior segment dysgenesis 2 (OMIM:610256); Aortic aneurysm, familial thoracic 11, susceptibility to (OMIM:617349); Congenital primary aphakia (ORPHA:83461); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); Peters anomaly (ORPHA:708)
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins (OMIM:265380); Congenital alveolar

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	capillary dysplasia (ORPHA:210122)
FOXF2	—
FOXG1	Rett syndrome, congenital variant (OMIM:613454); ORPHA:598164; Non-specific early-onset epileptic encephalopathy (ORPHA:442835); FOXG1 syndrome due to 14q12 microdeletion (ORPHA:261144)
FOXH1	Septopreoptic holoprosencephaly (ORPHA:280195); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925)
FOXI1	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct (OMIM:600791); Pendred syndrome (OMIM:274600); Pendred syndrome (ORPHA:705); ORPHA:402041
FOXJ1	Ciliary dyskinesia, primary, 43 (OMIM:618699); Primary ciliary dyskinesia (ORPHA:244)
FOXK1	—
FOXK2	—
FOXL2	Blepharophimosis, epicanthus inversus, and ptosis (OMIM:110100); Premature ovarian failure 3 (OMIM:608996); ORPHA:99915; Blepharophimosis-ptosis-epicanthus inversus syndrome plus (ORPHA:572333); ORPHA:572361; ORPHA:572354
FOXM1	—
FOXN1	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant (OMIM:618806); Immune defect due to absence of thymus (OMIM:242700); T-CELL IMMUNODEFICIENCY, CONGENITAL ALOPECIA, AND NAIL DYSTROPHY (OMIM:601705); T-cell immunodeficiency with thymic aplasia (ORPHA:83471); ORPHA:676039; Severe combined immunodeficiency due to FOXN1 deficiency (ORPHA:169095)
FOXO1	Rhabdomyosarcoma 2, alveolar (OMIM:268220); ORPHA:99756
FOXP1	Intellectual developmental disorder with language impairment with or without autistic features (OMIM:613670); MALT lymphoma (ORPHA:52417); FOXP1 Syndrome (ORPHA:391372); ORPHA:585877
FOXP2	Speech-language disorder-1 (OMIM:602081); Isolated childhood apraxia of speech (ORPHA:209908); 7q31 microdeletion syndrome (ORPHA:251061)
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked (OMIM:304790); Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome (ORPHA:37042)
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19 (OMIM:618241); Isolated complex I deficiency (ORPHA:2609)
FPGS	—
FPGT	—
FPR1	ORPHA:447740
FRAS1	Fraser syndrome (OMIM:219000); Fraser syndrome (ORPHA:2052); ORPHA:93100
FREM1	Manitoba oculotrichoanal syndrome (OMIM:248450); Bifid nose with or without anorectal and renal anomalies (OMIM:608980); Trigonocephaly 2 (OMIM:614485); Oculotrichoanal syndrome (ORPHA:2717); Non-syndromic metopic craniosynostosis (ORPHA:3366); ORPHA:93100; BNAR syndrome (ORPHA:217266)
FREM2	Fraser syndrome 2 (OMIM:617666); Cryptophthalmos, unilateral or bilateral, isolated (OMIM:123570); Fraser syndrome (ORPHA:2052); ORPHA:93100; ORPHA:98949
FREM3	—
FRK	—
FRMD5	Neurodevelopmental disorder with eye movement abnormalities and ataxia (OMIM:620094)
FRMD7	Nystagmus 1, congenital, X-linked (OMIM:310700)
FRMPD4	Intellectual developmental disorder, X-linked 104 (OMIM:300983); ORPHA:777
FRY	—
FRZB	Osteoarthritis susceptibility 1 (OMIM:165720)
FSBP	—
FSCB	—
FSCN2	Retinitis pigmentosa 30 (OMIM:607921); Retinitis pigmentosa (ORPHA:791)
FSHB	Hypogonadotropic hypogonadism 24 without anosmia (OMIM:229070); Isolated follicle stimulating hormone deficiency (ORPHA:52901)
FSHR	Ovarian hyperstimulation syndrome (OMIM:608115); Ovarian dysgenesis 1 (OMIM:233300); 46,XX gonadal dysgenesis (ORPHA:243); Ovarian hyperstimulation syndrome (ORPHA:64739)
FSIP1	—
FSIP2	Spermatogenic failure 34 (OMIM:618153); ORPHA:276234
FST	—
FSTL1	—
FTCD	Formiminotransferase deficiency (OMIM:229100); Formiminoglutamic aciduria (ORPHA:51208)
FTH1	Hemochromatosis, type 5 (OMIM:615517); Neurodegeneration with brain iron accumulation 9 (OMIM:620669); ORPHA:247790
FTHL17	—
FTO	Growth retardation, developmental delay, coarse facies, and earlydeath (OMIM:612938); Obesity, susceptibility to, BMIQ14 (OMIM:612460); ORPHA:210144
FTSJ1	Intellectual developmental disorder, X-linked 9 (OMIM:309549); ORPHA:777
FUCA1	Fucosidosis (OMIM:230000); Fucosidosis (ORPHA:349)
FUS	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia (OMIM:608030); Tremor, hereditary essential, 4 (OMIM:614782); Myxoid/round cell liposarcoma (ORPHA:99967); Frontotemporal dementia with motor neuron disease (ORPHA:275872); Juvenile amyotrophic lateral sclerosis (ORPHA:300605); Amyotrophic lateral sclerosis (ORPHA:803); ORPHA:79105
FUT1	Bombay phenotype (OMIM:616754)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
FUT2	Bombay phenotype (OMIM:616754); OMIM:612542
FUT3	OMIM:618983
FUT6	OMIM:613852
FUT7	—
FUT8	Congenital disorder of glycosylation with defective fucosylation 1 (OMIM:618005)
FUZ	Neural tube defects, susceptibility to (OMIM:182940); Caudal regression syndrome (ORPHA:3027); ORPHA:620158
FXN	Friedreich ataxia 2 (OMIM:601992); Friedreich ataxia 1 (OMIM:229300); Friedreich ataxia (ORPHA:95)
FXYD2	Hypomagnesemia 2, renal (OMIM:154020); ORPHA:34528
FXYD6	—
FYCO1	Cataract, autosomal recessive congenital 2 (OMIM:610019); ORPHA:98994; ORPHA:98991
FZD3	—
FZD4	Exudative vitreoretinopathy 1 (OMIM:133780); Familial exudative vitreoretinopathy (ORPHA:891); Retinopathy of prematurity (ORPHA:90050); Persistent hyperplastic primary vitreous (ORPHA:91495)
FZD6	Nail disorder, nonsyndromic congenital, 1 (OMIM:161050); ORPHA:280654
FZD9	—
G	—
G6PC1	Glycogen storage disease Ia (OMIM:232200); ORPHA:79258
G6PC2	—
G6PC3	Neutropenia, severe congenital, 4, autosomal recessive (OMIM:612541); ORPHA:331176
G6PD	Hemolytic anemia, G6PD deficient (favism) (OMIM:300908); OMIM:611162; ORPHA:466026
GAA	Glycogen storage disease II (OMIM:232300); Pompe disease, late-onset (OMIM:621314); Glycogen storage disease due to acid maltase deficiency, infantile onset (ORPHA:308552); ORPHA:420429
GABARAPL1	—
GABBR1	Neurodevelopmental disorder with language delay and variable cognitive abnormalities (OMIM:620502); ORPHA:178469
GABRA1	Developmental and epileptic encephalopathy 19 (OMIM:615744); OMIM:611136; Juvenile myoclonic epilepsy (ORPHA:307); Childhood absence epilepsy (ORPHA:64280); Dravet syndrome (ORPHA:33069)
GABRA2	Alcohol dependence (OMIM:103780); Epileptic encephalopathy, early infantile, 78 (OMIM:618557); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
GABRA6	—
GABRB3	Epileptic encephalopathy, early infantile, 43 (OMIM:617113); Epilepsy, childhood absence, susceptibility to, 5 (OMIM:612269); Lennox-Gastaut syndrome (ORPHA:2382); Childhood absence epilepsy (ORPHA:64280)
GABRD	Epilepsy, idiopathic generalized, 10 (OMIM:613060); Juvenile myoclonic epilepsy (ORPHA:307); Genetic epilepsy with febrile seizure plus (ORPHA:36387); 1p36 deletion syndrome (ORPHA:1606)
GABRG1	—
GABRG2	Epileptic encephalopathy, early infantile, 74 (OMIM:618396); Febrile seizures, familial, 8 (OMIM:607681); Genetic epilepsy with febrile seizure plus (ORPHA:36387); Non-specific early-onset epileptic encephalopathy (ORPHA:442835); Self-limited epilepsy with centrotemporal spikes (ORPHA:1945); Childhood absence epilepsy (ORPHA:64280); Dravet syndrome (ORPHA:33069)
GABRR2	—
GAD1	Developmental and epileptic encephalopathy 89 (OMIM:619124); ORPHA:210141
GAD2	—
GADD45A	—
GAK	—
GAL3ST1	—
GAL3ST2	—
GAL3ST3	—
GAL3ST4	—
GALC	Krabbe disease (OMIM:245200); Late-infantile/juvenile Krabbe disease (ORPHA:206443); Adult Krabbe disease (ORPHA:206448); Infantile Krabbe disease (ORPHA:206436)
GALE	Galactose epimerase deficiency (OMIM:230350); Thrombocytopenia 13, syndromic (OMIM:620776); ORPHA:308487; ORPHA:308473
GALK1	Galactokinase deficiency with cataracts (OMIM:230200); Galactokinase deficiency (ORPHA:79237)
GALNS	Mucopolysaccharidosis IVA (OMIM:253000); ORPHA:309297
GALNT11	—
GALNT12	OMIM:608812
GALNT13	—
GALNT14	—
GALNT17	—
GALNT18	—
GALNT2	Congenital disorder of glycosylation, type IIt (OMIM:618885)
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 1 (OMIM:211900); ORPHA:306661
GALNT5	—
GALNT6	—
GALNT7	—
GALNT8	—
GALNT9	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
GALNTL6	—
GALT	Galactosemia (OMIM:230400); Classic galactosemia (ORPHA:79239)
GAMT	Cerebral creatine deficiency syndrome 2 (OMIM:612736); Guanidinoacetate methyltransferase deficiency (ORPHA:382)
GAN	Giant axonal neuropathy 1, autosomal recessive (OMIM:256850); Giant axonal neuropathy (ORPHA:643)
GAP43	—
GAPDH	—
GARS1	Neuronopathy, distal hereditary motor, type VA (OMIM:600794); Spinal muscular atrophy, infantile, James type (OMIM:619042); Charcot-Marie-Tooth disease, axonal, type 2D (OMIM:601472); ORPHA:99938; Distal hereditary motor neuropathy type 5 (ORPHA:139536)
GAS1	Septopreoptic holoprosencephaly (ORPHA:280195); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925)
GAS2	Deafness, autosomal recessive 125 (OMIM:620877)
GAS2L2	Ciliary dyskinesia, primary, 41 (OMIM:618449); Primary ciliary dyskinesia (ORPHA:244)
GAS5	—
GATA1	Thrombocytopenia with beta-thalassemia, X-linked (OMIM:314050); Myeloproliferative syndrome, transient (OMIM:159595); Hemolytic anemia due to elevated adenosine deaminase (OMIM:301083); Thrombocytopenia, X-linked, with or without dyserythropoietic anemia (OMIM:300367); Anemia, X-linked, with or without neutropenia and/or platelet abnormalities (OMIM:300835); ORPHA:99887; Diamond-Blackfan anemia (ORPHA:124); ORPHA:86849; ORPHA:420611; Thrombocytopenia with congenital dyserythropoietic anemia (ORPHA:67044); Beta-thalassemia-X-linked thrombocytopenia syndrome (ORPHA:231393); ORPHA:363727; Congenital erythropoietic porphyria (ORPHA:79277)
GATA2	Myelodysplastic syndrome (OMIM:614286); Immunodeficiency 21 (OMIM:614172); Lymphedema, primary, with myelodysplasia (OMIM:614038); Leukemia, acute myeloid (OMIM:601626); GATA2 deficiency spectrum (ORPHA:228423)
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia (OMIM:146255); ORPHA:585936; Hypoparathyroidism-sensorineural deafness-renal disease syndrome (ORPHA:2237)
GATA4	Testicular anomalies with or without congenital heart disease (OMIM:615542); Tetralogy of Fallot (OMIM:187500); Ventricular septal defect 1 (OMIM:614429); Atrioventricular septal defect 4 (OMIM:614430); Atrial septal defect 2 (OMIM:607941); ORPHA:99067; ORPHA:99068; Hereditary atrial fibrillation (ORPHA:334); 46,XY partial gonadal dysgenesis (ORPHA:251510); Atrial septal defect, ostium secundum type (ORPHA:99103); Tetralogy of Fallot (ORPHA:3303); 8p23.1 microdeletion syndrome (ORPHA:251071); ORPHA:576232
GATA5	Congenital heart defects, multiple types, 5 (OMIM:617912); Hereditary atrial fibrillation (ORPHA:334); Tetralogy of Fallot (ORPHA:3303); Familial bicuspid aortic valve (ORPHA:402075)
GATA6	Conotruncal heart malformations (OMIM:217095); Pancreatic agenesis and congenital heart defects (OMIM:600001); Tetralogy of Fallot (OMIM:187500); Atrioventricular septal defect 5 (OMIM:614474); Atrial septal defect 9 (OMIM:614475); ORPHA:99067; Hereditary atrial fibrillation (ORPHA:334); Atrial septal defect, ostium secundum type (ORPHA:99103); Tetralogy of Fallot (ORPHA:3303); ORPHA:665058; ORPHA:665044; Congenital diaphragmatic hernia (ORPHA:2140); Pancreatic hypoplasia-diabetes-congenital heart disease syndrome (ORPHA:2255)
GATAD1	Cardiomyopathy, dilated, 2B (OMIM:614672); Familial isolated dilated cardiomyopathy (ORPHA:154)
GATM	Cerebral creatine deficiency syndrome 3 (OMIM:612718); Fanconi renotubular syndrome 1 (OMIM:134600); Primary Fanconi renotubular syndrome (ORPHA:3337); L-Arginine:glycine amidinotransferase deficiency (ORPHA:35704)
GBA1	Gaucher disease, perinatal lethal (OMIM:608013); Gaucher disease, type IIIC (OMIM:231005); Gaucher disease, type III (OMIM:231000); Dementia, lewy body (OMIM:127750); Gaucher disease, type II (OMIM:230900); Gaucher disease, type I (OMIM:230800); Parkinson disease, late-onset (OMIM:168600); Fetal Gaucher disease (ORPHA:85212); Gaucher disease type 3 (ORPHA:77261); Gaucher disease type 2 (ORPHA:77260); Gaucher disease type 1 (ORPHA:77259); Hereditary late-onset Parkinson disease (ORPHA:411602); Gaucher disease-ophthalmoplegia-cardiovascular calcification syndrome (ORPHA:2072)
GBE1	Polyglucosan body neuropathy, adult form (OMIM:263570); Glycogen storage disease IV (OMIM:232500); Adult polyglucosan body disease (ORPHA:206583); ORPHA:308638; ORPHA:308621; ORPHA:308684; ORPHA:308670; ORPHA:308655; ORPHA:308698; ORPHA:308712
GBGT1	—
GC	—
GCDH	Glutaric acidemia I (OMIM:231670); Glutaryl-CoA dehydrogenase deficiency (ORPHA:25)
GCGR	Mahvash disease (OMIM:619290); GCGR-related hyperglucagonemia (ORPHA:438274)
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia (OMIM:128230); Hyperphenylalaninemia, BH4-deficient, B (OMIM:233910); Autosomal dominant dopa-responsive dystonia (ORPHA:98808); ORPHA:2102
GCK	Diabetes mellitus, noninsulin-dependent (OMIM:125853); Maturity-onset diabetes of the young, type II (OMIM:125851); Diabetes mellitus, permanent neonatal 1 (OMIM:606176); Hyperinsulinemic hypoglycemia, familial, 3 (OMIM:602485); Isolated permanent neonatal diabetes mellitus (ORPHA:99885); MODY (ORPHA:552); Congenital glucokinase-related hyperinsulinism (ORPHA:79299)
GCKR	OMIM:613463
GCLC	OMIM:608446; Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency (OMIM:230450); Glutamate-cysteine ligase deficiency (ORPHA:33574); Cystic fibrosis (ORPHA:586)
GCLM	OMIM:608446

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
GCM2	Hypoparathyroidism, familial isolated 2 (OMIM:618883); Hyperparathyroidism 4 (OMIM:617343); Familial isolated hyperparathyroidism (ORPHA:99879); Familial isolated hypoparathyroidism due to agenesis of parathyroid gland (ORPHA:2239)
GCNT1	—
GCNT2	Cataract 13 with adult i phenotype (OMIM:116700); OMIM:110800; ORPHA:98994
GDA	—
GDAP1	Charcot-Marie-Tooth disease, recessive intermediate A (OMIM:608340); Charcot-Marie-Tooth disease, axonal, type 2K (OMIM:607831); Charcot-Marie-Tooth disease, type 4A (OMIM:214400); Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, autosomalrecessive (OMIM:607706); Autosomal recessive Charcot-Marie-Tooth disease with hoarseness (ORPHA:101097); Charcot-Marie-Tooth disease type 4A (ORPHA:99948); Autosomal dominant Charcot-Marie-Tooth disease type 2K (ORPHA:99944); ORPHA:217055; ORPHA:101102
GDF1	Congenital heart defects, multiple types, 6 (OMIM:613854); Right atrial isomerism (Ivemark) (OMIM:208530); Tetralogy of Fallot (ORPHA:3303); ORPHA:97548; ORPHA:216718
GDF3	Microphthalmia, isolated, with coloboma 6 (OMIM:613703); Klippel-Feil syndrome 3, autosomal dominant (OMIM:613702); Microphthalmia, isolated 7 (OMIM:613704); ORPHA:98938; Isolated Klippel-Feil syndrome (ORPHA:2345)
GDF5	Symphalangism, proximal, 1B (OMIM:615298); Brachydactyly, type A2 (OMIM:112600); Acromesomelic dysplasia, Hunter-Thompson type (OMIM:201250); Brachydactyly, type A1, C (OMIM:615072); Multiple synostoses syndrome 2 (OMIM:610017); Brachydactyly, type C (OMIM:113100); Acromesomelic dysplasia 2A (OMIM:200700); OMIM:612400; Fibular hypoplasia and complex brachydactyly (OMIM:228900); Proximal symphalangism (ORPHA:3250); Multiple synostoses syndrome (ORPHA:3237); Acromesomelic dysplasia, Hunter-Thompson type (ORPHA:968); Fibular aplasia-complex brachydactyly syndrome (ORPHA:2639); Angel-shaped phalango-epiphyseal dysplasia (ORPHA:63442); Brachydactyly type A2 (ORPHA:93396); Brachydactyly type C (ORPHA:93384); Brachydactyly type A1 (ORPHA:93388); Acromesomelic dysplasia, Grebe type (ORPHA:2098)
GDF6	Multiple synostoses syndrome 4 (OMIM:617898); Microphthalmia, isolated 4 (OMIM:613094); Leber congenital amaurosis 17 (OMIM:615360); Klippel-Feil syndrome 1, autosomal dominant (OMIM:118100); Microphthalmia, isolated, with coloboma 6 (OMIM:613703); Multiple synostoses syndrome (ORPHA:3237); Leber congenital amaurosis (ORPHA:65); ORPHA:98938; Isolated Klippel-Feil syndrome (ORPHA:2345)
GDF9	Premature ovarian failure 14 (OMIM:618014)
GDI1	Intellectual developmental disorder, X-linked 41 (OMIM:300849); ORPHA:777
GDNF	Hirschsprung disease, susceptibility to, 3 (OMIM:613711); Hirschsprung disease (ORPHA:388); Congenital central hypoventilation syndrome (ORPHA:661)
GFAP	Alexander disease (OMIM:203450); Alexander disease type II (ORPHA:363722); Alexander disease type I (ORPHA:363717)
GFI1	Neutropenia, severe congenital, 2, autosomal dominant (OMIM:613107); Neutropenia, nonimmune chronic idiopathic, of adults (OMIM:607847); Autosomal dominant severe congenital neutropenia (ORPHA:486)
GFI1B	Bleeding disorder, platelet-type, 17 (OMIM:187900); ORPHA:734; Autosomal dominant macrothrombocytopenia (ORPHA:140957)
GFM1	Combined oxidative phosphorylation deficiency 1 (OMIM:609060); Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1 (ORPHA:137681)
GFM2	Combined oxidative phosphorylation deficiency 39 (OMIM:618397); Combined oxidative phosphorylation defect type 39 (ORPHA:565624)
GFPT1	Myasthenic syndrome, congenital, with tubular aggregates 1 (OMIM:610542); Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency (OMIM:608931); Congenital myasthenic syndrome with glycosylation defect (ORPHA:353327)
GFRA1	Renal hypodysplasia/aplasia 4 (OMIM:619887); Renal agenesis, bilateral (ORPHA:1848)
GFRA2	—
GGACT	—
GGCX	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency (OMIM:610842); VITAMIN K-DEPENDENT CLOTTING FACTORS, COMBINED DEFICIENCY OF, 1 (OMIM:277450); Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency (ORPHA:91135); Pseudoxanthoma elasticum-like skin manifestations with retinitis pigmentosa (ORPHA:436274); Hereditary combined deficiency of vitamin K-dependent clotting factors (ORPHA:98434)
GGH	—
GH1	Growth hormone deficiency, isolated, type IB (OMIM:612781); Growth hormone deficiency, isolated, type IA (OMIM:262400); Pituitary dwarfism IV (OMIM:262650); Isolated growth hormone deficiency, type II (OMIM:173100); ORPHA:231671; ORPHA:231679; ORPHA:231662; ORPHA:629
GH2	—
GHR	Hypercholesterolemia, familial, 1 (OMIM:143890); Laron syndrome (OMIM:262500); Growth hormone insensitivity, partial (OMIM:604271); Short stature due to partial GHR deficiency (ORPHA:314802); Laron syndrome (ORPHA:633)
GHRH	—
GHRHR	Growth hormone deficiency, isolated, type IV (OMIM:618157); ORPHA:684247
GHRL	OBESITY (OMIM:601665)
GHRLOS	—
GHSR	Growth hormone deficiency, isolated partial (OMIM:615925); Short stature due to GHSR deficiency (ORPHA:314811)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
GIGYF2	Parkinson disease 11 (OMIM:607688); Hereditary late-onset Parkinson disease (ORPHA:411602)
GIMAP8	—
GIP	—
GIPC3	Deafness, autosomal recessive 15 (OMIM:601869); ORPHA:90636
GIPR	—
GJA3	Cataract, zonular pulverulent 3 (OMIM:601885); ORPHA:98991; ORPHA:98993; ORPHA:98984
GJA5	Atrial fibrillation, familial, 11 (OMIM:614049); Atrial standstill 1 (OMIM:108770); Chromosome 1q21.1 deletion syndrome, 1.35-mb (OMIM:612474); Hereditary atrial fibrillation (ORPHA:334); Tetralogy of Fallot (ORPHA:3303)
GJA8	Cataract 1, multiple types (OMIM:116200); Chromosome 1q21.1 deletion syndrome, 1.35-mb (OMIM:612474); Cataract-microcornea syndrome (ORPHA:1377); ORPHA:98994; ORPHA:98991; ORPHA:98984; ORPHA:98985; ORPHA:91490
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1 (OMIM:302800); X-linked Charcot-Marie-Tooth disease type 1 (ORPHA:101075); X-linked progressive cerebellar ataxia (ORPHA:1175)
GJB2	Vohwinkel syndrome (OMIM:124500); Ichthyosis, hystrix-like, with deafness (OMIM:602540); Bart-Pumphrey syndrome (OMIM:149200); Keratitis-Ichthyosis-Deafness syndrome, autosomal dominant (OMIM:148210); Deafness, autosomal recessive 1A (OMIM:220290); Deafness, autosomal dominant nonsyndromic sensorineural 3 (OMIM:601544); Keratoderma, palmoplantar, with deafness (OMIM:148350); ORPHA:90635; ORPHA:90636; ORPHA:166286; Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome (ORPHA:2698); KID syndrome (ORPHA:477); Keratoderma hereditarium mutilans (ORPHA:494); Palmoplantar keratoderma-deafness syndrome (ORPHA:2202)
GJB3	Deafness, autosomal dominant 2B (OMIM:612644); Erythrokeratodermia variabilis et progressiva 1 (OMIM:133200); Deafness, autosomal recessive 1A (OMIM:220290); Erythrokeratodermia variabilis (ORPHA:317); ORPHA:90635; ORPHA:90636; ORPHA:139512
GJB4	Erythrokeratodermia variabilis et progressiva 2 (OMIM:617524); Erythrokeratodermia variabilis (ORPHA:317)
GJB6	Clouston syndrome (OMIM:129500); Deafness, autosomal dominant 3B (OMIM:612643); Deafness, autosomal recessive 1B (OMIM:612645); Deafness, autosomal recessive 1A (OMIM:220290); ORPHA:90635; ORPHA:90636; Hidrotic ectodermal dysplasia (ORPHA:189); KID syndrome (ORPHA:477)
GJC2	Lymphatic malformation 3 (OMIM:613480); Leukodystrophy, hypomyelinating, 2 (OMIM:608804); Spastic paraplegia 44, autosomal recessive (OMIM:613206); ORPHA:280282; Autosomal recessive spastic paraplegia type 44 (ORPHA:320401); GJC2-related late-onset primary lymphedema (ORPHA:568051)
GJC3	—
GJD2	—
GK	Glycerol kinase deficiency (OMIM:307030); ORPHA:284411; ORPHA:284414
GLA	Fabry disease (OMIM:301500); Fabry disease (ORPHA:324)
GLB1	GM1-gangliosidosis, type I (OMIM:230500); Mucopolysaccharidosis type IVB (Morquio) (OMIM:253010); GM1-gangliosidosis, type II (OMIM:230600); GM1-gangliosidosis, type III (OMIM:230650); ORPHA:309310; GM1 gangliosidosis type 1 (ORPHA:79255); ORPHA:79256; ORPHA:79257
GLCCI1	OMIM:614400
GLDC	Glycine encephalopathy (OMIM:605899); ORPHA:289857; ORPHA:289863; ORPHA:289860
GLE1	Congenital arthrogryposis with anterior horn cell disease (OMIM:611890); Lethal congenital contracture syndrome 1 (OMIM:253310); ORPHA:53696; Lethal congenital contracture syndrome type 1 (ORPHA:1486); Amyotrophic lateral sclerosis (ORPHA:803)
GLI2	Holoprosencephaly 9 (OMIM:610829); Culler-Jones syndrome (OMIM:615849); Septopreoptic holoprosencephaly (ORPHA:280195); Combined pituitary hormone deficiencies, genetic forms (ORPHA:95494); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925); ORPHA:420584
GLI3	Pallister-Hall syndrome (OMIM:146510); Polydactyly, postaxial, types A1 and B (OMIM:174200); Greig cephalopolysyndactyly syndrome (OMIM:175700); Polydactyly, preaxial IV (OMIM:174700); Greig cephalopolysyndactyly syndrome (ORPHA:380); Acrocallosal syndrome (ORPHA:36); Pallister-Hall syndrome (ORPHA:672); ORPHA:93338; ORPHA:93335; ORPHA:93334; Isolated tibial hemimelia (ORPHA:93322)
GLIS2	Nephronophthisis 7 (OMIM:611498); ORPHA:329469; ORPHA:93592
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism (OMIM:610199); ORPHA:79118
GLMN	Glomuvenous malformations (OMIM:138000); Glomuvenous malformation (ORPHA:83454)
GLO1	—
GLP1R	—
GLRA1	Hyperekplexia 1 (OMIM:149400); Hereditary hyperekplexia (ORPHA:3197)
GLRB	Hyperekplexia 2 (OMIM:614619); Hereditary hyperekplexia (ORPHA:3197)
GLRX2	—
GLRX5	Spasticity, childhood-onset, with hyperglycinemia (OMIM:616859); Anemia, sideroblastic, 3, pyridoxine-refractory (OMIM:616860); Childhood-onset spasticity with hyperglycinemia (ORPHA:401866); ORPHA:255132
GLUL	Glutamine deficiency, congenital (OMIM:610015); Developmental and epileptic encephalopathy 116 (OMIM:620806); ORPHA:71278
GLYCTK	D-glyceric aciduria (OMIM:220120); D-glyceric aciduria (ORPHA:941)
GM2A	GM2-gangliosidosis, AB variant (OMIM:272750); GM2 gangliosidosis, AB variant (ORPHA:309246)
GMDS	—
GML	—
GMPS	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
GNAI2	Ventricular tachycardia, familial (OMIM:192605)
GNAI3	Auriculocondylar syndrome 1 (OMIM:602483); Auriculocondylar syndrome (ORPHA:137888)
GNAQ	Nevi flammei, familial multiple (OMIM:163000); Sturge-Weber syndrome (OMIM:185300); Sturge-Weber syndrome (ORPHA:3205); ORPHA:79483; Uveal melanoma (ORPHA:39044); Familial multiple nevi flammei (ORPHA:624); ORPHA:675359
GNAS	Pseudohypoparathyroidism, type IB (OMIM:603233); McCune-Albright syndrome, somatic, mosaic (OMIM:174800); Osseous heteroplasia, progressive (OMIM:166350); Pituitary adenoma 3, multiple types, somatic (OMIM:617686); Pseudohypoparathyroidism, type IA (OMIM:103580); ACTH-independent macronodular adrenal hyperplasia (OMIM:219080); Pseudopseudohypoparathyroidism (OMIM:612463); Pseudohypoparathyroidism, type IC (OMIM:612462); Pseudohypoparathyroidism type 1A (ORPHA:79443); Pseudohypoparathyroidism type 1C (ORPHA:79444); Pseudopseudohypoparathyroidism (ORPHA:79445); Progressive osseous heteroplasia (ORPHA:2762); ORPHA:93276; ORPHA:93277; Cushing syndrome due to bilateral macronodular adrenocortical disease (ORPHA:189427); Mazabraud syndrome (ORPHA:57782); McCune-Albright syndrome (ORPHA:562); Pseudohypoparathyroidism type 1B (ORPHA:94089)
GNAT1	Night blindness, congenital stationary, autosomal dominant 3 (OMIM:610444); Night blindness, congenital stationary, type 1G (OMIM:616389); ORPHA:714096
GNAT2	Achromatopsia 4 (OMIM:613856); Progressive cone dystrophy (ORPHA:1871); Achromatopsia (ORPHA:49382)
GNB1L	—
GNB3	Night blindness, congenital stationary, type 1H (OMIM:617024); Hypertension, essential (OMIM:145500)
GNE	Nonaka myopathy (OMIM:605820); Sialuria (OMIM:269921); Thrombocytopenia 12 with or without myopathy (OMIM:620757); ORPHA:438207; Sialuria (ORPHA:3166); GNE myopathy (ORPHA:602)
GNG7	—
GNMT	GLYCINE N-METHYLTRANSFERASE DEFICIENCY (OMIM:606664); ORPHA:289891
GNPAT	Rhizomelic chondrodysplasia punctata, type 2 (OMIM:222765); ORPHA:309796
GNPTAB	Mucolipidosis II alpha/beta (OMIM:252500); Mucolipidosis III alpha/beta (OMIM:252600); Mucolipidosis type III alpha/beta (ORPHA:423461); Mucolipidosis type II (ORPHA:576)
GNPTG	Mucolipidosis III gamma (OMIM:252605); ORPHA:423470
GNRH1	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Hypogonadotropic hypogonadism 12 with or without anosmia (OMIM:614841); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432)
GNS	Mucopolysaccharidosis, type IIID (OMIM:252940); ORPHA:79272
GOLGA3	—
GOLGA5	ORPHA:146
GON4L	Li-Takada-Miyake syndrome (OMIM:621212)
GOPC	ORPHA:171709
GORAB	Geroderma osteodysplasticum (OMIM:231070); Geroderma osteodysplastica (ORPHA:2078)
GOSR2	Muscular dystrophy, congenital, with or without seizures (OMIM:620166); Epilepsy, progressive myoclonic, 6 (OMIM:614018); ORPHA:280620
GOT1	OMIM:614419
GP1BA	Bernard-Soulier syndrome (OMIM:231200); Bernard-Soulier syndrome, type A2, autosomal dominant (OMIM:153670); Pseudo-Von willebrand disease (OMIM:177820); Nonarteritic anterior ischemic optic neuropathy, susceptibility to (OMIM:258660); Bernard-Soulier syndrome (ORPHA:274); ORPHA:52530; Fetal and neonatal alloimmune thrombocytopenia (ORPHA:853); Autosomal dominant macrothrombocytopenia (ORPHA:140957)
GP1BB	Bernard-Soulier syndrome (OMIM:231200); Bernard-Soulier syndrome (ORPHA:274); Fetal and neonatal alloimmune thrombocytopenia (ORPHA:853); Autosomal dominant macrothrombocytopenia (ORPHA:140957); 22q11.2 deletion syndrome (ORPHA:567)
GP6	Bleeding disorder, platelet-type, 11 (OMIM:614201); ORPHA:98885
GP9	Bernard-Soulier syndrome (OMIM:231200); Bernard-Soulier syndrome (ORPHA:274)
GPAM	—
GPATCH8	—
GPBAR1	—
GPC3	Wilms tumor 1 (OMIM:194070); Simpson-Golabi-Behmel syndrome, type 1 (OMIM:312870); Simpson-Golabi-Behmel syndrome (ORPHA:373); Nephroblastoma (ORPHA:654)
GPC4	Wilms tumor 1 (OMIM:194070); Keipert syndrome (OMIM:301026); Simpson-Golabi-Behmel syndrome (ORPHA:373); Keipert syndrome (ORPHA:2662)
GPC6	Omodysplasia 1 (OMIM:258315); Autosomal recessive omodysplasia (ORPHA:93329)
GPD1	Hypertriglyceridemia, transient infantile (OMIM:614480); ORPHA:300293
GPD1L	Brugada syndrome 2 (OMIM:611777); Brugada syndrome (ORPHA:130)
GPD2	Diabetes mellitus, noninsulin-dependent (OMIM:125853)
GPHN	Molybdenum cofactor deficiency, complementation group C (OMIM:615501); Hyperekplexia 1 (OMIM:149400); Hereditary hyperekplexia (ORPHA:3197); ORPHA:308400
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerasedeficiency (OMIM:613470); Hemolytic anemia due to glucophosphate isomerase deficiency (ORPHA:712)
GPIHBP1	Hyperlipoproteinemia, type ID (OMIM:615947); ORPHA:535458
GPLD1	—
GPNMB	Amyloidosis, primary localized cutaneous, 3 (OMIM:617920); ORPHA:319635
GPR12	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
GPR132	—
GPR139	—
GPR143	Nystagmus 6, congenital, X-linked (OMIM:300814); Albinism, ocular, type I (OMIM:300500); X-linked recessive ocular albinism (ORPHA:54)
GPR157	—
GPR179	Night blindness, congenital stationary, type 1E (OMIM:614565); ORPHA:714079
GPR37	—
GPR78	—
GPS1	—
GPSM2	Chudley-Mccullough syndrome (OMIM:604213); ORPHA:90636; ORPHA:314597
GPT	—
GRB10	Silver-Russell syndrome due to maternal uniparental disomy of chromosome 7 (ORPHA:96182)
GREM1	Hereditary mixed polyposis syndrome (ORPHA:157794)
GRHL1	—
GRHL2	Corneal dystrophy, posterior polymorphous, 4 (OMIM:618031); Deafness, autosomal dominant nonsyndromic sensorineural 28 (OMIM:608641); Ectodermal dysplasia/short stature syndrome (OMIM:616029); ORPHA:90635; ORPHA:423454; Posterior polymorphous corneal dystrophy (ORPHA:98973)
GRHPR	Hyperoxaluria, primary, type II (OMIM:260000); Primary hyperoxaluria type 2 (ORPHA:93599)
GRIA1	Intellectual developmental disorder, autosomal dominant 67 (OMIM:619927); Intellectual developmental disorder, autosomal recessive 76 (OMIM:619931); ORPHA:178469; ORPHA:88616
GRIA3	Intellectual developmental disorder, X-linked syndromic, Wu type (OMIM:300699); X-linked intellectual disability due to GRIA3 mutations (ORPHA:364028)
GRID1	—
GRID2	Spinocerebellar ataxia, autosomal recessive 18 (OMIM:616204); Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency (ORPHA:363432)
GRIK1	—
GRIK2	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures (OMIM:619580); Intellectual developmental disorder, autosomal recessive 6 (OMIM:611092); ORPHA:88616; ORPHA:528084
GRIK4	—
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive (OMIM:617820); Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant (OMIM:614254); Developmental and epileptic encephalopathy 101 (OMIM:619814); ORPHA:178469; Bilateral generalized polymicrogyria (ORPHA:208447); ORPHA:88616; Early infantile developmental and epileptic encephalopathy (ORPHA:1934)
GRIN2A	Epilepsy, focal, with speech disorder and with or without impaired intellectual development (OMIM:245570); Landau-Kleffner syndrome (ORPHA:98818); Rolandic epilepsy-speech dyspraxia syndrome (ORPHA:163721); Self-limited epilepsy with centrotemporal spikes (ORPHA:1945); Developmental and epileptic encephalopathy with spike-wave activation in sleep (ORPHA:725); Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation (ORPHA:289266)
GRIN2B	Intellectual developmental disorder, autosomal dominant 6, with or without seizures (OMIM:613970); Developmental and epileptic encephalopathy 27 (OMIM:616139); Infantile epileptic spasms syndrome (ORPHA:697160); ORPHA:589547
GRIN3A	—
GRIP1	Fraser syndrome 3 (OMIM:617667); Fraser syndrome (ORPHA:2052)
GRK1	Oguchi disease-2 (OMIM:613411); Oguchi disease (ORPHA:75382)
GRK3	—
GRK4	—
GRK6	—
GRM1	Spinocerebellar ataxia 44 (OMIM:617691); Spinocerebellar ataxia, autosomal recessive 13 (OMIM:614831); Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency (ORPHA:324262); ORPHA:404507; ORPHA:631095
GRM5	—
GRM6	Night blindness, congenital stationary, type 1B (OMIM:257270); ORPHA:714079
GRM7	Neurodevelopmental disorder with seizures, hypotonia, and brain imaging abnormalities (OMIM:618922); ORPHA:88616; Early infantile developmental and epileptic encephalopathy (ORPHA:1934)
GRN	Ceroid lipofuscinosis, neuronal, 11 (OMIM:614706); Frontotemporal lobar degeneration with TDP43 inclusions (OMIM:607485); Behavioral variant of frontotemporal dementia (ORPHA:275864); Progressive non-fluent aphasia (ORPHA:100070); Semantic dementia (ORPHA:100069); ORPHA:314629
GRPR	—
GRXCR1	Deafness, autosomal recessive 25 (OMIM:613285); ORPHA:90636
GS1	—
GSC	Short stature, auditory canal atresia, mandibular hypoplasia, andskeletal abnormalities (OMIM:602471); ORPHA:397623
GSDME	Deafness, autosomal dominant nonsyndromic sensorineural 5 (OMIM:600994); ORPHA:90635
GSE1	—
GSN	Amyloidosis, Finnish type (OMIM:105120); AGel amyloidosis (ORPHA:85448)
GSPT2	—
GSR	Hemolytic anemia due to glutathione reductase deficiency (OMIM:618660); ORPHA:90030
GSS	Glutathione synthetase deficiency (OMIM:266130); Hemolytic anemia due to glutathione synthetase deficiency

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	(OMIM:231900); ORPHA:289849; ORPHA:289846
GSTA2	—
GSTA3	—
GSTM1	—
GSTM2	—
GSTM5	—
GSTO1	—
GSTP1	—
GSTZ1	Maleylacetoacetate isomerase deficiency (OMIM:617596)
GTF2A1L	—
GTF2H5	Trichothiodystrophy 3, photosensitive (OMIM:616395); Trichothiodystrophy (ORPHA:33364)
GTF2I	Williams syndrome (ORPHA:904)
GTF2IRD1	Williams syndrome (ORPHA:904)
GTF3C2	—
GTPBP2	Jaberi-Elahi syndrome (OMIM:617988)
GUCA1A	Cone dystrophy 3 (OMIM:602093); Central areolar choroidal dystrophy (ORPHA:75377); Cone rod dystrophy (ORPHA:1872); Progressive cone dystrophy (ORPHA:1871)
GUCA1ANB	—
GUCA1B	Retinitis pigmentosa 48 (OMIM:613827); Retinitis pigmentosa (ORPHA:791)
GUCY2C	Diarrhea 6 (OMIM:614616); Meconium ileus (OMIM:614665); ORPHA:314373; ORPHA:314376; ORPHA:103908
GUCY2D	Leber congenital amaurosis, type I (OMIM:204000); Choroidal dystrophy, central areolar 1 (OMIM:215500); Cone-Rod dystrophy 6 (OMIM:601777); Night blindness, congenital stationary, type 1I (OMIM:618555); Leber congenital amaurosis (ORPHA:65); Central areolar choroidal dystrophy (ORPHA:75377); Cone rod dystrophy (ORPHA:1872)
GUCY2F	—
GUSB	Mucopolysaccharidosis VII (OMIM:253220); Mucopolysaccharidosis type 7 (ORPHA:584)
GUSBP3	—
GUSBP5	—
GYG1	Polyglucosan body myopathy 2 (OMIM:616199); Glycogen storage disease XV (OMIM:613507); Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency (ORPHA:263297); ORPHA:456369
GYG2	—
GYPA	OMIM:111300; OMIM:611162
GYPC	OMIM:616089; OMIM:611162; Hereditary elliptocytosis (ORPHA:288)
GYPE	—
GYS1	Glycogen storage disease 0, muscle (OMIM:611556); Glycogen storage disease due to muscle and heart glycogen synthase deficiency (ORPHA:137625)
GYS2	Glycogen storage disease 0, liver (OMIM:240600); Glycogen storage disease due to hepatic glycogen synthase deficiency (ORPHA:2089)
H1	—
H2BC4	—
H3	—
H6PD	Cortisone reductase deficiency 1 (OMIM:604931); Hyperandrogenism due to cortisone reductase deficiency (ORPHA:168588)
HABP2	Thyroid cancer, nonmedullary, 5 (OMIM:616535); Thrombophiliavenous thromboembolism, included (OMIM:188050); Familial papillary or follicular thyroid carcinoma (ORPHA:319487)
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures (OMIM:616756); Neuroblastoma (ORPHA:635); Spastic paraplegia-severe developmental delay-epilepsy syndrome (ORPHA:464282)
HACL1	—
HADH	3-Hydroxyacyl-Coa dehydrogenase deficiency (OMIM:231530); Hyperinsulinemic hypoglycemia, familial, 4 (OMIM:609975); Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency (ORPHA:71212)
HADHA	Mitochondrial trifunctional protein deficiency (OMIM:609015); Long-Chain 3-hydroxyacyl-coa dehydrogenase deficiency (OMIM:609016); Mitochondrial trifunctional protein deficiency (ORPHA:746); ORPHA:243367; Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (ORPHA:5)
HADHB	Mitochondrial trifunctional protein deficiency 2 (OMIM:620300); Mitochondrial trifunctional protein deficiency (ORPHA:746)
HAFML	—
HAL	Histidinemia (OMIM:235800); Histidinemia (ORPHA:2157)
HAMP	Hemochromatosis, type 2B (OMIM:613313); ORPHA:648581; HJV or HAMP-related hemochromatosis (ORPHA:79230)
HAND1	—
HAND2	Familial isolated dilated cardiomyopathy (ORPHA:154)
HAPLN1	—
HARS1	Charcot-Marie-Tooth disease, axonal, type 2W (OMIM:616625); Usher syndrome, type IIIB (OMIM:614504); Autosomal dominant Charcot-Marie-Tooth disease type 2W (ORPHA:488333); Usher syndrome type 3 (ORPHA:231183)
HARS2	Perrault syndrome 2 (OMIM:614926); ORPHA:642976; ORPHA:642945
HAVCR1	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
HAX1	Neutropenia, severe congenital 3, autosomal recessive (OMIM:610738); ORPHA:99749
HBB	BETA-THALASSEMIA (OMIM:613985); Fetal hemoglobin quantitative trait locus 1 (OMIM:141749); Methemoglobinemia, Beta type (OMIM:617971); Erythrocytosis 6 (OMIM:617980); Heinz body anemias (OMIM:140700); Sickle cell anemia (OMIM:603903); Thalassemia-beta, dominant inclusion-body (OMIM:603902); OMIM:611162; Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome (ORPHA:46532); ORPHA:330041; Sickle cell anemia (ORPHA:232); ORPHA:695147; ORPHA:695140; Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome (ORPHA:251380); ORPHA:251375; ORPHA:251370; ORPHA:251365; ORPHA:699822; Hemoglobin D disease (ORPHA:90039); ORPHA:715125; ORPHA:715128; ORPHA:715135; ORPHA:700090; ORPHA:715140; ORPHA:715143; ORPHA:715157; ORPHA:700107; ORPHA:700111; Hemoglobin E disease (ORPHA:2133); ORPHA:2132; Delta-beta-thalassemia (ORPHA:231237); Hemoglobin C-beta-thalassemia syndrome (ORPHA:231242); Hemoglobin E-beta-thalassemia syndrome (ORPHA:231249); Beta-thalassemia major (ORPHA:231214); Beta-thalassemia intermedia (ORPHA:231222); Unstable beta globin chain variant disease (ORPHA:231226); ORPHA:247511
HBD	ORPHA:330032; ORPHA:699822; Delta-beta-thalassemia (ORPHA:231237)
HBG2	Cyanosis, transient neonatal (OMIM:613977); Fetal hemoglobin quantitative trait locus 1 (OMIM:141749); Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome (ORPHA:46532); Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome (ORPHA:251380); ORPHA:707792; ORPHA:280615
HBM	—
HCCS	Linear skin defects with multiple congenital anomalies 1 (OMIM:309801); Microphthalmia with linear skin defects syndrome (ORPHA:2556)
HCFC1	Methylmalonic acidemia and homocysteinemia, Cblx type (OMIM:309541); ORPHA:369962; ORPHA:777
HCG4P8	—
HCN1	Epileptic encephalopathy, early infantile, 24 (OMIM:615871); Generalized epilepsy with febrile seizures plus, type 10 (OMIM:618482); Genetic epilepsy with febrile seizure plus (ORPHA:36387); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
HCN2	Febrile seizures, familial, 2 (OMIM:602477)
HCN3	—
HCN4	Epilepsy, idiopathic generalized, susceptibility to, 18 (OMIM:619521); Brugada syndrome 8 (OMIM:613123); Sick sinus syndrome 2 (OMIM:163800); Brugada syndrome (ORPHA:130); ORPHA:166282
HCRT	Narcolepsy 1 (OMIM:161400); Narcolepsy type 2 (ORPHA:83465); Narcolepsy type 1 (ORPHA:2073)
HCRTR2	—
HDAC4	Chromosome 2q37 deletion syndrome (OMIM:600430); Neurodevelopmental disorder with central hypotonia and dysmorphic facies (OMIM:619797); 2q37 microdeletion syndrome (ORPHA:1001)
HDAC6	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia (OMIM:300863); X-linked dominant chondrodysplasia, Chassaing-Lacombe type (ORPHA:163966)
HDAC8	Cornelia de Lange syndrome 5 (OMIM:300882); Cornelia de Lange syndrome (ORPHA:199); Wilson-Turner syndrome (ORPHA:3459)
HDAC9	Auriculocondylar syndrome 4 (OMIM:620457)
HDC	Gilles de la tourette syndrome (OMIM:137580)
HDGFL2	—
HDLBP	—
HDX	—
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A (OMIM:613925); Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development (OMIM:613926); Megalencephalic leukoencephalopathy with subcortical cysts 1 (OMIM:604004); Macrocephaly-intellectual disability-autism syndrome (ORPHA:210548); Megalencephalic leukoencephalopathy with subcortical cysts (ORPHA:2478)
HEPH	—
HERC1	Macrocephaly, dysmorphic facies, and psychomotor retardation (OMIM:617011); Megalencephaly-severe kyphoscoliosis-overgrowth syndrome (ORPHA:457359)
HES7	Spondylocostal dysostosis 4, autosomal recessive (OMIM:613686); Autosomal recessive spondylocostal dysostosis (ORPHA:2311)
HESX1	Septooptic dysplasia (OMIM:182230); Septo-optic dysplasia spectrum (ORPHA:3157); Pituitary stalk interruption syndrome (ORPHA:95496); Combined pituitary hormone deficiencies, genetic forms (ORPHA:95494); Hypothyroidism due to deficient transcription factors involved in pituitary development or function (ORPHA:226307); Kallmann syndrome (ORPHA:478)
HEXA	Tay-Sachs disease (OMIM:272800); ORPHA:309178; ORPHA:309192; ORPHA:309185
HEXB	Sandhoff disease (OMIM:268800); Sandhoff disease, juvenile form (ORPHA:309162); Sandhoff disease, adult form (ORPHA:309169); Sandhoff disease, infantile form (ORPHA:309155)
HEY1	—
HEY2	Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387)
HFE	Hemochromatosis, type 1 (OMIM:235200); ORPHA:443057; ORPHA:443062; Symptomatic form of HFE-related hemochromatosis (ORPHA:465508); Cystic fibrosis (ORPHA:586); ORPHA:648581
HGD	Alkaptonuria (OMIM:203500); Alkaptonuria (ORPHA:56)
HGF	Deafness, congenital neurosensory, autosomal recessive 39 (OMIM:608265); ORPHA:90636
HGSNAT	Mucopolysaccharidosis type IIIC (OMIM:252930); Retinitis pigmentosa 73 (OMIM:616544); Retinitis pigmentosa (ORPHA:791); ORPHA:79271
HIBCH	3-hydroxyisobutyryl-CoA hydrolase deficiency (OMIM:250620); Neurodegeneration due to 3-hydroxyisobutyryl-

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	CoA hydrolase deficiency (ORPHA:88639)
HIF1A	—
HIGD2A	—
HIGD2B	—
HINT1	Neuromyotonia and axonal neuropathy, autosomal recessive (OMIM:137200); Autosomal recessive axonal neuropathy with neuromyotonia (ORPHA:324442)
HIP1	—
HJV	Hemochromatosis, type 2A (OMIM:602390); ORPHA:648581; HJV or HAMP-related hemochromatosis (ORPHA:79230)
HK1	Hemolytic anemia, nonspherocytic, due to hexokinase deficiency (OMIM:235700); Retinitis pigmentosa 79 (OMIM:617460); Neurodevelopmental disorder with visual defects and brain anomalies (OMIM:618547); Neuropathy, hereditary motor and sensory, Russe type (OMIM:605285); Charcot-Marie-Tooth disease type 4G (ORPHA:99953); ORPHA:90031
HK2	—
HLA	—
HLCS	Holocarboxylase synthetase deficiency (OMIM:253270); Holocarboxylase synthetase deficiency (ORPHA:79242)
HLX	ORPHA:527468
HMBS	Porphyria, acute intermittent (OMIM:176000); Encephalopathy, porphyria-related (OMIM:620704); Leukoencephalopathy, porphyria-related (OMIM:620711); Acute intermittent porphyria (ORPHA:79276)
HMCN1	Macular degeneration, age-related, 1 (OMIM:603075)
HMGA2	Silver-Russell syndrome 5 (OMIM:618908); ORPHA:276148; ORPHA:99970; Well-differentiated liposarcoma (ORPHA:99971); Silver-Russell syndrome due to a point mutation (ORPHA:397590); 12q14 microdeletion syndrome (ORPHA:94063)
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency (OMIM:246450); 3-hydroxy-3-methylglutaric aciduria (ORPHA:20)
HMGCR	OMIM:620410; Muscular dystrophy, limb-girdle, autosomal recessive 28 (OMIM:620375); ORPHA:653725
HMGCS2	HMG-CoA synthase-2 deficiency (OMIM:605911); 3-hydroxy-3-methylglutaryl-CoA synthase deficiency (ORPHA:35701)
HMMR	Breast cancer (OMIM:114480)
HMOX1	Pulmonary disease, chronic obstructive (OMIM:606963); Heme oxygenase-1 deficiency (OMIM:614034); ORPHA:562509; Cystic fibrosis (ORPHA:586)
HMX1	Oculoauricular syndrome (OMIM:612109); ORPHA:157962
HMX2	—
HNF1A	Diabetes mellitus, noninsulin-dependent (OMIM:125853); Hepatic adenomas, familial (OMIM:142330); Diabetes mellitus, insulin-dependent-1 (OMIM:222100); Diabetes mellitus, insulin-dependent, 20 (OMIM:612520); Renal cell carcinoma, nonpapillary (OMIM:144700); Maturity-onset diabetes of the young, type III (OMIM:600496); Hyperinsulinism due to HNF1A deficiency (ORPHA:324575); ORPHA:319303; ORPHA:404511; MODY (ORPHA:552)
HNF1B	Renal cysts and diabetes syndrome (OMIM:137920); Diabetes mellitus, noninsulin-dependent (OMIM:125853); Renal cell carcinoma, nonpapillary (OMIM:144700); ORPHA:97364; ORPHA:97363; Mayer-Rokitansky-Küster-Hauser syndrome type 2 (ORPHA:2578); ORPHA:93172; ORPHA:93173; ORPHA:1331; HNF1B-related autosomal dominant tubulointerstitial kidney disease (ORPHA:93111); Medullary sponge kidney (ORPHA:1309); 17q12 microdeletion syndrome (ORPHA:261265)
HNF4A	Diabetes mellitus, noninsulin-dependent (OMIM:125853); Maturity-onset diabetes of the young, type 1 (OMIM:125850); Fanconi renotubular syndrome 4 with maturity-onset diabetes of the young (OMIM:616026); Congenital hyperinsulinism due to HNF4A deficiency (ORPHA:263455); ORPHA:544628; MODY (ORPHA:552)
HNMT	Asthma, susceptibility to (OMIM:600807); Intellectual developmental disorder, autosomal recessive 51 (OMIM:616739); ORPHA:88616
HNRNPH2	Intellectual developmental disorder, X-linked syndromic, Bain type (OMIM:300986); ORPHA:662198
HNRNPH3	—
HNRNPU	Epileptic encephalopathy, early infantile, 54 (OMIM:617391); 1q44 microdeletion syndrome (ORPHA:238769)
HNRNPUL2	—
HOGA1	Hyperoxaluria, primary, type III (OMIM:613616); Primary hyperoxaluria type 3 (ORPHA:93600)
HOOK2	—
HOTAIRM1	—
HOTTIP	—
HOXA	—
HOXA1	Athabaskan brainstem dysgenesis syndrome (OMIM:601536); ORPHA:69737; ORPHA:69739
HOXA10	—
HOXA11	Radioulnar synostosis with amegakaryocytic thrombocytopenia (OMIM:605432); Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome (ORPHA:71289)
HOXA13	Hand-foot-genital syndrome (OMIM:140000); Preaxial deficiency, postaxial polydactyly, and hypospadias (OMIM:176305); Hand-foot-genital syndrome (ORPHA:2438); ORPHA:2957
HOXA2	Microtia, hearing impairment, and cleft palate (OMIM:612290); Microtia (ORPHA:83463); ORPHA:140963
HOXA3	—
HOXA4	—
HOXB1	Facial paresis, hereditary congenital, 3 (OMIM:614744); ORPHA:306530
HOXB13	OMIM:610997; ORPHA:1331

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
HOXB3	—
HOXB6	—
HOXB7	—
HOXB8	—
HOXC13	Ectodermal dysplasia 9, hair/nail type (OMIM:614931); ORPHA:69084
HOXD10	Vertical talus, congenital (OMIM:192950); ORPHA:295203; ORPHA:295201
HOXD13	Brachydactyly-Syndactyly syndrome (OMIM:610713); Synpolydactyly 1 (OMIM:186000); Brachydactyly, type E (OMIM:113300); Syndactyly, type V (OMIM:186300); Brachydactyly, type D (OMIM:113200); VACTERL/VATER association (ORPHA:887); Brachydactyly type E (ORPHA:93387); Syndactyly type 5 (ORPHA:93406); Brachydactyly-syndactyly, Zhao type (ORPHA:93409); ORPHA:295191; ORPHA:295195
HOXD3	—
HOXD4	—
HP	OMIM:614081
HPCAL4	—
HPD	Hawkinsinuria (OMIM:140350); Tyrosinemia, type III (OMIM:276710); ORPHA:69723; Hawkinsinuria (ORPHA:2118)
HPGD	Hypertrophic osteoarthropathy, primary, autosomal recessive 1 (OMIM:259100); Digital clubbing, isolated congenital (OMIM:119900); Pachydermoperiostosis (ORPHA:2796); Cranio-osteoarthropathy (ORPHA:1525); Isolated nail clubbing (ORPHA:217059)
HPN	—
HPRT1	Lesch-Nyhan syndrome (OMIM:300322); Gout, hprt-related (OMIM:300323); Lesch-Nyhan syndrome (ORPHA:510); Hypoxanthine guanine phosphoribosyltransferase partial deficiency (ORPHA:79233)
HPS1	Hermansky-Pudlak syndrome 1 (OMIM:203300); ORPHA:231500
HPS3	Hermansky-Pudlak syndrome 3 (OMIM:614072); ORPHA:231512
HPS4	Hermansky-Pudlak syndrome 4 (OMIM:614073); ORPHA:231500
HPS5	Hermansky-Pudlak syndrome 5 (OMIM:614074); ORPHA:231512
HPS6	Hermansky-Pudlak syndrome 6 (OMIM:614075); ORPHA:231512
HPSE2	Urofacial syndrome (OMIM:236730); Urofacial syndrome (ORPHA:2704)
HR	Atrichia with papular lesions (OMIM:209500); Alopecia universalis congenita (OMIM:203655); ORPHA:86819; Alopecia universalis (ORPHA:701); Marie Unna hereditary hypotrichosis (ORPHA:444)
HRAS	Epidermal nevus, somatic (OMIM:162900); Costello syndrome (OMIM:218040); Thyroid cancer, nonmedullary, 2 (OMIM:188470); Schimmelpenning-Feuerstein-Mims syndrome (OMIM:163200); Bladder cancer (OMIM:109800); Melanocytic nevus syndrome, congenital (OMIM:137550); Woolly hair nevus (ORPHA:79414); Phakomatosis pigmentokeratotica (ORPHA:2874); ORPHA:146; Linear nevus sebaceus syndrome (ORPHA:2612); Costello syndrome (ORPHA:3071)
HRAT17	—
HRG	Thrombophilia due to histidine-rich glycoprotein deficiency (OMIM:613116); ORPHA:217467
HSBP1	—
HSD11B1	Cortisone reductase deficiency 2 (OMIM:614662); Hyperandrogenism due to cortisone reductase deficiency (ORPHA:168588)
HSD11B2	Apparent mineralocorticoid excess (OMIM:218030); Apparent mineralocorticoid excess (ORPHA:320)
HSD17B10	HSD10 mitochondrial disease (OMIM:300438); HSD10 disease, atypical type (ORPHA:85295); HSD10 disease, neonatal type (ORPHA:391457); HSD10 disease, infantile type (ORPHA:391428)
HSD17B3	Pseudohermaphroditism, male, with gynecomastia (OMIM:264300); 46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency (ORPHA:752)
HSD17B4	D-bifunctional protein deficiency (OMIM:261515); Perrault syndrome 1 (OMIM:233400); ORPHA:300; ORPHA:642976; ORPHA:642945
HSD3B1	—
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency (OMIM:201810); Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency (ORPHA:90791)
HSD3B7	Bile acid synthesis defect, congenital, 1 (OMIM:607765); Congenital bile acid synthesis defect type 1 (ORPHA:79301)
HSF1	—
HSF4	Cataract, lamellar (OMIM:116800); ORPHA:98994; ORPHA:441452
HSP90AA1	—
HSPA5	—
HSPA9	Anemia, sideroblastic, autosomal dominant (OMIM:182170); Even-Plus syndrome (OMIM:616854); ORPHA:260305; ORPHA:496751
HSPB1	Neuronopathy, distal hereditary motor, type IIB (OMIM:608634); Charcot-Marie-Tooth disease, axonal, type 2F (OMIM:606595); Autosomal dominant Charcot-Marie-Tooth disease type 2F (ORPHA:99940); ORPHA:139525
HSPB3	Neuronopathy, distal hereditary motor, type IIC (OMIM:613376); ORPHA:139525
HSPB6	—
HSPB7	—
HSPB8	Myopathy, myofibrillar, 13, with rimmed vacuoles (OMIM:621078); Neuronopathy, distal hereditary motor, type IIA (OMIM:158590); Charcot-Marie-Tooth disease, axonal, type 2L (OMIM:608673); ORPHA:99945; ORPHA:139525; ORPHA:476093
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type (OMIM:224410); Schwartz-jampel syndrome, type 1 (OMIM:255800); Dyssegmental dysplasia, Silverman-Handmaker type (ORPHA:1865); Schwartz-Jampel syndrome (ORPHA:800); 1p36 deletion syndrome (ORPHA:1606)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
HTN3	—
HTR1A	Periodic fever, menstrual cycle-dependent (OMIM:614674); ORPHA:498251
HTR1B	—
HTR2A	Obsessive-Compulsive disorder 1 (OMIM:164230); Major depressive disorder (OMIM:608516); Schizophrenia (OMIM:181500)
HTR2B	—
HTR2C	—
HTR3A	—
HTR3B	—
HTR3C	—
HTR7	—
HTRA1	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2 (OMIM:616779); Autosomal recessive cerebral arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL) (OMIM:600142); Cerebral autosomal recessive arteriopathy-subcortical infarcts-leukoencephalopathy (ORPHA:199354); ORPHA:482077; ORPHA:252212; ORPHA:252128
HTRA2	Parkinson disease 13 (OMIM:610297); 3-methylglutaconic aciduria, type VIII (OMIM:617248); Young-onset Parkinson disease (ORPHA:2828); ORPHA:505208
HTT	Huntington disease (OMIM:143100); Lopes-Maciel-Rodan syndrome (OMIM:617435); Huntington disease (ORPHA:399); ORPHA:528084; Juvenile Huntington disease (ORPHA:248111)
HUWE1	Intellectual developmental disorder, X-linked syndromic, Turner type (OMIM:309590); ORPHA:528084
HVCN1	—
HYAL1	Mucopolysaccharidosis type IX (OMIM:601492); Hyaluronidase deficiency (ORPHA:67041)
HYCC1	Leukodystrophy, hypomyelinating, 5 (OMIM:610532); Hypomyelination-congenital cataract syndrome (ORPHA:85163)
HYDIN	Ciliary dyskinesia, primary, 5 (OMIM:608647); Primary ciliary dyskinesia (ORPHA:244)
HYLS1	Hydrolethalus syndrome 1 (OMIM:236680); Isolated Joubert syndrome (ORPHA:475); Hydrolethalus (ORPHA:2189)
IAH1	—
IAPP	—
ICAM1	OMIM:611162
ICAM4	OMIM:111250
ICOS	Immunodeficiency, common variable, 1 (OMIM:607594); Immunodeficiency, common variable, 2 (OMIM:240500); ORPHA:695183
ID3	—
ID4	—
IDH1	Glioma susceptibility 1 (OMIM:137800); ORPHA:251579; ORPHA:251576; Ollier disease (ORPHA:296); ORPHA:86845; Maffucci syndrome (ORPHA:163634); Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria (ORPHA:99646)
IDH2	D-2-hydroxyglutaric aciduria 2 (OMIM:613657); ORPHA:251598; ORPHA:251589; Ollier disease (ORPHA:296); ORPHA:86845; Maffucci syndrome (ORPHA:163634); D-2-hydroxyglutaric aciduria (ORPHA:79315); ORPHA:251663; ORPHA:251656; ORPHA:251627; ORPHA:251630; ORPHA:251604; ORPHA:251601
IDH3B	Retinitis pigmentosa 46 (OMIM:612572); Retinitis pigmentosa (ORPHA:791)
IDO1	—
IDS	Mucopolysaccharidosis, type II (OMIM:309900); ORPHA:217093; ORPHA:217085
IDUA	Hurler syndrome (OMIM:607014); Scheie syndrome (OMIM:607016); Hurler-Scheie syndrome (OMIM:607015); Scheie syndrome (ORPHA:93474); Hurler syndrome (ORPHA:93473); Hurler-Scheie syndrome (ORPHA:93476)
IER3	—
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome (OMIM:614231); ORPHA:306558
IFIH1	Singleton-Merten syndrome 1 (OMIM:182250); Aicardi-Goutieres syndrome 7 (OMIM:615846); Immunodeficiency 95 (OMIM:619773); Aicardi-Goutières syndrome (ORPHA:51); ORPHA:689231; ORPHA:85191
IFITM3	OMIM:614680
IFITM5	Osteogenesis imperfecta, type V (OMIM:610967); ORPHA:216828
IFNAR1	Immunodeficiency 106, susceptibility to viral infections (OMIM:619935)
IFNAR2	OMIM:610424; Immunodeficiency 45 (OMIM:616669); ORPHA:431166
IFNG	OMIM:609423; Aplastic anemia (OMIM:609135); Immunodeficiency 69, mycobacteriosis (OMIM:618963); OMIM:609532; Tuberous sclerosis-2 (OMIM:613254); OMIM:607948; ORPHA:699618; Idiopathic aplastic anemia (ORPHA:88); Tuberous sclerosis complex (ORPHA:805)
IFNGR1	OMIM:610424; Immunodeficiency 27A, mycobacteriosis, AR (OMIM:209950); Immunodeficiency 27B (OMIM:615978); OMIM:607948; Helicobacter pylori infection, susceptibility to (OMIM:600263); ORPHA:99898; Behçet disease (ORPHA:117); ORPHA:319569; ORPHA:319581
IFNGR2	Immunodeficiency 28 (OMIM:614889); ORPHA:319574; ORPHA:319547; ORPHA:319589
IFNL4	—
IFRD1	Spinocerebellar ataxia type 18 (ORPHA:98771)
IFT122	Cranioectodermal dysplasia (OMIM:218330); Cranioectodermal dysplasia (ORPHA:1515); ORPHA:93268
IFT140	Retinitis pigmentosa 80 (OMIM:617781); Polycystic kidney disease 9, susceptibility to (OMIM:621164); Cranioectodermal dysplasia 5 (OMIM:621180); Short-rib thoracic dysplasia 9 with or without polydactyly

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	(OMIM:266920); Leber congenital amaurosis (ORPHA:65); Retinitis pigmentosa (ORPHA:791); Autosomal dominant polycystic kidney disease (ORPHA:730); ORPHA:140969; Jeune syndrome (ORPHA:474)
IFT172	Retinitis pigmentosa 71 (OMIM:616394); Short-Rib thoracic dysplasia 10 with or without polydactyly (OMIM:615630); Bardet-Biedl syndrome 20 (OMIM:619471); Bardet-Biedl syndrome (ORPHA:110); Retinitis pigmentosa (ORPHA:791); ORPHA:140969; Jeune syndrome (ORPHA:474)
IFT43	Short-Rib thoracic dysplasia 18 with polydactyly (OMIM:617866); Retinitis pigmentosa 81 (OMIM:617871); Cranioectodermal dysplasia 3 (OMIM:614099); Cranioectodermal dysplasia (ORPHA:1515); Retinitis pigmentosa (ORPHA:791)
IFT80	Asphyxiating thoracic dystrophy 2 (OMIM:611263); Short rib-polydactyly syndrome, Verma-Naumoff type (ORPHA:93271); ORPHA:93268; Jeune syndrome (ORPHA:474)
IFT88	Retinitis pigmentosa (ORPHA:791)
IGBP1	Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia (OMIM:300472); Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome (ORPHA:52055)
IGF1	Insulin-Like growth factor I deficiency (OMIM:608747); Growth delay due to insulin-like growth factor type 1 deficiency (ORPHA:73272)
IGF1R	Insulin-Like growth factor I, resistance to (OMIM:270450); Growth delay due to insulin-like growth factor I resistance (ORPHA:73273)
IGF2	Silver-Russell syndrome 3 (OMIM:616489); Silver-Russell syndrome 1 (OMIM:180860); Beckwith-Wiedemann syndrome (OMIM:130650); Wilms tumor 1 (OMIM:194070); Silver-Russell syndrome due to an imprinting defect of 11p15 (ORPHA:231140); Silver-Russell syndrome due to 11p15 microduplication (ORPHA:231144); ORPHA:231117; Silver-Russell syndrome due to a point mutation (ORPHA:397590); Isolated hemihyperplasia (ORPHA:2128)
IGF2BP2	Diabetes mellitus, noninsulin-dependent (OMIM:125853)
IGF2R	Hepatocellular carcinoma (OMIM:114550)
IGFALS	Acid-labile subunit, deficiency of (OMIM:615961); Short stature due to primary acid-labile subunit deficiency (ORPHA:140941)
IGFBP7	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis (OMIM:614224); ORPHA:284247
IGHMBP2	Spinal muscular atrophy, distal, autosomal recessive, 1 (OMIM:604320); Charcot-Marie-Tooth disease, axonal, type 2S (OMIM:616155); ORPHA:443073; ORPHA:98920
IGLL1	Agammaglobulinemia 2, autosomal recessive (OMIM:613500); Autosomal non-syndromic agammaglobulinemia (ORPHA:33110)
IGSF1	Hypothyroidism, central, and testicular enlargement (OMIM:300888); ORPHA:329235
IHH	Brachydactyly, type A1 (OMIM:112500); Acrocapitofemoral dysplasia (OMIM:607778); Acrocapitofemoral dysplasia (ORPHA:63446); Brachydactyly type A1 (ORPHA:93388)
IK	—
IKBIP	—
IKBKB	Immunodeficiency 15 (OMIM:615592); Immunodeficiency 15A (OMIM:618204); ORPHA:700205; ORPHA:397787
IKBKG	Incontinentia pigmenti (OMIM:308300); Autoinflammatory disease, systemic, X-linked (OMIM:301081); Ectodermal dysplasia and immunodeficiency 1 (OMIM:300291); Immunodeficiency 33 (OMIM:300636); ORPHA:699605; Hypohidrotic ectodermal dysplasia with immunodeficiency (ORPHA:98813); Incontinentia pigmenti (ORPHA:464); ORPHA:69088
IKZF1	Immunodeficiency, common variable, 13 (OMIM:616873); ORPHA:695172; Stevens-Johnson syndrome (ORPHA:36426); ORPHA:585909; ORPHA:317473; ORPHA:697414
IKZF3	Immunodeficiency 84 (OMIM:619437); ORPHA:699593; ORPHA:699590; ORPHA:67038
IL10	OMIM:609423; OMIM:614395; Rheumatoid arthritis (OMIM:180300); ORPHA:238569; Behçet disease (ORPHA:117); Systemic lupus erythematosus (ORPHA:536)
IL10RA	Inflammatory bowel disease 28, early onset, autosomal recessive (OMIM:613148); ORPHA:238569
IL10RB	OMIM:610424; Inflammatory bowel disease 25, early onset, autosomal recessive (OMIM:612567); ORPHA:238569
IL11	—
IL11RA	Craniosynostosis and dental anomalies (OMIM:614188); ORPHA:284149
IL12B	Immunodeficiency 29 (OMIM:614890); Takayasu arteritis (ORPHA:3287); ORPHA:319558
IL12RB1	Immunodeficiency 30 (OMIM:614891); Primary biliary cholangitis (ORPHA:186); Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency (ORPHA:319552)
IL12RB2	—
IL13	Asthma, susceptibility to (OMIM:600807); OMIM:607154
IL17F	Candidiasis, familial, 6, autosomal dominant (OMIM:613956); Chronic mucocutaneous candidiasis (ORPHA:1334)
IL17RA	Immunodeficiency 51 (OMIM:613953); Chronic mucocutaneous candidiasis (ORPHA:1334)
IL18BP	Hepatitis, fulminant viral, susceptibility to (OMIM:618549)
IL18RAP	—
IL19	—
IL1B	Gastric cancer, somatic (OMIM:613659)
IL1R1	Chronic recurrent multifocal osteomyelitis (OMIM:259680)
IL1RAPL1	Intellectual developmental disorder, X-linked 21 (OMIM:300143); ORPHA:777
IL1RN	Interleukin 1 receptor antagonist deficiency (OMIM:612852); OMIM:612628; Gastric cancer, somatic (OMIM:613659); ORPHA:210115

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
IL20RB	—
IL21R	Immunodeficiency 56 (OMIM:615207); ORPHA:357329
IL23R	OMIM:605606; OMIM:612261; Behçet disease (ORPHA:117)
IL2RA	Immunodeficiency 41 with lymphoproliferation and autoimmunity (OMIM:606367); Diabetes mellitus, insulin-dependent, 10 (OMIM:601942); Oligoarticular juvenile idiopathic arthritis (ORPHA:85410); Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis (ORPHA:85408); ORPHA:169100
IL2RG	Severe combined immunodeficiency, X-linked (OMIM:300400); Combined immunodeficiency, X-linked (OMIM:312863); T-B+ severe combined immunodeficiency due to gamma chain deficiency (ORPHA:276); Omenn syndrome (ORPHA:39041)
IL3	—
IL31RA	Amyloidosis, primary localized cutaneous, 2 (OMIM:613955); ORPHA:353220
IL36RN	Psoriasis 14, pustular (OMIM:614204); Generalized pustular psoriasis (ORPHA:247353); ORPHA:163927; ORPHA:163931; ORPHA:404546
IL4	—
IL4I1	—
IL4R	—
IL6	Diabetes mellitus, noninsulin-dependent (OMIM:125853); OMIM:604302; Arteriovenous malformation of the brain, somatic (OMIM:108010); Inflammatory bowel disease 1, Crohn disease (OMIM:266600); Diabetes mellitus, insulin-dependent-1 (OMIM:222100); Kaposi sarcoma, susceptibility to (OMIM:148000); Systemic-onset juvenile idiopathic arthritis (ORPHA:85414)
IL6R	Hyper-IgE recurrent infection syndrome 5, autosomal recessive (OMIM:618944); OMIM:614752; OMIM:614689; ORPHA:656326
IL6ST	Hyper-IgE recurrent infection syndrome 4, autosomal recessive (OMIM:618523); Stuve-Wiedemann syndrome 2 (OMIM:619751); Hyper-IgE recurrent infection syndrome 4A, autosomal dominant (OMIM:619752); Immunodeficiency 94 with autoinflammation and dysmorphic facies (OMIM:619750); ORPHA:656300; ORPHA:656313; ORPHA:656283
IL7R	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-positive (OMIM:608971); T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency (ORPHA:169154); Omenn syndrome (ORPHA:39041)
IL9R	—
ILDR1	Deafness, neurosensory, autosomal recessive 42 (OMIM:609646); ORPHA:90636
ILK	—
IMMP2L	—
IMPDH1	Retinitis pigmentosa 10 (OMIM:180105); Leber congenital amaurosis 11 (OMIM:613837); Leber congenital amaurosis (ORPHA:65); Retinitis pigmentosa (ORPHA:791)
IMPDH2	OMIM:617995; Autosomal dominant dopa-responsive dystonia (ORPHA:98808)
IMPG2	Macular dystrophy, vitelliform, 5 (OMIM:616152); Retinitis pigmentosa 56 (OMIM:613581); Adult-onset foveomacular vitelliform dystrophy (ORPHA:99000); Retinitis pigmentosa (ORPHA:791)
INF2	Glomerulosclerosis, focal segmental, 5 (OMIM:613237); Charcot-Marie-Tooth disease, dominant intermediate E (OMIM:614455); ORPHA:93114; Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
ING1	Squamous cell carcinoma, head and neck (OMIM:275355); ORPHA:494547; ORPHA:494550; ORPHA:502363; ORPHA:502366; ORPHA:500478; ORPHA:500464
INHA	—
INMT	—
INPP5B	—
INPP5D	—
INPP5E	Joubert syndrome 1 (OMIM:213300); Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome (OMIM:610156); Joubert syndrome with ocular defect (ORPHA:220493); MORM syndrome (ORPHA:75858); Joubert syndrome with hepatic defect (ORPHA:1454); Isolated Joubert syndrome (ORPHA:475)
INS	Diabetes mellitus, insulin-dependent, 2 (OMIM:125852); Hyperproinsulinemia (OMIM:616214); Diabetes mellitus, permanent neonatal 4 (OMIM:618858); Maturity-onset diabetes of the young, type 10 (OMIM:613370); Isolated permanent neonatal diabetes mellitus (ORPHA:99885); MODY (ORPHA:552)
INSL3	Cryptorchidism, unilateral or bilateral (OMIM:219050)
INSL6	—
INSR	Pineal hyperplasia, insulin-resistant diabetes mellitus, and somatic abnormalities (OMIM:262190); Diabetes mellitus, insulin-resistant, with acanthosis nigricans (OMIM:610549); Donohue syndrome (OMIM:246200); Hyperinsulinemic hypoglycemia, familial, 5 (OMIM:609968); Hyperinsulinism due to INSR deficiency (ORPHA:263458); Rabson-Mendenhall syndrome (ORPHA:769); Donohue syndrome (ORPHA:508); Insulin-resistance syndrome type A (ORPHA:2297)
INSRR	—
INVS	Nephronophthisis 2 (OMIM:602088); Senior-Loken syndrome (ORPHA:3156); ORPHA:93591
IPCEF1	—
IQANK1	—
IQCB1	Senior-Loken syndrome 5 (OMIM:609254); Senior-Loken syndrome (ORPHA:3156); Leber congenital amaurosis (ORPHA:65)
IQCG	—
IQGAP2	—
IQGAP3	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
IQSEC2	Intellectual developmental disorder, X-linked 1 (OMIM:309530); OMIM:301164; Microduplication Xp11.22p11.23 syndrome (ORPHA:217377); Smith-Magenis syndrome (ORPHA:819); Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome (ORPHA:397933)
IRAK1BP1	—
IRAK3	OMIM:611064
IRAK4	Immunodeficiency 67 (OMIM:607676); Transient predisposition to invasive pyogenic bacterial infection (ORPHA:70592)
IRF1	Immunodeficiency 117, mycobacteriosis, autosomal recessive (OMIM:620668); Lung cancer, susceptibility to (OMIM:211980); Gastric cancer, somatic (OMIM:613659); ORPHA:699615
IRF4	OMIM:611724; Immunodeficiency 131 (OMIM:621097); Whipple disease (ORPHA:3452)
IRF5	OMIM:612245; OMIM:612251; Limited cutaneous systemic sclerosis (ORPHA:220402); Diffuse cutaneous systemic sclerosis (ORPHA:220393); Primary biliary cholangitis (ORPHA:186); Systemic lupus erythematosus (ORPHA:536)
IRF6	van der Woude syndrome 1 (OMIM:119300); Orofacial cleft 6, susceptibility to (OMIM:608864); Popliteal pterygium syndrome (OMIM:119500); Isolated cleft lip (ORPHA:199302); Cleft lip/palate (ORPHA:199306); Van der Woude syndrome (ORPHA:888); Autosomal dominant popliteal pterygium syndrome (ORPHA:1300); ORPHA:708014; Cleft lip and alveolus (ORPHA:141291); Oligodontia (ORPHA:99798)
IRF8	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive (OMIM:226990); Immunodeficiency 32A (OMIM:614893); Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency (ORPHA:319600)
IRGM	OMIM:607948; Inflammatory bowel disease 19 (OMIM:612278)
IRS1	Diabetes mellitus, noninsulin-dependent (OMIM:125853)
IRS2	Diabetes mellitus, noninsulin-dependent (OMIM:125853)
IRS4	Hypothyroidism, congenital, nongoitrous, 9 (OMIM:301035)
IRX4	—
IRX5	Hamamy syndrome (OMIM:611174); ORPHA:314555
ISCU	Myopathy with exercise intolerance, Swedish type (OMIM:255125); ORPHA:43115
ISL1	Classic bladder exstrophy (ORPHA:93930)
IT1	—
ITCH	Autoimmune disease, multisystem, with facial dysmorphism (OMIM:613385); Syndromic multisystem autoimmune disease due to Itch deficiency (ORPHA:228426)
ITFG1	—
ITFG2	—
ITGA1	—
ITGA2	OMIM:621267; Fetal and neonatal alloimmune thrombocytopenia (ORPHA:853)
ITGA2B	OMIM:621266; Glanzmann thrombasthenia (OMIM:273800); Bleeding disorder, platelet-type, 16 (OMIM:187800); Fetal and neonatal alloimmune thrombocytopenia (ORPHA:853); Glanzmann thrombasthenia (ORPHA:849); Autosomal dominant macrothrombocytopenia (ORPHA:140957)
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital (OMIM:614748); ORPHA:306504
ITGA4	—
ITGA6	Epidermolysis bullosa, junctional, with pyloric atresia (OMIM:226730); Epidermolysis bullosa, junctional 6, with pyloric atresia (OMIM:619817); Junctional epidermolysis bullosa with pyloric atresia (ORPHA:79403)
ITGA7	Muscular dystrophy, congenital, due to integrin alpha-7 deficiency (OMIM:613204); ORPHA:34520; Congenital fiber-type disproportion myopathy (ORPHA:2020)
ITGA9	—
ITGAM	Systemic lupus erythematosus (ORPHA:536)
ITGB1	—
ITGB2	Leukocyte adhesion deficiency, type I (OMIM:116920); ORPHA:99842
ITGB3	Glanzmann thrombasthenia 2 (OMIM:619267); OMIM:621264; Bleeding disorder, platelet-type, 24, autosomal dominant (OMIM:619271); Fetal and neonatal alloimmune thrombocytopenia (ORPHA:853); Glanzmann thrombasthenia (ORPHA:849); Autosomal dominant macrothrombocytopenia (ORPHA:140957)
ITGB3BP	—
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia (OMIM:226730); Epidermolysis bullosa, junctional 5A, intermediate (OMIM:619816); Junctional epidermolysis bullosa with pyloric atresia (ORPHA:79403); Intermediate generalized junctional epidermolysis bullosa (ORPHA:79402); Epidermolysis bullosa simplex with pyloric atresia (ORPHA:158684); Localized junctional epidermolysis bullosa (ORPHA:251393); Aplasia cutis congenita (ORPHA:1114)
ITGB6	Amelogenesis imperfecta, type IH (OMIM:616221); Alopecia-intellectual disability syndrome (ORPHA:2850); ORPHA:100031; ORPHA:100032
ITIH4	—
ITIH6	—
ITK	Lymphoproliferative syndrome 1 (OMIM:613011); ORPHA:538963
ITM2B	Cerebral amyloid angiopathy, itm2b-related, 1 (OMIM:176500); Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities (OMIM:616079); Dementia, familial danish (OMIM:117300); ORPHA:97346; ORPHA:97345; ORPHA:397758
ITPA	Epileptic encephalopathy, early infantile, 35 (OMIM:616647); Inosine triphosphatase deficiency (OMIM:613850); ORPHA:457375

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ITPKC	—
ITPR1	Gillespie syndrome (OMIM:206700); Spinocerebellar ataxia 15 (OMIM:606658); Spinocerebellar ataxia 29, congenital nonprogressive (OMIM:117360); Spinocerebellar ataxia type 29 (ORPHA:208513); Spinocerebellar ataxia type 15/16 (ORPHA:98769); Aniridia-cerebellar ataxia-intellectual disability syndrome (ORPHA:1065)
ITPR2	Anhidrosis, isolated, with normal sweat glands (OMIM:106190); ORPHA:468666
ITSN2	—
IVD	Isovaleric acidemia (OMIM:243500); Isovaleric acidemia (ORPHA:33)
IYD	Thyroid hormonogenesis, genetic defect in, 4 (OMIM:274800); Familial thyroid dyshormonogenesis (ORPHA:95716)
JAG1	Deafness, congenital heart defects, and posterior embryotoxon (OMIM:617992); Charcot-Marie-Tooth disease, axonal, type 2HH (OMIM:619574); Tetralogy of Fallot (OMIM:187500); Alagille syndrome 1 (OMIM:118450); ORPHA:261619; Tetralogy of Fallot (ORPHA:3303); ORPHA:261600
JAG2	Muscular dystrophy, limb-girdle, autosomal recessive 27 (OMIM:619566)
JAK2	Budd-Chiari syndrome (OMIM:600880); Erythrocytosis, familial, 1 (OMIM:133100); Myelofibrosis with myeloid metaplasia, somatic (OMIM:254450); Thrombocythemia 3 (OMIM:614521); Polycythemia vera (OMIM:263300); Leukemia, acute myeloid (OMIM:601626); Familial thrombocytosis (ORPHA:71493); Essential thrombocythemia (ORPHA:3318); Budd-Chiari syndrome (ORPHA:131); ORPHA:667662; Primary myelofibrosis (ORPHA:824); Polycythemia vera (ORPHA:729)
JAK3	Scid, autosomal recessive, T-Negative/b-Positive type (OMIM:600802); T-B+ severe combined immunodeficiency due to JAK3 deficiency (ORPHA:35078)
JAM3	Hemorrhagic destruction of the brain, subependymal calcification,and cataracts (OMIM:613730); ORPHA:306547
JCAD	—
JMJD1C	ORPHA:91352; 22q11.2 deletion syndrome (ORPHA:567)
JPH2	Cardiomyopathy, dilated, 2E (OMIM:619492); Cardiomyopathy, familial hypertrophic, 17 (OMIM:613873); Familial isolated dilated cardiomyopathy (ORPHA:154)
JPH3	Huntington disease-like 2 (OMIM:606438); Huntington disease-like 2 (ORPHA:98934)
JRK	Juvenile myoclonic epilepsy (ORPHA:307); Childhood absence epilepsy (ORPHA:64280)
JUN	—
JUNB	—
JUP	Arrhythmogenic right ventricular dysplasia 12 (OMIM:611528); Naxos disease (OMIM:601214); Lethal acantholytic erosive disorder (ORPHA:158687); ORPHA:293888; ORPHA:293899; ORPHA:293910; Naxos disease (ORPHA:34217)
KALRN	—
KANK1	Cerebral palsy, spastic quadriplegic, 2 (OMIM:612900); ORPHA:210141
KARS1	Deafness, congenital, and adult-onset progressive leukoencephalopathy (OMIM:619196); Leukoencephalopathy, progressive, infantile-onset, with or without deafness (OMIM:619147); Deafness, autosomal recessive 89 (OMIM:613916); Charcot-marie-tooth disease, recessive intermediate B (OMIM:613641); ORPHA:90636; Early-onset progressive leukoencephalopathy-central nervous system calcification-deafness-visual impairment syndrome (ORPHA:3240); ORPHA:254334; ORPHA:652532
KAT2A	—
KAT6A	Arboleda-Tham syndrome (OMIM:616268); KAT6-related intellectual disability-craniofacial anomalies-cardiac defects syndrome (ORPHA:457193); ORPHA:370026
KAT6B	Genitopatellar syndrome (OMIM:606170); Ohdo syndrome, Say-Barber-Biesecker-Young-Simpson variant (OMIM:603736); Genitopatellar syndrome (ORPHA:85201); Blepharophimosis-intellectual disability syndrome, SBBYS type (ORPHA:3047)
KATNAL2	—
KBTBD11	—
KBTBD13	Nemaline myopathy 6 (OMIM:609273); Childhood-onset nemaline myopathy (ORPHA:171439)
KBTBD6	—
KCNA1	Episodic ataxia, type 1 (OMIM:160120); ORPHA:199326; Episodic ataxia type 1 (ORPHA:37612); Hereditary continuous muscle fiber activity (ORPHA:972); Paroxysmal kinesigenic dyskinesia (ORPHA:98809); Early infantile developmental and epileptic encephalopathy (ORPHA:1934)
KCNA4	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum (OMIM:618284)
KCNA5	Atrial fibrillation, familial, 7 (OMIM:612240); Hereditary atrial fibrillation (ORPHA:334)
KCNA6	—
KCNAB1	—
KCNAB2	1p36 deletion syndrome (ORPHA:1606)
KCNC3	Spinocerebellar ataxia 13 (OMIM:605259); Spinocerebellar ataxia type 13 (ORPHA:98768)
KCND2	—
KCND3	Brugada syndrome 9 (OMIM:616399); Spinocerebellar ataxia 19 (OMIM:607346); Brugada syndrome (ORPHA:130); Spinocerebellar ataxia type 19/22 (ORPHA:98772)
KCNE1	Jervell and Lange-Nielsen syndrome 2 (OMIM:612347); Long QT syndrome 5 (OMIM:613695); Romano-Ward syndrome (ORPHA:101016); Hereditary atrial fibrillation (ORPHA:334); Jervell and Lange-Nielsen syndrome (ORPHA:90647)
KCNE2	Atrial fibrillation, familial, 4 (OMIM:611493); Long QT syndrome 6 (OMIM:613693); Romano-Ward syndrome (ORPHA:101016); Hereditary atrial fibrillation (ORPHA:334)
KCNE3	Brugada syndrome 6 (OMIM:613119); Brugada syndrome (ORPHA:130); Hypokalemic periodic paralysis

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	(ORPHA:681)
KCNE4	—
KCNE5	Brugada syndrome (ORPHA:130); Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome (ORPHA:86818)
KCNH2	Short QT syndrome 1 (OMIM:609620); Long QT syndrome 2 (OMIM:613688); Congenital short QT syndrome (ORPHA:51083); Romano-Ward syndrome (ORPHA:101016)
KCNH3	—
KCNIP1	—
KCNIP4	—
KCNJ1	Bartter syndrome, type 2 (OMIM:241200); ORPHA:620220
KCNJ10	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct (OMIM:600791); SESAME syndrome (OMIM:612780); Pendred syndrome (OMIM:274600); EAST syndrome (ORPHA:199343); Paroxysmal kinesigenic dyskinesia (ORPHA:98809); Pendred syndrome (ORPHA:705)
KCNJ11	Diabetes mellitus, noninsulin-dependent (OMIM:125853); Maturity-onset diabetes of the young, type 13 (OMIM:616329); Diabetes, permanent neonatal 2, with or without neurologic features (OMIM:618856); Diabetes mellitus, transient neonatal, 3 (OMIM:610582); Hyperinsulinemic hypoglycemia, familial, 2 (OMIM:601820); Isolated permanent neonatal diabetes mellitus (ORPHA:99885); Transient neonatal diabetes mellitus (ORPHA:99886); ORPHA:99989; Autosomal recessive hyperinsulinism due to Kir6.2 deficiency (ORPHA:79644); Autosomal dominant hyperinsulinism due to Kir6.2 deficiency (ORPHA:276580); ORPHA:276603; MODY (ORPHA:552); DEND syndrome (ORPHA:79134)
KCNJ12	—
KCNJ13	Leber congenital amaurosis 16 (OMIM:614186); Snowflake vitreoretinal degeneration (OMIM:193230); Leber congenital amaurosis (ORPHA:65); ORPHA:91496
KCNJ2	Atrial fibrillation, familial, 9 (OMIM:613980); Short QT syndrome 3 (OMIM:609622); Andersen cardiodysrhythmic periodic paralysis (OMIM:170390); Congenital short QT syndrome (ORPHA:51083); Hereditary atrial fibrillation (ORPHA:334); Andersen-Tawil syndrome (ORPHA:37553)
KCNJ5	Long QT syndrome 13 (OMIM:613485); Hyperaldosteronism, familial, type III (OMIM:613677); Romano-Ward syndrome (ORPHA:101016); Hereditary atrial fibrillation (ORPHA:334); Andersen-Tawil syndrome (ORPHA:37553); Familial hyperaldosteronism type III (ORPHA:251274)
KCNJ6	Keppen-Lubinsky syndrome (OMIM:614098); Keppen-Lubinsky syndrome (ORPHA:435628)
KCNJ8	Cantú syndrome (ORPHA:1517); Brugada syndrome (ORPHA:130)
KCNK18	Migraine, with or without aura, susceptibility to, 13 (OMIM:613656)
KCNK3	Developmental delay with sleep apnea (OMIM:621402); Pulmonary hypertension, primary, 4 (OMIM:615344); ORPHA:275777
KCNK9	Birk-Barel syndrome (OMIM:612292); Birk-Barel syndrome (ORPHA:166108)
KCNMA1	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy (OMIM:609446); Cerebellar atrophy, developmental delay, and seizures (OMIM:617643); Liang-Wang syndrome (OMIM:618729); Epilepsy, idiopathic generalized, susceptibility to, 16 (OMIM:618596); ORPHA:664438; Generalized epilepsy-paroxysmal dyskinesia syndrome (ORPHA:79137)
KCNMB1	HYPERTENSION, DIASTOLIC, RESISTANCE TO (OMIM:608622)
KCNMB4	—
KCNN2	Dystonia 34, myoclonic (OMIM:619724); Neurodevelopmental disorder with or without variable movement or behavioral abnormalities (OMIM:619725)
KCNN3	Zimmermann-Laband syndrome 3 (OMIM:618658); Zimmermann-Laband syndrome (ORPHA:3473)
KCNQ1	Jervell and Lange-Nielsen syndrome 1 (OMIM:220400); Beckwith-Wiedemann syndrome (OMIM:130650); Short QT syndrome 2 (OMIM:609621); Atrial fibrillation, familial, 3 (OMIM:607554); Long QT syndrome 1 (OMIM:192500); Congenital short QT syndrome (ORPHA:51083); Romano-Ward syndrome (ORPHA:101016); Hereditary atrial fibrillation (ORPHA:334); Jervell and Lange-Nielsen syndrome (ORPHA:90647)
KCNQ1OT1	Beckwith-Wiedemann syndrome (OMIM:130650); ORPHA:231117; Isolated hemihyperplasia (ORPHA:2128)
KCNQ2	Seizures, benign familial neonatal, 1 (OMIM:121200); Epileptic encephalopathy, early infantile, 7 (OMIM:613720); Self-limited infantile epilepsy (ORPHA:306); ORPHA:178469; Self-limited neonatal epilepsy (ORPHA:1949); KCNQ2-related developmental and epileptic encephalopathy (ORPHA:439218); Epilepsy of infancy with migrating focal seizures (ORPHA:293181); Self-limited neonatal-infantile epilepsy (ORPHA:140927)
KCNQ3	Epilepsy, benign neonatal, 2 (OMIM:121201); Juvenile myoclonic epilepsy (ORPHA:307); Self-limited infantile epilepsy (ORPHA:306); Self-limited neonatal epilepsy (ORPHA:1949)
KCNQ4	Deafness, autosomal dominant nonsyndromic sensorineural 2 (OMIM:600101); ORPHA:90635
KCNQ5	Intellectual developmental disorder, autosomal dominant 46 (OMIM:617601); ORPHA:178469
KCNRG	—
KCNS1	—
KCNS3	—
KCNT1	Epilepsy, nocturnal frontal lobe, 5 (OMIM:615005); Developmental and epileptic encephalopathy 14 (OMIM:614959); Sleep-related hypermotor epilepsy (ORPHA:98784); Epilepsy of infancy with migrating focal seizures (ORPHA:293181)
KCNV1	—
KCNV2	Retinal cone dystrophy 3B (OMIM:610356); ORPHA:209932
KCTD13	—
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions (OMIM:611726); Progressive myoclonic epilepsy type 3 (ORPHA:263516); ORPHA:699708

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KDM3A	—
KDM5A	El Hayek-Chahrour neurodevelopmental syndrome (OMIM:620820)
KDM5C	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type (OMIM:300534); KDM5C-related syndromic X-linked intellectual disability (ORPHA:85279)
KDM6A	Kabuki Syndrome 1 (OMIM:147920); Kabuki syndrome 2 (OMIM:300867); Kabuki syndrome (ORPHA:2322)
KDM6B	Stolerman neurodevelopmental syndrome (OMIM:618505)
KDR	Hemangioma, capillary infantile (OMIM:602089); Tetralogy of Fallot (ORPHA:3303)
KEL	OMIM:110900
KERA	Cornea plana 2 (OMIM:217300); ORPHA:53691
KHDC3L	Hydatidiform mole, recurrent, 2 (OMIM:614293); ORPHA:254693; ORPHA:254688
KHK	Fructosuria, essential (OMIM:229800); Essential fructosuria (ORPHA:2056)
KIAA0232	—
KIAA0319	—
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development (OMIM:152950); Microcephaly-lymphedema-chorioretinopathy syndrome (ORPHA:2526)
KIF17	—
KIF1A	Spastic paraplegia 30, autosomal recessive (OMIM:610357); Neuropathy, hereditary sensory and autonomic, type II (OMIM:201300); Neuropathy, hereditary sensory, type IIC (OMIM:614213); NESCAV syndrome (OMIM:614255); Spastic paraplegia 30, autosomal recessive (OMIM:620607); Autosomal spastic paraplegia type 30 (ORPHA:101010); ORPHA:662367; Hereditary sensory and autonomic neuropathy type 2 (ORPHA:970)
KIF1B	PHEOCHROMOCYTOMA (OMIM:171300); Neuroblastoma, susceptibility to (OMIM:256700); Charcot-Marie-Tooth disease, axonal, type 2A1 (OMIM:118210); ORPHA:99946; Hereditary pheochromocytoma-paraganglioma (ORPHA:29072)
KIF21A	Fibrosis of extraocular muscles, congenital, 1 (OMIM:135700); Fetal akinesia deformation sequence (ORPHA:994); Congenital fibrosis of extraocular muscles (ORPHA:45358)
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2 (OMIM:603546); Spondyloepimetaphyseal dysplasia with joint laxity, leptodactylic type (ORPHA:93360)
KIF27	—
KIF3C	—
KIF5A	Amyotrophic lateral sclerosis, susceptibility to, 25 (OMIM:617921); Spastic paraplegia 10, autosomal dominant (OMIM:604187); Myoclonus, intractable, neonatal (OMIM:617235); ORPHA:324611; Autosomal dominant spastic paraplegia type 10 (ORPHA:100991)
KIF7	Al-Gazali-Bakalinova syndrome (OMIM:607131); Hydrolethalus syndrome 2 (OMIM:614120); Acrocallosal syndrome (OMIM:200990); Acrocallosal syndrome (ORPHA:36); Orofaciodigital syndrome type 6 (ORPHA:2754); Multiple epiphyseal dysplasia-macrocephaly-facial dysmorphism syndrome (ORPHA:166024); Hydrolethalus (ORPHA:2189)
KIFBP	Goldberg-Shprintzen syndrome (OMIM:609460); Goldberg-Shprintzen megacolon syndrome (ORPHA:66629)
KIR2DL3	—
KIRREL3	ORPHA:178469
KISS1	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Hypogonadotropic hypogonadism 13 with or without anosmia (OMIM:614842); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432); ORPHA:650097
KISS1R	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Precocious puberty, central (OMIM:176400); Hypogonadotropic hypogonadism 8 with or without anosmia (OMIM:614837); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432); ORPHA:650097; ORPHA:650077
KIT	Testicular tumor, somatic (OMIM:273300); Mastocytosis, cutaneous (OMIM:154800); Piebald trait (OMIM:172800); Gastrointestinal stromal tumor (OMIM:606764); Leukemia, acute myeloid (OMIM:601626); Cutaneous mastocytoma (ORPHA:79455); ORPHA:158769; ORPHA:158766; Gastrointestinal stromal tumor (ORPHA:44890); ORPHA:158778; ORPHA:158775; ORPHA:158772; Piebaldism (ORPHA:2884); Systemic mastocytosis with associated hematologic neoplasm (ORPHA:98849); ORPHA:98834; ORPHA:98829; ORPHA:842; ORPHA:102724; Bullous diffuse cutaneous mastocytosis (ORPHA:280785); Pseudoxanthomatous diffuse cutaneous mastocytosis (ORPHA:280794); ORPHA:90389; ORPHA:566396; ORPHA:566393
KITLG	OMIM:611664; Hyperpigmentation, familial progressive (OMIM:145250); Deafness, autosomal dominant 69 (OMIM:616697); Waardenburg syndrome, type 2F (OMIM:619947); ORPHA:90635; Waardenburg syndrome type 2 (ORPHA:895); ORPHA:363494; ORPHA:280628; ORPHA:79146
KL	Tumoral calcinosis, hyperphosphatemic, familial, 3 (OMIM:617994); ORPHA:306661
KLC1	—
KLC4	Neurodegeneration, early-childhood-onset, with retinitis pigmentosa, sensorineural hearing loss, and demyelinating peripheral neuropathy (OMIM:621129)
KLF1	Anemia, congenital dyserythropoietic, type IVb (OMIM:620969); BLOOD GROUP--LUTHERAN INHIBITOR; INLU (OMIM:111150); Anemia, dyserythropoietic congenital, type IV (OMIM:613673); Hereditary persistence of fetal hemoglobin (OMIM:613566); Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome (ORPHA:46532); Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome (ORPHA:251380); ORPHA:293825
KLF10	—
KLF11	Maturity-onset diabetes of the young, type VII (OMIM:610508); MODY (ORPHA:552)
KLF2	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
KLF6	Prostate cancer (OMIM:176807); Gastric cancer, somatic (OMIM:613659)
KLF8	—
KLHDC4	—
KLHDC8B	Lymphoma, hodgkin (OMIM:236000); ORPHA:98843
KLHL10	Spermatogenic failure 11 (OMIM:615081); Male infertility with azoospermia or oligozoospermia due to single gene mutation (ORPHA:399805)
KLHL3	Pseudohypoaldosteronism, type IID (OMIM:614495); ORPHA:300525
KLHL7	Retinitis pigmentosa 42 (OMIM:612943); PERCHING syndrome (OMIM:617055); Retinitis pigmentosa (ORPHA:791); ORPHA:603689; ORPHA:603684; ORPHA:157820; ORPHA:603694
KLHL9	KLHL9-related early-onset distal myopathy (ORPHA:399081)
KLK1	OMIM:615953
KLK12	—
KLK4	Amelogenesis imperfecta, hypomaturation type, iia1 (OMIM:204700); ORPHA:100033
KLKB1	Prekallikrein deficiency (OMIM:612423); ORPHA:749
KMT2D	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome (OMIM:620186); Kabuki Syndrome 1 (OMIM:147920); Kabuki syndrome (ORPHA:2322); ORPHA:589856
KMT5A	—
KMT5B	Intellectual developmental disorder, autosomal dominant 51 (OMIM:617788); ORPHA:684226
KNG1	Angioedema, hereditary, 6 (OMIM:619363); Flaujeac factor deficiency (OMIM:228960); ORPHA:599418; ORPHA:483
KNL1	Microcephaly 4, primary, autosomal recessive (OMIM:604321); Autosomal recessive primary microcephaly (ORPHA:2512)
KPNA1	—
KRAS	Cardiofaciocutaneous syndrome 2 (OMIM:615278); Cardiofaciocutaneous syndrome 1 (OMIM:115150); OMIM:621582; Arteriovenous malformation of the brain, somatic (OMIM:108010); Pancreatic cancer (OMIM:260350); Schimmelpenning-Feuerstein-Mims syndrome (OMIM:163200); Breast cancer (OMIM:114480); Bladder cancer (OMIM:109800); Noonan syndrome 3 (OMIM:609942); Lung cancer, susceptibility to (OMIM:211980); Gastric cancer, somatic (OMIM:613659); Oculoectodermal syndrome (OMIM:600268); Leukemia, acute myeloid (OMIM:601626); Oculoectodermal syndrome (ORPHA:3339); Lynch syndrome (ORPHA:144); ORPHA:146; Linear nevus sebaceus syndrome (ORPHA:2612); ORPHA:86834; Cardiofaciocutaneous syndrome (ORPHA:1340); Familial pancreatic carcinoma (ORPHA:1333); Encephalocraniocutaneous lipomatosis (ORPHA:2396); ORPHA:268114; Noonan syndrome (ORPHA:648); ORPHA:251615
KRT1	Epidermolytic hyperkeratosis (OMIM:113800); Palmoplantar keratoderma, nonepidermolytic (OMIM:600962); Ichthyosis, annular epidermolytic 2 (OMIM:620148); Ichthyosis hystrix, Curth-Macklin type (OMIM:146590); Keratosis palmoplantaris striata III (OMIM:607654); Palmoplantar keratoderma, epidermolytic, 2 (OMIM:620411); Autosomal dominant epidermolytic ichthyosis (ORPHA:312); ORPHA:538574; Ichthyosis hystrix of Curth-Macklin (ORPHA:79503); KRT1-related diffuse nonepidermolytic keratoderma (ORPHA:530838); Striate palmoplantar keratoderma (ORPHA:50942); ORPHA:281139; ORPHA:281190; Epidermolytic palmoplantar keratoderma (ORPHA:2199)
KRT10	Erythroderma, ichthyosiform, congenital reticular (OMIM:609165); Epidermolytic hyperkeratosis 2 (OMIM:620150); Ichthyosis hystrix gravior (OMIM:146600); Ichthyosis, cyclic, with epidermolytic hyperkeratosis (OMIM:607602); Epidermolytic hyperkeratosis 2B, autosomal recessive (OMIM:620707); Autosomal dominant epidermolytic ichthyosis (ORPHA:312); ORPHA:512103; ORPHA:281139; ORPHA:281190
KRT12	Meesmann corneal dystrophy 1 (OMIM:122100); ORPHA:98954
KRT13	White sponge nevus 2 (OMIM:615785); ORPHA:171723
KRT14	Epidermolysis bullosa simplex, Weber-Cockayne type (OMIM:131800); Epidermolysis bullosa simplex, Dowling-Meara type (OMIM:131760); Epidermolysis bullosa simplex 1B, generalized intermediate (OMIM:131900); Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive (OMIM:601001); Naegeli-Franceschetti-Jadassohn syndrome (OMIM:161000); Dermatopathia pigmentosa reticularis (OMIM:125595); Localized epidermolysis bullosa simplex (ORPHA:79400); Autosomal dominant generalized epidermolysis bullosa simplex, severe form (ORPHA:79396); Epidermolysis bullosa simplex with mottled pigmentation (ORPHA:79397); Autosomal dominant generalized epidermolysis bullosa simplex, intermediate form (ORPHA:79399); Autosomal recessive generalized epidermolysis bullosa simplex (ORPHA:89838); ORPHA:86920; Naegeli-Franceschetti-Jadassohn syndrome (ORPHA:69087)
KRT16	Palmoplantar keratoderma, nonepidermolytic, focal 1 (OMIM:613000); Pachyonychia congenita, type 1 (OMIM:167200); Pachyonychia congenita (ORPHA:2309); ORPHA:448264; Epidermolytic palmoplantar keratoderma (ORPHA:2199)
KRT17	Steatocystoma multiplex (OMIM:184500); Pachyonychia congenita 2 (OMIM:167210); Sebocystomatosis (ORPHA:841); Pachyonychia congenita (ORPHA:2309)
KRT18	Cirrhosis, familial (OMIM:215600)
KRT2	Ichthyosis, Bullous type (OMIM:146800); Superficial epidermolytic ichthyosis (ORPHA:455)
KRT3	Meesmann corneal dystrophy 2 (OMIM:618767); ORPHA:98954
KRT37	—
KRT38	—
KRT4	White sponge nevus 1 (OMIM:193900); ORPHA:171723
KRT5	Epidermolysis bullosa simplex 2E, with migratory circinate erythema (OMIM:609352); Epidermolysis bullosa simplex, Dowling-Meara type (OMIM:131760); Dowling-Degos disease (OMIM:179850); Epidermolysis bullosa simplex with mottled pigmentation (OMIM:131960); Epidermolysis bullosa simplex 2A, generalized severe

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	(OMIM:619555); Epidermolysis bullosa simplex 2B, generalized intermediate (OMIM:619588); Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive (OMIM:619599); Epidermolysis bullosa simplex 2C, localized (OMIM:619594); Localized epidermolysis bullosa simplex (ORPHA:79400); Autosomal dominant generalized epidermolysis bullosa simplex, severe form (ORPHA:79396); Epidermolysis bullosa simplex with mottled pigmentation (ORPHA:79397); Autosomal dominant generalized epidermolysis bullosa simplex, intermediate form (ORPHA:79399); Epidermolysis bullosa simplex with circinate migratory erythema (ORPHA:158681); Dowling-Degos disease (ORPHA:79145)
KRT6A	Pachyonychia congenita 3 (OMIM:615726); Pachyonychia congenita, type 1 (OMIM:167200); Pachyonychia congenita (ORPHA:2309)
KRT74	Hypotrichosis 3 (OMIM:613981); Woolly hair, autosomal dominant (OMIM:194300); Ectodermal dysplasia 7, Hair/nail type (OMIM:614929); Woolly hair (ORPHA:170); Hypotrichosis simplex of the scalp (ORPHA:90368); ORPHA:69084
KRT75	OMIM:612318
KRT8	—
KRT81	Monilethrix 2 (OMIM:621169); Monilethrix (ORPHA:573)
KRT83	Erythrokeratodermia variabilis et progressiva 5 (OMIM:617756); Monilethrix 3 (OMIM:621170); Progressive symmetric erythrokeratodermia (ORPHA:316); Monilethrix (ORPHA:573)
KRT85	Ectodermal dysplasia 4, hair/nail type (OMIM:602032); ORPHA:69084
KRT86	Monilethrix (OMIM:158000); Monilethrix (ORPHA:573)
KRT9	Palmoplantar keratoderma, epidermolytic (OMIM:144200); Epidermolytic palmoplantar keratoderma (ORPHA:2199)
KRTAP1	—
KRTCAP3	—
KYNU	Vertebral, cardiac, renal, and limb defects syndrome 2 (OMIM:617661); Hydroxykynureninuria (OMIM:236800); ORPHA:521438; Hydroxykynureninuria (ORPHA:79155)
L1CAM	Hydrocephalus due to congenital stenosis of aqueduct of sylvius (OMIM:307000); MASA syndrome (OMIM:303350); Corpus callosum, partial agenesis of, X-linked (OMIM:304100); X-linked complicated corpus callosum dysgenesis (ORPHA:1497); X-linked complicated spastic paraplegia type 1 (ORPHA:306617); MASA syndrome (ORPHA:2466); Hydrocephalus with stenosis of the aqueduct of Sylvius (ORPHA:2182)
L2HGDH	L-2-hydroxyglutaric aciduria (OMIM:236792); L-2-hydroxyglutaric aciduria (ORPHA:79314)
L3MBTL1	—
LAMA1	Poretti-Boltshauser syndrome (OMIM:615960); Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome (ORPHA:370022)
LAMA2	Muscular dystrophy, limb-girdle, autosomal recessive 23 (OMIM:618138); Muscular dystrophy, congenital, merosin deficient or partially deficient (OMIM:607855); Laminin subunit alpha 2-related congenital muscular dystrophy (ORPHA:258); ORPHA:565837
LAMA3	Laryngoonychocutaneous syndrome (OMIM:245660); Epidermolysis bullosa, junctional, Herlitz type (OMIM:226700); Epidermolysis bullosa, junctional 2B, severe (OMIM:619784); Epidermolysis bullosa, junctional 2A, intermediate (OMIM:619783); Severe generalized junctional epidermolysis bullosa (ORPHA:79404); Intermediate generalized junctional epidermolysis bullosa (ORPHA:79402); Laryngo-onycho-cutaneous syndrome (ORPHA:2407)
LAMA4	Cardiomyopathy, dilated, 1JJ (OMIM:615235); Familial isolated dilated cardiomyopathy (ORPHA:154)
LAMA5	Nephrotic syndrome, type 26 (OMIM:620049); Bent bone dysplasia syndrome 2 (OMIM:620076); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656); ORPHA:521450
LAMB1	Lissencephaly 5 (OMIM:615191); Leukoencephalopathy without lacunae, adult-onset (OMIM:621424); Cobblestone lissencephaly without muscular or ocular involvement (ORPHA:352682)
LAMB2	Pierson syndrome (OMIM:609049); Nephrotic syndrome, type 5, with or without ocular abnormalities (OMIM:614199); Pierson syndrome (ORPHA:2670); Synaptic congenital myasthenic syndrome (ORPHA:98915)
LAMB3	Epidermolysis bullosa, junctional, Non-Herlitz type (OMIM:226650); Epidermolysis bullosa, junctional, Herlitz type (OMIM:226700); Amelogenesis imperfecta, type IA (OMIM:104530); Severe generalized junctional epidermolysis bullosa (ORPHA:79404); Intermediate generalized junctional epidermolysis bullosa (ORPHA:79402); ORPHA:100031
LAMB4	—
LAMC2	Epidermolysis bullosa, junctional, Herlitz type (OMIM:226700); Epidermolysis bullosa, junctional 3A, intermediate (OMIM:619785); Epidermolysis bullosa, junctional 3B, severe (OMIM:619786); Severe generalized junctional epidermolysis bullosa (ORPHA:79404); Intermediate generalized junctional epidermolysis bullosa (ORPHA:79402)
LAMC3	Cortical malformations, occipital (OMIM:614115); ORPHA:280640
LAMP2	Danon disease (OMIM:300257); Danon disease (ORPHA:34587)
LAMTOR1	—
LAMTOR2	Immunodeficiency due to defect in mapbp-interacting protein (OMIM:610798); Primary immunodeficiency syndrome due to P14/LAMTOR2 deficiency (ORPHA:90023)
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1 (OMIM:236670); Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6 (OMIM:608840); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6 (OMIM:613154); Congenital muscular dystrophy with intellectual disability (ORPHA:370968); Walker-Warburg syndrome (ORPHA:899); Muscle-eye-brain disease (ORPHA:588)
LARGE2	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
LAS1L	Wilson-Turner syndrome (OMIM:309585); ORPHA:404521; Wilson-Turner syndrome (ORPHA:3459)
LBCS	—
LBR	Greenberg skeletal dysplasia (OMIM:215140); Reynolds syndrome (OMIM:613471); Pelger-Huet anomaly (OMIM:169400); Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly (OMIM:618019); Greenberg dysplasia (ORPHA:1426); Reynolds syndrome (ORPHA:779); ORPHA:448267
LCA5	Leber congenital amaurosis 5 (OMIM:604537); Leber congenital amaurosis (ORPHA:65); Severe early-childhood-onset retinal dystrophy (ORPHA:364055)
LCAT	Lecithin:cholesterol acyltransferase deficiency (OMIM:245900); Fish-Eye disease (OMIM:136120); Fish-eye disease (ORPHA:79292); ORPHA:79293
LCE3B	—
LCE5A	—
LCK	Immunodeficiency 22 (OMIM:615758); ORPHA:280142
LCN10	—
LCT	Lactase deficiency, congenital (OMIM:223000); ORPHA:53690
LDB1	—
LDB3	Myopathy, myofibrillar, 4 (OMIM:609452); Cardiomyopathy, dilated, 2L (OMIM:621237); Cardiomyopathy, dilated, 1C, with or without left ventricular noncompaction (OMIM:601493); ORPHA:54260; Familial isolated dilated cardiomyopathy (ORPHA:154); ORPHA:293888; ORPHA:293899; ORPHA:293910; Late-onset distal myopathy, Markesbery-Griggs type (ORPHA:98912)
LDHA	Glycogen storage disease XI (OMIM:612933); Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency (ORPHA:284426)
LDHB	Lactate dehydrogenase B deficiency (OMIM:614128); ORPHA:284435
LDLR	Hypercholesterolemia, familial, 1 (OMIM:143890); Homozygous familial hypercholesterolemia (ORPHA:391665)
LDLRAD2	—
LDLRAP1	Hypercholesterolemia, autosomal recessive (OMIM:603813); Homozygous familial hypercholesterolemia (ORPHA:391665)
LECT2	—
LEF1	Ectodermal dysplasia 17 with or without limb malformations (OMIM:621224); ORPHA:708043
LEFTY2	ORPHA:157769
LEMD3	Buschke-Ollendorff syndrome (OMIM:166700); Isolated osteopoikilosis (ORPHA:166119); Melorheostosis with osteopoikilosis (ORPHA:1879); 12q14 microdeletion syndrome (ORPHA:94063)
LENG1	—
LEP	Leptin deficiency or dysfunction (OMIM:614962); Obesity due to congenital leptin deficiency (ORPHA:66628)
LEPR	Leptin receptor deficiency (OMIM:614963); Obesity due to leptin receptor gene deficiency (ORPHA:179494)
LETM1	Wolf-Hirschhorn syndrome (OMIM:194190); Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction (OMIM:620089); Wolf-Hirschhorn syndrome (ORPHA:280)
LFNG	Spondylocostal dysostosis 3, autosomal recessive (OMIM:609813); Autosomal recessive spondylocostal dysostosis (ORPHA:2311)
LGALS13	—
LGALS2	OMIM:608446
LGI1	Developmental and epileptic encephalopathy 121 (OMIM:621475); Epilepsy, familial temporal lobe, 1 (OMIM:600512); Epilepsy with auditory features (ORPHA:101046)
LGI4	Arthrogryposis multiplex congenita, neurogenic, with myelin defect (OMIM:617468); Hypomyelination neuropathy-arthrogryposis syndrome (ORPHA:2680)
LGR5	—
LHB	Hypogonadotropic hypogonadism 23 without anosmia (OMIM:228300); ORPHA:325448
LHFPL5	Deafness, autosomal recessive 67 (OMIM:610265); ORPHA:90636
LHX1	17q12 microdeletion syndrome (ORPHA:261265)
LHX3	Deafness, sensorineural, with pituitary dwarfism (OMIM:221750); Non-acquired combined pituitary hormone deficiency-sensorineural hearing loss-spine abnormalities syndrome (ORPHA:231720); Hypothyroidism due to deficient transcription factors involved in pituitary development or function (ORPHA:226307)
LHX4	Pituitary hormone deficiency, combined, 4 (OMIM:262700); Pituitary stalk interruption syndrome (ORPHA:95496); Combined pituitary hormone deficiencies, genetic forms (ORPHA:95494); Hypothyroidism due to deficient transcription factors involved in pituitary development or function (ORPHA:226307); ORPHA:85442
LHX8	—
LIAS	Hyperglycinemia, lactic acidosis, and seizures (OMIM:614462); ORPHA:401859
LIF	—
LIFR	Stuve-Wiedemann syndrome (OMIM:601559); Stüve-Wiedemann syndrome (ORPHA:3206)
LIG1	Immunodeficiency 96 (OMIM:619774)
LIG3	Mitochondrial DNA depletion syndrome 20 (MNGIE type) (OMIM:619780); Mitochondrial neurogastrointestinal encephalomyopathy (ORPHA:298)
LIG4	Multiple myeloma (OMIM:254500); LIG4 syndrome (OMIM:606593); LIG4 syndrome (ORPHA:99812); Dubowitz syndrome (ORPHA:235); Omenn syndrome (ORPHA:39041)
LIM2	Cataract 19, multiple types (OMIM:615277); ORPHA:98994
LIMASI	—
LIMK1	Williams syndrome (ORPHA:904)
LINS1	Intellectual developmental disorder, autosomal recessive 27 (OMIM:614340); ORPHA:88616

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
LIPA	Lysosomal acid lipase deficiency (OMIM:278000); Wolman disease (OMIM:620151); Wolman disease (ORPHA:75233); Cholesteryl ester storage disease (ORPHA:75234)
LIPC	Diabetes mellitus, noninsulin-dependent (OMIM:125853); OMIM:612797; Hepatic lipase deficiency (OMIM:614025); Hyperlipidemia due to hepatic triacylglycerol lipase deficiency (ORPHA:140905)
LIPG	—
LIPH	Hypotrichosis 7 (OMIM:604379); Woolly hair (ORPHA:170); Hypotrichosis simplex (ORPHA:55654)
LIPI	—
LIPN	Ichthyosis, congenital, autosomal recessive 8 (OMIM:613943); Lamellar ichthyosis (ORPHA:313)
LITAF	Charcot-Marie-Tooth disease, demyelinating, type 1C (OMIM:601098); ORPHA:101083
LIX1	—
LLGL1	—
LMAN1	Combined deficiency of factor V and factor VIII type 1 (OMIM:227300); Combined deficiency of factor V and factor VIII (ORPHA:35909)
LMBR1	Laurin-Sandrow syndrome (OMIM:135750); Triphalangeal thumb-polysyndactyly syndrome (OMIM:190605); Acheiropody (OMIM:200500); Syndactyly, type IV (OMIM:186200); Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome (ORPHA:988); Isolated acheiropodia (ORPHA:931); Laurin-Sandrow syndrome (ORPHA:2378); ORPHA:93336; Isolated radial hemimelia (ORPHA:93321); Syndactyly type 4 (ORPHA:93405)
LMBRD1	Methylmalonic aciduria and homocystinuria, Cblf type (OMIM:277380); Methylmalonic acidemia with homocystinuria type cblF (ORPHA:79284)
LMF1	Lipase deficiency, combined (OMIM:246650); ORPHA:535453
LMNA	Emery-Dreifuss muscular dystrophy 3, autosomal recessive (OMIM:616516); Heart-hand syndrome, Slovenian type (OMIM:610140); Malouf syndrome (OMIM:212112); Charcot-Marie-Tooth disease, axonal, type 2B1 (OMIM:605588); Mandibuloacral dysplasia (OMIM:248370); Cardiomyopathy, dilated, 1A (OMIM:115200); Hutchinson-Gilford progeria syndrome (OMIM:176670); LMNA-related congenital muscular dystrophy (OMIM:613205); Lipodystrophy, familial partial, type 2 (OMIM:151660); Emery-Dreifuss muscular dystrophy 2, autosomal dominant (OMIM:181350); Restrictive dermopathy 2 (OMIM:619793); ORPHA:54260; Atypical Werner syndrome (ORPHA:79474); Familial isolated dilated cardiomyopathy (ORPHA:154); Autosomal semi-dominant severe lipodystrophic laminopathy (ORPHA:280365); Heart-hand syndrome, Slovenian type (ORPHA:168796); Autosomal recessive Emery-Dreifuss muscular dystrophy (ORPHA:98855); Charcot-Marie-Tooth disease type 2B1 (ORPHA:98856); Autosomal dominant Emery-Dreifuss muscular dystrophy (ORPHA:98853); ORPHA:293888; ORPHA:293899; Congenital muscular dystrophy due to LMNA mutation (ORPHA:157973); Hutchinson-Gilford progeria syndrome (ORPHA:740); Familial partial lipodystrophy, Dunnigan type (ORPHA:2348); Mandibuloacral dysplasia with type A lipodystrophy (ORPHA:90153); ORPHA:293910; Familial dilated cardiomyopathy with conduction defect due to LMNA mutation (ORPHA:300751); Familial partial lipodystrophy, Köbberling type (ORPHA:79084); Restrictive dermopathy (ORPHA:1662); LMNA-related cardiocutaneous progeria syndrome (ORPHA:363618); ORPHA:675396; Dilated cardiomyopathy-hypergonadotropic hypogonadism syndrome (ORPHA:2229)
LMNB1	Microcephaly 26, primary, autosomal dominant (OMIM:619179); Adult-onset autosomal dominant leukodystrophy (ORPHA:99027); Autosomal dominant primary microcephaly (ORPHA:2514)
LMNB2	Epilepsy, progressive myoclonic, 9 (OMIM:616540); Microcephaly 27, primary, autosomal dominant (OMIM:619180); Lipodystrophy, partial, acquired, susceptibility to (OMIM:608709); Acquired partial lipodystrophy (ORPHA:79087); ORPHA:457265
LMO2	—
LMO4	—
LMTK3	—
LMX1B	Focal segmental glomerulosclerosis 10 (OMIM:256020); Nail-Patella syndrome (OMIM:161200); 9q33.3q34.11 microdeletion syndrome (ORPHA:495818); Nail-patella-like renal disease (ORPHA:2613); Nail-patella syndrome (ORPHA:2614)
LNC	—
LNCAROD	—
LNCTSI	—
LORICRIN	Vohwinkel syndrome, variant form (OMIM:604117); Progressive symmetric erythrokeratodermia (ORPHA:316); Keratoderma hereditarium mutilans with ichthyosis (ORPHA:79395)
LOX	Aortic aneurysm, familial thoracic 10 (OMIM:617168); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387)
LOXHD1	Deafness, autosomal recessive 77 (OMIM:613079); ORPHA:90636
LOXL1	Exfoliation syndrome (OMIM:177650)
LOXL3	Myopia 28, autosomal recessive (OMIM:619781); Autosomal recessive Stickler syndrome (ORPHA:250984)
LPA	OMIM:618807
LPAR6	Hypotrichosis 8 (OMIM:278150); Woolly hair (ORPHA:170); Hypotrichosis simplex (ORPHA:55654)
LPIN1	Myoglobinuria, acute recurrent, autosomal recessive (OMIM:268200); Genetic recurrent myoglobinuria (ORPHA:99845)
LPIN2	Majeed syndrome (OMIM:609628); Majeed syndrome (ORPHA:77297)
LPIN3	—
LPL	Type I hyperlipoproteinemia (OMIM:238600); Hyperlipidemia, familial combined, 3 (OMIM:144250); ORPHA:309015
LPO	—
LPP	Leukemia, acute myeloid (OMIM:601626)
LRAT	Leber congenital amaurosis 14 (OMIM:613341); Retinitis pigmentosa (OMIM:268000); Leber congenital

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	amaurosis (ORPHA:65); Retinitis pigmentosa (ORPHA:791); Severe early-childhood-onset retinal dystrophy (ORPHA:364055)
LRBA	Immunodeficiency, common variable, 8, with autoimmunity (OMIM:614700); ORPHA:445018
LRFN5	—
LRIT3	Night blindness, congenital stationary, type 1F (OMIM:615058); ORPHA:714079
LRP1	Developmental dysplasia of the hip 3 (OMIM:620690); Keratosis pilaris atrophicans (OMIM:604093); Keratosis follicularis spinulosa decalvans (ORPHA:2340); Atrophoderma vermiculata (ORPHA:79100)
LRP1B	—
LRP2	Donnai-Barrow syndrome (OMIM:222448); Donnai-Barrow syndrome (ORPHA:2143)
LRP4	Myasthenic syndrome, congenital, 17 (OMIM:616304); Sclerosteosis 2 (OMIM:614305); Cenani-Lenz syndactyly syndrome (OMIM:212780); Sclerosteosis (ORPHA:3152); Cenani-Lenz syndrome (ORPHA:3258); Postsynaptic congenital myasthenic syndrome (ORPHA:98913)
LRP5	Polycystic liver disease 4 with or without kidney cysts (OMIM:617875); Osteopetrosis, autosomal dominant 1 (OMIM:607634); Exudative vitreoretinopathy 1 (OMIM:133780); Osteoporosis-pseudoglioma syndrome (OMIM:259770); BONE MINERAL DENSITY QUANTITATIVE TRAIT LOCUS 1 (OMIM:601884); Exudative vitreoretinopathy 4 (OMIM:601813); Hyperostosis, endosteal (OMIM:144750); Endosteal hyperostosis, Worth type (ORPHA:2790); Osteoporosis-pseudoglioma syndrome (ORPHA:2788); ORPHA:2783; Osteosclerosis-developmental delay-craniosynostosis syndrome (ORPHA:178377); Familial exudative vitreoretinopathy (ORPHA:891); Retinopathy of prematurity (ORPHA:90050); Isolated polycystic liver disease (ORPHA:2924); ORPHA:498481; Hyperostosis corticalis generalisata (ORPHA:3416)
LRP6	Osteopetrosis, autosomal dominant 4 (OMIM:621449); Exudative vitreoretinopathy 8 (OMIM:621268); Tooth agenesis, selective, 7 (OMIM:616724); Coronary artery disease, autosomal dominant 2 (OMIM:610947); Autosomal dominant hypohidrotic ectodermal dysplasia (ORPHA:1810); Oligodontia (ORPHA:99798)
LRP8	OMIM:608446
LRPPRC	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian) (OMIM:220111); Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type (ORPHA:70472)
LRRC4	—
LRRC41	—
LRRC59	—
LRRC8A	Agammaglobulinemia 5, autosomal dominant (OMIM:613506); Autosomal non-syndromic agammaglobulinemia (ORPHA:33110)
LRRFIP2	—
LRRIQ1	—
LRRK2	Parkinson disease 8, autosomal dominant (OMIM:607060); Young-onset Parkinson disease (ORPHA:2828); Hereditary late-onset Parkinson disease (ORPHA:411602)
LRRTM1	—
LRSAM1	Charcot-Marie-Tooth disease, axonal, type 2P (OMIM:614436); ORPHA:300319
LRTOMT	Deafness, autosomal recessive 63 (OMIM:611451); ORPHA:90636
LTBP1	Cutis laxa, autosomal recessive, type IIE (OMIM:619451); Autosomal recessive cutis laxa type 1 (ORPHA:90349)
LTBP2	Glaucoma 3, primary infantile, B (OMIM:600975); Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma (OMIM:251750); Glaucoma 3, primary congenital, D (OMIM:613086); Weill-Marchesani syndrome 3 (OMIM:614819); Congenital glaucoma (ORPHA:98976); ORPHA:238763; Weill-Marchesani syndrome (ORPHA:3449)
LTBP3	Geleophysic dysplasia 3 (OMIM:617809); Dental anomalies and short stature (OMIM:601216); ORPHA:2899; Acromicric dysplasia (ORPHA:969); Geleophysic dysplasia (ORPHA:2623)
LTBP4	Cutis laxa, autosomal recessive, type IC (OMIM:613177); Duchenne muscular dystrophy (ORPHA:98896); ORPHA:221145
LTBR	—
LTC4S	Leukotriene C4 synthase deficiency (OMIM:614037)
LTN1	—
LUCAT1	—
LUM	—
LURAP1L	—
LXN	—
LY96	—
LYN	Autoinflammatory disease, systemic, with vasculitis (OMIM:620376)
LYST	Chediak-Higashi syndrome (OMIM:214500); Chédiak-Higashi syndrome (ORPHA:167); Attenuated Chédiak-Higashi syndrome (ORPHA:352723)
LYZ	Amyloidosis, hereditary systemic 5 (OMIM:620658); ORPHA:93561
LZTFL1	Bardet-Biedl syndrome 17 (OMIM:615994); Bardet-Biedl syndrome (ORPHA:110)
LZTS1	Esophageal cancer, somatic (OMIM:133239)
MACF1	Lissencephaly 9 with complex brainstem malformation (OMIM:618325); Posterior-predominant lissencephaly-broad flat pons and medulla-midline crossing defects syndrome (ORPHA:572013)
MACROD2	—
MACROH2A1	Brachydactyly-elbow wrist dysplasia syndrome (ORPHA:1275)
MAD1L1	Mosaic variegated aneuploidy syndrome 7 with inflammation and tumor predisposition (OMIM:620189); Prostate cancer (OMIM:176807)
MADD	Deeah syndrome (OMIM:619004); Neurodevelopmental disorder with dysmorphic facies, impaired speech and

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	hypotonia (OMIM:619005); ORPHA:686495; ORPHA:528084
MAEA	—
MAF	Cataract 21, multiple types (OMIM:610202); Ayme-Gripp syndrome (OMIM:601088); Aymé-Gripp syndrome (ORPHA:1272); Cataract-microcornea syndrome (ORPHA:1377); ORPHA:98989; ORPHA:98984
MAFA	Insulinomatosis and diabetes mellitus (OMIM:147630)
MAFB	Multicentric carpotarsal osteolysis syndrome (OMIM:166300); Duane retraction syndrome 3 with or without deafness (OMIM:617041); ORPHA:529574; Multicentric carpo-tarsal osteolysis with or without nephropathy (ORPHA:2774); Duane retraction syndrome (ORPHA:233)
MAGEC1	—
MAGEE2	—
MAGEL2	Prader-Willi syndrome (OMIM:176270); Schaaf-Yang syndrome (OMIM:615547); Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2 (ORPHA:177904); Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1 (ORPHA:177901); Prader-Willi syndrome due to imprinting mutation (ORPHA:177910); Fetal akinesia deformation sequence (ORPHA:994); Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15 (ORPHA:98754); Schaaf-Yang syndrome (ORPHA:398069)
MAGI2	Nephrotic syndrome, type 15 (OMIM:617609); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
MAGT1	Congenital disorder of glycosylation, type Icc (OMIM:301031); Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia (OMIM:300853); ORPHA:317476
MAK16	—
MALT1	Immunodeficiency 12 (OMIM:615468); MALT lymphoma (ORPHA:52417); ORPHA:397964
MAML2	—
MAMLD1	Hypospadias 2, X-linked (OMIM:300758); X-linked myotubular myopathy-abnormal genitalia syndrome (ORPHA:456328); Non-syndromic posterior hypospadias (ORPHA:95706)
MAN1A2	—
MAN1B1	Rafiq syndrome (OMIM:614202); ORPHA:88616; MAN1B1-CDG (ORPHA:397941)
MAN2A1	—
MAN2B1	Alpha-mannosidosis (OMIM:248500); Alpha-mannosidosis, infantile form (ORPHA:309282); Alpha-mannosidosis, adult form (ORPHA:309288)
MANBA	Mannosidosis, beta (OMIM:248510); Beta-mannosidosis (ORPHA:118)
MAOA	Brunner syndrome (OMIM:300615); Monoamine oxidase A deficiency (ORPHA:3057)
MAOB	—
MAP2	—
MAP2K1	Cardiofaciocutaneous syndrome 3 (OMIM:615279); Cardiofaciocutaneous syndrome 1 (OMIM:115150); Melorheostosis, isolated (OMIM:155950); Noonan syndrome 1 (OMIM:163950); Langerhans cell histiocytosis (ORPHA:389); Cardiofaciocutaneous syndrome (ORPHA:1340)
MAP2K2	Cardiofaciocutaneous syndrome 4 (OMIM:615280); Cardiofaciocutaneous syndrome 1 (OMIM:115150); Cardiofaciocutaneous syndrome (ORPHA:1340); Neurofibromatosis-Noonan syndrome (ORPHA:638)
MAP2K3	—
MAP2K4	—
MAP2K7	—
MAP3K1	46,xy sex reversal 6 (OMIM:613762); 46,XY partial gonadal dysgenesis (ORPHA:251510); 46,XY complete gonadal dysgenesis (ORPHA:242)
MAP3K14	Immunodeficiency 112 (OMIM:620449); ORPHA:447731
MAP3K15	—
MAP3K8	Lung cancer, susceptibility to (OMIM:211980)
MAP4K5	—
MAP6	—
MAP7D1	—
MAP9	—
MAPK1	Noonan syndrome 13 (OMIM:619087); Distal 22q11.2 microdeletion syndrome (ORPHA:261330)
MAPK10	Lennox-Gastaut syndrome (ORPHA:2382)
MAPK8IP1	Diabetes mellitus, noninsulin-dependent (OMIM:125853)
MAPKAPK3	Macular dystrophy, patterned, 3 (OMIM:617111); ORPHA:466718
MAPT	Pick disease of brain (OMIM:172700); Supranuclear palsy, progressive atypical (OMIM:260540); Supranuclear palsy, progressive, 1 (OMIM:601104); Parkinson disease, late-onset (OMIM:168600); Frontotemporal dementia (OMIM:600274); Progressive supranuclear palsy-pure akinesia with gait freezing syndrome (ORPHA:240094); Progressive supranuclear palsy-predominant parkinsonism syndrome (ORPHA:240085); Classic progressive supranuclear palsy syndrome (ORPHA:240071); Behavioral variant of frontotemporal dementia (ORPHA:275864); Progressive non-fluent aphasia (ORPHA:100070); Semantic dementia (ORPHA:100069); Progressive supranuclear palsy-progressive non-fluent aphasia syndrome (ORPHA:240112); Progressive supranuclear palsy-corticobasal syndrome (ORPHA:240103)
MARS1	Charcot-Marie-Tooth disease, axonal, type 2U (OMIM:616280); Interstitial lung and liver disease (OMIM:615486); Trichothiodystrophy 9, nonphotosensitive (OMIM:619692); Spastic paraplegia 70, autosomal recessive (OMIM:620323); Autosomal recessive spastic paraplegia type 70 (ORPHA:401835); ORPHA:397735; ORPHA:440427
MARS2	Combined oxidative phosphorylation deficiency 25 (OMIM:616430); Spastic ataxia 3, autosomal recessive (OMIM:611390); ORPHA:447954; Autosomal recessive spastic ataxia with leukoencephalopathy (ORPHA:314603)
MARVELD2	Deafness, autosomal recessive 49 (OMIM:610153); ORPHA:90636

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
MASP1	3MC syndrome 1 (OMIM:257920); 3MC syndrome (ORPHA:293843)
MASP2	Masp2 deficiency (OMIM:613791); ORPHA:331187
MAST3	Developmental and epileptic encephalopathy 108 (OMIM:620115)
MASTL	ORPHA:168629
MAT1A	Methionine adenosyltransferase I/III deficiency (OMIM:250850); ORPHA:168598
MATN3	Epiphyseal dysplasia, multiple, 5 (OMIM:607078); Osteoarthritis of distal interphalangeal joints (OMIM:140600); Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type (OMIM:608728); Spondyloepimetaphyseal dysplasia, matrilin-3 type (ORPHA:156728); Multiple epiphyseal dysplasia type 5 (ORPHA:93311)
MATR3	Amyotrophic lateral sclerosis 21 (OMIM:606070); Amyotrophic lateral sclerosis (ORPHA:803); Vocal cord and pharyngeal distal myopathy (ORPHA:600)
MAX	PHEOCHROMOCYTOMA (OMIM:171300); Polydactyly-macrocephaly syndrome (OMIM:620712); Hereditary pheochromocytoma-paraganglioma (ORPHA:29072)
MBD1	—
MBD3	—
MBD4	Tumor predisposition syndrome 2 (OMIM:619975); Uveal melanoma, susceptibility to, 1 (OMIM:606660); ORPHA:661526
MBD5	Intellectual developmental disorder, autosomal dominant 1 (OMIM:156200); ORPHA:178469; 2q23.1 microdeletion syndrome (ORPHA:228402)
MBL2	Mannose-Binding lectin deficiency (OMIM:614372)
MBNL1	—
MBOAT7	Intellectual developmental disorder, autosomal recessive 57 (OMIM:617188); ORPHA:88616
MBTPS2	IFAP syndrome with or without BRESHECK syndrome (OMIM:308205); Palmoplantar keratoderma, mutilating, with periorificial keratotic plaques, X-linked (OMIM:300918); Keratosis follicularis spinulosa decalvans, X-linked (OMIM:308800); Osteogenesis imperfecta, type XIX (OMIM:301014); BRESEK syndrome (ORPHA:85284); ORPHA:216812; ORPHA:216820; ORPHA:216796; Keratosis follicularis spinulosa decalvans (ORPHA:2340); Mutilating palmoplantar keratoderma with periorificial keratotic plaques (ORPHA:659); Ichthyosis follicularis-alopecia-photophobia syndrome (ORPHA:2273)
MC1R	OMIM:613099; OMIM:613098; SKIN/HAIR/EYE PIGMENTATION, VARIATION IN, 2 (OMIM:266300); Albinism, oculocutaneous, type II (OMIM:203200); Oculocutaneous albinism type 2 (ORPHA:79432); Familial melanoma (ORPHA:618)
MC2R	Glucocorticoid deficiency 1 (OMIM:202200); Familial glucocorticoid deficiency (ORPHA:361)
MC3R	BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 9; BMIQ9 (OMIM:602025)
MC4R	Body mass index quantitative trait locus 20 (OMIM:618406); Obesity due to melanocortin 4 receptor deficiency (ORPHA:71529)
MCC	Colorectal cancer (OMIM:114500)
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency (OMIM:210200); 3-methylcrotonyl-CoA carboxylase deficiency (ORPHA:6)
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency (OMIM:210210); 3-methylcrotonyl-CoA carboxylase deficiency (ORPHA:6)
MCEE	Methylmalonyl-Coa epimerase deficiency (OMIM:251120); ORPHA:308425
MCF2L2	—
MCFD2	Combined deficiency of factor V and factor VIII type 2 (OMIM:613625); Combined deficiency of factor V and factor VIII (ORPHA:35909)
MCHR1	—
MCM4	Immunodeficiency 54 (OMIM:609981); ORPHA:75391
MCM5	Meier-Gorlin syndrome 8 (OMIM:617564)
MCM6	Lactase persistence/nonpersistence (OMIM:223100)
MCM7	Autosomal recessive primary microcephaly (ORPHA:2512)
MCM9	Ovarian dysgenesis 4 (OMIM:616185); ORPHA:444048
MCOLN1	Lisch epithelial corneal dystrophy (OMIM:620763); Mucolipidosis IV (OMIM:252650); Mucolipidosis type IV (ORPHA:578)
MCPH1	Microcephaly, primary autosomal recessive, 1 (OMIM:251200); Autosomal recessive primary microcephaly (ORPHA:2512)
MCRS1	—
MDGA2	Developmental and epileptic encephalopathy 122 (OMIM:621608)
MDK	—
MDM2	OMIM:614401; Lessel-Kubisch syndrome (OMIM:618681); ORPHA:99970; Well-differentiated liposarcoma (ORPHA:99971); Li-Fraumeni syndrome (ORPHA:524)
MDN1	—
ME2	—
MECOM	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2 (OMIM:616738); ORPHA:402020; Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome (ORPHA:71289)
MECP2	Intellectual developmental disorder, X-linked syndromic 13 (OMIM:300055); Autism susceptibility, X-linked 3 (OMIM:300496); Intellectual developmental disorder, X-linked syndromic, Lubs type (OMIM:300260); Rett syndrome (OMIM:312750); Encephalopathy, neonatal severe, due to mecp2 mutations (OMIM:300673); Atypical Rett syndrome (ORPHA:3095); X-linked intellectual disability-psychosis-macroorchidism syndrome (ORPHA:3077); Rett syndrome (ORPHA:778); ORPHA:777; Systemic lupus erythematosus (ORPHA:536); MECP2-related severe neonatal encephalopathy (ORPHA:209370); Proximal Xq28 duplication syndrome

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	(ORPHA:1762)
MED12	Lujan-Fryns syndrome (OMIM:309520); Hardikar syndrome (OMIM:301068); Ohdo syndrome, X-linked (OMIM:300895); Opitz-Kaveggia syndrome (OMIM:305450); Hardikar syndrome (ORPHA:1415); FG syndrome type 1 (ORPHA:93932); Blepharophimosis-intellectual disability syndrome, MKB type (ORPHA:293707); Lujan-Fryns syndrome (ORPHA:776); ORPHA:777
MED12L	Nizon-Isidor syndrome (OMIM:618872); ORPHA:528084
MED13L	Impaired intellectual development and distinctive facial features with or without cardiac defects (OMIM:616789); ORPHA:216718; Developmental delay-facial dysmorphism syndrome due to MED13L deficiency (ORPHA:369891)
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy (OMIM:613668); ORPHA:402364
MED23	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy (OMIM:614249); ORPHA:88616
MED25	Basel-Vanagaite-Smirin-Yosef syndrome (OMIM:616449); Charcot-Marie-Tooth disease, axonal, type 2B2 (OMIM:605589); Basel-Vanagaite-Smirin-Yosef syndrome (ORPHA:464738); ORPHA:88616
MEF2A	Coronary artery disease, autosomal dominant, 1 (OMIM:608320)
MEF2B	—
MEF2C	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language (OMIM:613443); 5q14.3 microdeletion syndrome (ORPHA:228384); ORPHA:576227; ORPHA:664416
MEF2D	—
MEFV	Neutrophilic dermatosis, acute febrile (OMIM:608068); Familial Mediterranean fever, AD (OMIM:134610); Familial Mediterranean fever, AR (OMIM:249100); Familial Mediterranean fever (ORPHA:342); Sweet syndrome (ORPHA:3243); Intermittent hydrarthrosis (ORPHA:329967); Behçet disease (ORPHA:117)
MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset (OMIM:614399); Congenital myopathy 10B, mild variant (OMIM:620249); ORPHA:439212
MEGF11	—
MEGF8	Carpenter syndrome 2 (OMIM:614976); Carpenter syndrome (ORPHA:65759)
MEIS1	—
MEN1	Hyperparathyroidism 1 (OMIM:145000); Multiple endocrine neoplasia 1 (OMIM:131100); Familial isolated hyperparathyroidism (ORPHA:99879); Insulinoma (ORPHA:97279); Multiple endocrine neoplasia type 1 (ORPHA:652); Prolactinoma (ORPHA:2965); ORPHA:314790; ORPHA:314786; Pituitary gigantism (ORPHA:99725)
MEPE	—
MERTK	Retinitis pigmentosa 38 (OMIM:613862); Retinitis pigmentosa (ORPHA:791)
MESD	Osteogenesis imperfecta, type XX (OMIM:618644); ORPHA:216804
MESP2	Spondylocostal dysostosis, autosomal recessive 1 (OMIM:277300); Spondylocostal dysostosis 2, autosomal recessive (OMIM:608681); Autosomal recessive spondylocostal dysostosis (ORPHA:2311)
MEST	—
MET	Osteofibrous dysplasia, susceptibility to (OMIM:607278); Arthrogryposis, distal, type 11 (OMIM:620019); Deafness, autosomal recessive 97 (OMIM:616705); Renal cell carcinoma, papillary, 1, familial and somatic (OMIM:605074); Hepatocellular carcinoma (OMIM:114550); ORPHA:90636; ORPHA:47044; Pediatric hepatocellular carcinoma (ORPHA:33402); ORPHA:319298; ORPHA:488265
METTL21A	—
METTL8	—
MFAP4	—
MFF	Encephalopathy due to defective mitochondrial and peroxisomal fission 2 (OMIM:617086); MFF-related encephalopathy due to mitochondrial and peroxisomal fission defect (ORPHA:485421)
MFGE8	—
MFN2	Charcot-marie-tooth disease, axonal, type 2A2A (OMIM:609260); Charcot-Marie-Tooth disease, axonal, type 2A2B (OMIM:617087); Hereditary motor and sensory neuropathy VIA (OMIM:601152); Lipomatosis, multiple symmetric, with or without peripheral neuropathy (OMIM:151800); Autosomal dominant Charcot-Marie-Tooth disease type 2A2 (ORPHA:99947); ORPHA:90120; ORPHA:90118; Multiple symmetric lipomatosis (ORPHA:2398); ORPHA:64751
MFRP	Nanophthalmos 2 (OMIM:609549); Microphthalmia, isolated 5 (OMIM:611040); ORPHA:251279; Nanophthalmos (ORPHA:35612)
MFSD8	Ceroid lipofuscinosis, neuronal, 7 (OMIM:610951); Macular dystrophy with central cone involvement (OMIM:616170); ORPHA:228366; Cone rod dystrophy (ORPHA:1872)
MGAM	—
MGAT1	—
MGAT2	Congenital disorder of glycosylation, type IIa (OMIM:212066); MGAT2-CDG (ORPHA:79329)
MGAT3	—
MGAT4A	—
MGAT4B	—
MGAT4C	—
MGAT5	—
MGAT5B	—
MGC16275	—
MGC32805	—
MGLL	—
MGMT	ORPHA:251579; ORPHA:251576; Familial melanoma (ORPHA:618)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
MGP	Keutel syndrome (OMIM:245150); Keutel syndrome (ORPHA:85202)
MGST2	—
MGST3	—
MICA	—
MICAL1	Epilepsy, familial temporal lobe, 1 (OMIM:600512); Epilepsy with auditory features (ORPHA:101046)
MID1	Opitz GBBB syndrome (OMIM:300000); Opitz GBBB syndrome (ORPHA:2745)
MIF	OMIM:604302; Systemic-onset juvenile idiopathic arthritis (ORPHA:85414); Cystic fibrosis (ORPHA:586)
MIF4GD	—
MILR1	—
MINPP1	Thyroid cancer, nonmedullary, 2 (OMIM:188470); Pontocerebellar hypoplasia, type 16 (OMIM:619527); Familial papillary or follicular thyroid carcinoma (ORPHA:319487); Pontocerebellar hypoplasia type 7 (ORPHA:284339)
MIP	Cataract 15, multiple types (OMIM:615274); ORPHA:98994; ORPHA:98991; ORPHA:98993; ORPHA:98989; ORPHA:98985; ORPHA:441452
MIPOL1	—
MITF	Tietz albinism-deafness syndrome (OMIM:103500); Melanoma, cutaneous malignant, susceptibility to, 8 (OMIM:614456); Coloboma, osteopetrosis, microphthalmia, macrocephaly, albinism, and deafness (OMIM:617306); Waardenburg syndrome, type 2A (OMIM:193510); Waardenburg-Shah syndrome (ORPHA:897); Waardenburg syndrome type 2 (ORPHA:895); ORPHA:293822; ORPHA:319298; Tietz syndrome (ORPHA:42665); ORPHA:404511; Familial melanoma (ORPHA:618)
MKKS	Mckusick-Kaufman syndrome (OMIM:236700); Bardet-Biedl syndrome 6 (OMIM:605231); Bardet-Biedl syndrome (ORPHA:110); McKusick-Kaufman syndrome (ORPHA:2473)
MKRN2	—
MKS1	Bardet-Biedl syndrome 13 (OMIM:615990); Joubert syndrome 28 (OMIM:617121); Meckel syndrome 1 (OMIM:249000); Joubert syndrome with ocular defect (ORPHA:220493); Bardet-Biedl syndrome (ORPHA:110); Meckel syndrome (ORPHA:564); Isolated Joubert syndrome (ORPHA:475)
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts 1 (OMIM:604004); Megalencephalic leukoencephalopathy with subcortical cysts (ORPHA:2478)
MLH1	Colorectal cancer, hereditary nonpolyposis, type 2 (OMIM:609310); Muir-Torre syndrome (OMIM:158320); Mismatch repair cancer syndrome 1 (OMIM:276300); Lynch syndrome (ORPHA:144); ORPHA:252202
MLH3	Endometrial carcinoma, somatic (OMIM:608089); Colorectal cancer, hereditary nonpolyposis, type 7 (OMIM:614385); Colorectal cancer (OMIM:114500)
MLLT1	—
MLLT10	Leukemia, acute myeloid (OMIM:601626); ORPHA:99861
MLLT11	—
MLLT3	ORPHA:402017
MLPH	Griscelli syndrome, type 3 (OMIM:609227); Griscelli syndrome type 3 (ORPHA:79478)
MLX	Takayasu arteritis (ORPHA:3287)
MLXIP	—
MLXIPL	Williams-Beuren syndrome (OMIM:194050)
MLYCD	Malonyl-CoA decarboxylase deficiency (OMIM:248360); Malonic aciduria (ORPHA:943)
MMAA	Methylmalonic aciduria, vitamin B12-responsive, cblA type (OMIM:251100); ORPHA:79310
MMAB	Methylmalonic aciduria, vitamin B12-responsive, cblB type (OMIM:251110); ORPHA:79311
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type (OMIM:277400); Methylmalonic acidemia with homocystinuria, type cblC (ORPHA:79282)
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type (OMIM:277410); Methylmalonic aciduria, cblD type (OMIM:620953); Homocystinuria-megaloblastic anemia, cblD type (OMIM:620952); ORPHA:308380; ORPHA:308442; Methylmalonic acidemia with homocystinuria, type cblD (ORPHA:79283)
MME	Spinocerebellar ataxia 43 (OMIM:617018); Charcot-Marie-Tooth disease, axonal, type 2T (OMIM:617017); ORPHA:497757; Spinocerebellar ataxia type 43 (ORPHA:497764); ORPHA:495274; Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization (ORPHA:69063)
MMEL1	Primary biliary cholangitis (ORPHA:186)
MMP1	Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form (ORPHA:79408)
MMP13	Metaphyseal chondrodysplasia, Spahr type (OMIM:250400); Spondyloepimetaphyseal dysplasia, Missouri type (OMIM:602111); Metaphyseal chondrodysplasia, Spahr type (ORPHA:2501); Metaphyseal anadysplasia (ORPHA:1040); Spondyloepimetaphyseal dysplasia, Missouri type (ORPHA:93356)
MMP14	Winchester syndrome (OMIM:277950); Multicentric osteolysis-nodulosis-arthropathy spectrum (ORPHA:371428)
MMP2	Multicentric osteolysis, nodulosis, and arthropathy (OMIM:259600); Multicentric osteolysis-nodulosis-arthropathy spectrum (ORPHA:371428)
MMP20	Amelogenesis imperfecta, hypomaturation type, iia2 (OMIM:612529); ORPHA:100033
MMP26	—
MMP3	OMIM:614466
MMP8	—
MMP9	Metaphyseal anadysplasia 2 (OMIM:613073); Metaphyseal anadysplasia (ORPHA:1040)
MMUT	Methylmalonic aciduria, mut type (OMIM:251000); Vitamin B12-unresponsive methylmalonic acidemia type mut0 (ORPHA:289916); Vitamin B12-unresponsive methylmalonic acidemia type mut- (ORPHA:79312)
MN1	Meningioma, familial, susceptibility to (OMIM:607174); CEBALID syndrome (OMIM:618774); ORPHA:693549; ORPHA:263662

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
MNX1	Currarino syndrome (OMIM:176450); Currarino syndrome (ORPHA:1552)
MOB1B	—
MOCOS	Xanthinuria, type II (OMIM:603592); ORPHA:93602
MOCS1	Molybdenum cofactor deficiency, complementation group A (OMIM:252150); ORPHA:308386
MOCS2	Molybdenum cofactor deficiency, complementation group B (OMIM:252160); ORPHA:308393
MOG	Narcolepsy 7 (OMIM:614250); Narcolepsy type 1 (ORPHA:2073)
MOGS	Congenital disorder of glycosylation, type IIB (OMIM:606056); MOGS-CDG (ORPHA:79330)
MOK	—
MPC1	Mitochondrial pyruvate carrier deficiency (OMIM:614741); ORPHA:447784
MPDU1	Congenital disorder of glycosylation, type IF (OMIM:609180); MPDU1-CDG (ORPHA:79323)
MPDZ	Hydrocephalus, congenital, 2, with or without brain or eye anomalies (OMIM:615219); ORPHA:269505
MPG	—
MPHOSPH8	—
MPI	Congenital disorder of glycosylation, type Ib (OMIM:602579); MPI-CDG (ORPHA:79319)
MPL	Amegakaryocytic thrombocytopenia, congenital (OMIM:604498); Myelofibrosis with myeloid metaplasia, somatic (OMIM:254450); Thrombocythemia 2 (OMIM:601977); Familial thrombocytosis (ORPHA:71493); Essential thrombocythemia (ORPHA:3318); Congenital amegakaryocytic thrombocytopenia (ORPHA:3319); Primary myelofibrosis (ORPHA:824); ORPHA:397692
MPLKIP	Trichothiodystrophy 4, nonphotosensitive (OMIM:234050); Trichothiodystrophy (ORPHA:33364)
MPO	Alzheimer disease (OMIM:104300); Myeloperoxidase deficiency (OMIM:254600); ORPHA:2587
MPRIP	—
MPST	—
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) (OMIM:256810); Charcot-Marie-Tooth disease, axonal, type 2EE (OMIM:618400); ORPHA:255229
MPZ	Roussy-Levy hereditary areflexic dystasia (OMIM:180800); Charcot-Marie-Tooth disease, axonal, type 2I (OMIM:607677); Neuropathy, congenital hypomyelinating, 2 (OMIM:618184); Charcot-Marie-Tooth disease, type 1B (OMIM:118200); Hypertrophic neuropathy of dejerine-sottas (OMIM:145900); Charcot-Marie-Tooth disease, dominant intermediate D (OMIM:607791); Charcot-Marie-Tooth disease, type 2J (OMIM:607736); Charcot-Marie-Tooth disease type 1B (ORPHA:101082); ORPHA:538574; ORPHA:99942; ORPHA:99943; ORPHA:324585; Roussy-Lévy syndrome (ORPHA:3115); ORPHA:64748; ORPHA:100046
MR1	—
MRAP	Glucocorticoid deficiency 2 (OMIM:607398); Familial glucocorticoid deficiency (ORPHA:361)
MRC1	—
MRE11	Ataxia-telangiectasia-like disorder 1 (OMIM:604391); ORPHA:240760; Hereditary breast and/or ovarian cancer syndrome (ORPHA:145); Ataxia-telangiectasia-like disorder (ORPHA:251347)
MREG	—
MRNIP	—
MROH8	—
MRPL3	Combined oxidative phosphorylation deficiency 9 (OMIM:614582); ORPHA:319509
MRPL43	—
MRPL48	—
MRPS16	Combined oxidative phosphorylation deficiency 2 (OMIM:610498); ORPHA:254920
MRPS18C	—
MRPS22	Combined oxidative phosphorylation deficiency 5 (OMIM:611719); Ovarian dysgenesis 7 (OMIM:618117); ORPHA:137908; 46,XX gonadal dysgenesis (ORPHA:243)
MRRF	—
MRTFA	Immunodeficiency 66 (OMIM:618847); ORPHA:619941; ORPHA:402023
MS4A1	Immunodeficiency, common variable, 5 (OMIM:613495)
MS4A12	—
MS4A2	—
MSC	—
MSH2	Muir-Torre syndrome (OMIM:158320); Mismatch repair cancer syndrome 2 (OMIM:619096); Lynch syndrome I (OMIM:120435); Lynch syndrome (ORPHA:144); ORPHA:252202
MSH3	Endometrial carcinoma, somatic (OMIM:608089); Familial adenomatous polyposis 4 (OMIM:617100); MSH3-related polyposis (ORPHA:480536)
MSH6	Endometrial carcinoma, somatic (OMIM:608089); Colorectal cancer, hereditary nonpolyposis, type 5 (OMIM:614350); Mismatch repair cancer syndrome 3 (OMIM:619097); Lynch syndrome (ORPHA:144); ORPHA:252202
MSMB	ORPHA:1331
MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis (OMIM:616834); ORPHA:488168
MSR1	Barrett esophagus (OMIM:614266); ORPHA:1331
MSRA	—
MSRB3	Deafness, autosomal recessive 74 (OMIM:613718); ORPHA:90636
MST1	Primary sclerosing cholangitis (ORPHA:171)
MST1R	Nasopharyngeal carcinoma, susceptibility to, 3 (OMIM:617075)
MSTN	Muscle hypertrophy (OMIM:614160)
MSX1	Witkop syndrome (OMIM:189500); Orofacial cleft 5 (OMIM:608874); Tooth agenesis, selective, 1 (OMIM:106600); Isolated cleft lip (ORPHA:199302); Cleft lip/palate (ORPHA:199306); Cleft lip and alveolus (ORPHA:141291); Oligodontia (ORPHA:99798); Hypodontia-dysplasia of nails syndrome (ORPHA:2228)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
MSX2	Craniosynostosis 2 (OMIM:604757); Parietal foramina with cleidocranial dysplasia (OMIM:168550); Parietal foramina (OMIM:168500); Enlarged parietal foramina (ORPHA:60015); ORPHA:1541; ORPHA:251290
MTA1	—
MTA2	—
MTAP	Diaphyseal medullary stenosis with malignant fibrous histiocytoma (OMIM:112250); ORPHA:85182
MTBP	—
MTCL3	—
MTCP1	—
MTFMT	Mitochondrial complex I deficiency, nuclear type 27 (OMIM:618248); Combined oxidative phosphorylation deficiency 15 (OMIM:614947); ORPHA:319524
MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia (OMIM:617780); Neural tube defects, folate-sensitive (OMIM:601634); ORPHA:658813
MTHFR	Homocystinuria due to deficiency of n(5,10)-methylenetetrahydrofolatereductase activity (OMIM:236250); Thrombophiliavenous thromboembolism, included (OMIM:188050); Schizophrenia (OMIM:181500); Neural tube defects, folate-sensitive (OMIM:601634); Homocystinuria due to methylene tetrahydrofolate reductase deficiency (ORPHA:395); Isolated exencephaly (ORPHA:563612); Isolated anencephaly (ORPHA:563609)
MTHFS	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination (OMIM:618367); ORPHA:597874
MTM1	Myopathy, centronuclear, X-linked (OMIM:310400); X-linked myotubular myopathy-abnormal genitalia syndrome (ORPHA:456328); X-linked centronuclear myopathy (ORPHA:596)
MTMR14	Centronuclear myopathy 1 (OMIM:160150); Autosomal dominant centronuclear myopathy (ORPHA:169189)
MTMR2	Charcot-Marie-Tooth disease, type 4B1 (OMIM:601382); ORPHA:99955
MTMR9	—
MTNR1A	—
MTNR1B	Diabetes mellitus, noninsulin-dependent (OMIM:125853)
MTO1	Combined oxidative phosphorylation deficiency 10 (OMIM:614702); ORPHA:314637
MTPAP	Spastic ataxia 4, autosomal recessive (OMIM:613672); Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome (ORPHA:254343)
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type (OMIM:250940); Neural tube defects, folate-sensitive (OMIM:601634); ORPHA:2170
MTRR	Homocystinuria-megaloblastic anemia, cbl E type (OMIM:236270); Neural tube defects, folate-sensitive (OMIM:601634); Methylcobalamin deficiency type cblE (ORPHA:2169)
MTSS1	—
MTTP	ABETALIPOPROTEINEMIA (OMIM:200100); Abetalipoproteinemia (ORPHA:14)
MUC12	—
MUC15	—
MUC16	—
MUC17	—
MUC5B	Pulmonary fibrosis, idiopathic (OMIM:178500); ORPHA:171700; Idiopathic pulmonary fibrosis (ORPHA:2032); ORPHA:686465
MUC7	Asthma, susceptibility to (OMIM:600807)
MUS81	—
MUSK	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency (OMIM:616325); Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency (OMIM:608931); Fetal akinesia deformation sequence (OMIM:208150); Fetal akinesia deformation sequence (ORPHA:994); Postsynaptic congenital myasthenic syndrome (ORPHA:98913)
MUTYH	Familial adenomatous polyposis, 2 (OMIM:608456); Gastric cancer, somatic (OMIM:613659); MUTYH-related polyposis (ORPHA:247798); Familial colorectal cancer Type X (ORPHA:440437)
MVK	Mevalonic aciduria (OMIM:610377); Hyper-Igd syndrome (OMIM:260920); Porokeratosis 3, multiple types (OMIM:175900); Hyperimmunoglobulinemia D with periodic fever (ORPHA:343); Mevalonic aciduria (ORPHA:29); Porokeratosis of Mibelli (ORPHA:735); Disseminated superficial actinic porokeratosis (ORPHA:79152)
MVP	—
MXI1	Prostate cancer (OMIM:176807)
MYB	ORPHA:99861; ORPHA:86849; ORPHA:251671
MYBPC1	Arthrogryposis, distal, type 1B (OMIM:614335); Myopathy, congenital, with tremor (OMIM:618524); Lethal congenital contracture syndrome 4 (OMIM:614915); ORPHA:498693; ORPHA:137783; Distal arthrogryposis type 1 (ORPHA:1146)
MYBPC3	Cardiomyopathy, familial hypertrophic, 4 (OMIM:115197); Left ventricular noncompaction 10 (OMIM:615396); ORPHA:54260; Familial isolated dilated cardiomyopathy (ORPHA:154)
MYC	Burkitt lymphoma (OMIM:113970); ORPHA:99861; ORPHA:480541; Burkitt lymphoma (ORPHA:543)
MYCN	Feingold syndrome 1 (OMIM:164280); Megalencephaly-polydactyly syndrome (OMIM:620748); Neuroblastoma (ORPHA:635); Feingold syndrome type 1 (ORPHA:391641); ORPHA:357034; ORPHA:357027
MYCNOS	—
MYD88	Macroglobulinemia, Waldenstrom, somatic (OMIM:153600); Immunodeficiency 68 (OMIM:612260); Waldenström macroglobulinemia (ORPHA:33226); ORPHA:714046; Transient predisposition to invasive pyogenic bacterial infection (ORPHA:70592)
MYEF2	—
MYF5	Ophthalmoplegia, external, with rib and vertebral anomalies (OMIM:618155)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
MYF6	Autosomal dominant centronuclear myopathy (ORPHA:169189)
MYH11	Visceral myopathy 2 (OMIM:619350); Megacystis-microcolon-intestinal hypoperistalsis syndrome 2 (OMIM:619351); Aortic aneurysm, familial thoracic 4 (OMIM:132900); Familial aortic dissection (ORPHA:229); ORPHA:98829; Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); Megacystis-microcolon-intestinal hypoperistalsis syndrome (ORPHA:2241)
MYH14	Peripheral neuropathy, myopathy, hoarseness, and hearing loss (OMIM:614369); Deafness, autosomal dominant nonsyndromic sensorineural 4 (OMIM:600652); ORPHA:90635; MYH14-related peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome (ORPHA:397744)
MYH2	Myopathy, proximal, and ophthalmoplegia (OMIM:605637); ORPHA:79091; ORPHA:363677
MYH3	Arthrogryposis, distal, type 2B3 (OMIM:618436); Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A (OMIM:178110); Contractures, pterygia, and variable skeletal fusions syndrome 1B (OMIM:618469); Arthrogryposis, distal, type 2A (OMIM:193700); ORPHA:65743; Spondylocarpotarsal synostosis (ORPHA:3275); Freeman-Sheldon syndrome (ORPHA:2053); Sheldon-Hall syndrome (ORPHA:1147); Distal arthrogryposis type 1 (ORPHA:1146); Autosomal recessive multiple pterygium syndrome (ORPHA:2990)
MYH6	Cardiomyopathy, familial hypertrophic 1 (OMIM:192600); Atrial septal defect 3 (OMIM:614089); Sick sinus syndrome 3 (OMIM:614090); Cardiomyopathy, familial hypertrophic, 14 (OMIM:613251); Cardiomyopathy, dilated, 1EE (OMIM:613252); Atrial septal defect, ostium secundum type (ORPHA:99103); Familial isolated dilated cardiomyopathy (ORPHA:154); ORPHA:166282
MYH7	Myopathy, myosin storage, autosomal recessive (OMIM:255160); Cardiomyopathy, familial hypertrophic 1 (OMIM:192600); Myopathy, distal, 1 (OMIM:160500); Myopathy, myosin storage, autosomal dominant (OMIM:608358); Cardiomyopathy, dilated, 1S (OMIM:613426); ORPHA:636970; ORPHA:636965; ORPHA:54260; Familial isolated dilated cardiomyopathy (ORPHA:154); Laing distal myopathy (ORPHA:59135); Ebstein malformation of the tricuspid valve (ORPHA:1880); Classic multiminicore myopathy (ORPHA:324604)
MYH8	Arthrogryposis, distal, type 7 (OMIM:158300); Carney complex variant (OMIM:608837); Trismus-pseudocamptodactyly syndrome (ORPHA:3377); ORPHA:319340
MYH9	Deafness, autosomal dominant nonsyndromic sensorineural 17 (OMIM:603622); Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss (OMIM:155100); ORPHA:90635; MYH9-related syndromic thrombocytopenia (ORPHA:182050); ORPHA:477742
MYHAS	—
MYL1	Myopathy, congenital, with fast-twitch (type II) fiber atrophy (OMIM:618414); ORPHA:544602
MYL2	Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy (OMIM:619424); Cardiomyopathy, familial hypertrophic, 10 (OMIM:608758); Congenital fiber-type disproportion myopathy (ORPHA:2020)
MYL3	Cardiomyopathy, familial hypertrophic, 8 (OMIM:608751)
MYLK	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1 (OMIM:249210); Aortic aneurysm, familial thoracic 7 (OMIM:613780); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); Megacystis-microcolon-intestinal hypoperistalsis syndrome (ORPHA:2241)
MYLK2	Cardiomyopathy, familial hypertrophic 1 (OMIM:192600)
MYO15A	Deafness, neurosensory, autosomal recessive 3 (OMIM:600316); ORPHA:90636
MYO1A	Diarrhea 15, congenital (OMIM:621179); Deafness, autosomal dominant 48 (OMIM:607841); ORPHA:90635
MYO1C	ORPHA:90635
MYO1D	—
MYO1E	Focal segmental glomerulosclerosis 6 (OMIM:614131); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
MYO1F	—
MYO3A	Deafness, autosomal recessive 30 (OMIM:607101); Deafness, autosomal dominant 90 (OMIM:620722); ORPHA:90636
MYO5A	Griscelli syndrome, type 1 (OMIM:214450); Griscelli syndrome type 1 (ORPHA:79476); Griscelli syndrome type 3 (ORPHA:79478); Neuroectodermal melanolysosomal disease (ORPHA:33445)
MYO5B	Diarrhea 2, with microvillous atrophy (OMIM:251850); Cholestasis, progressive familial intrahepatic, 10 (OMIM:619868); ORPHA:79306; ORPHA:480491; Microvillus inclusion disease (ORPHA:2290)
MYO5C	—
MYO6	Deafness, autosomal dominant 22 (OMIM:606346); Deafness, autosomal recessive 37 (OMIM:607821); ORPHA:90635; ORPHA:90636; ORPHA:228012
MYO7A	Usher syndrome, type I (OMIM:276900); Deafness, neurosensory, autosomal recessive 2 (OMIM:600060); Deafness, autosomal dominant nonsyndromic sensorineural 11 (OMIM:601317); ORPHA:90635; ORPHA:90636; Usher syndrome type 2 (ORPHA:231178); Usher syndrome type 1 (ORPHA:231169)
MYO7B	—
MYO9B	—
MYOC	Glaucoma 1, open angle, A (OMIM:137750); Congenital glaucoma (ORPHA:98976); Juvenile glaucoma (ORPHA:98977)
MYOCD	Megabladder, congenital (OMIM:618719)
MYOCOS	—
MYOD1	Myopathy, congenital, with diaphragmatic defects, respiratory insufficiency, and dysmorphic facies (OMIM:618975); Fetal akinesia deformation sequence (ORPHA:994)
MYOM1	—
MYOT	Myopathy, myofibrillar, 3 (OMIM:609200); Distal myotilinopathy (ORPHA:98911); ORPHA:268129
MYOZ2	Cardiomyopathy, familial hypertrophic, 16 (OMIM:613838)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
MYPN	Cardiomyopathy, dilated, 1KK (OMIM:615248); Nemaline myopathy 11, autosomal recessive (OMIM:617336); Familial isolated dilated cardiomyopathy (ORPHA:154); Familial isolated restrictive cardiomyopathy (ORPHA:75249); Childhood-onset nemaline myopathy (ORPHA:171439); Cap myopathy (ORPHA:171881)
MYT1	—
MYT1L	Intellectual developmental disorder, autosomal dominant 39 (OMIM:616521); ORPHA:699850; MYT1L-related developmental delay-intellectual disability-obesity syndrome (ORPHA:647799)
NAA10	Microphthalmia, syndromic 1 (OMIM:309800); Ogden syndrome (OMIM:300855); Ogden syndrome (ORPHA:276432); Microphthalmia, Lenz type (ORPHA:568)
NAALADL2	—
NAGA	Schindler disease, type I (OMIM:609241); Kanzaki disease (OMIM:609242); Alpha-N-acetylgalactosaminidase deficiency type 2 (ORPHA:79280); Alpha-N-acetylgalactosaminidase deficiency type 3 (ORPHA:79281); Alpha-N-acetylgalactosaminidase deficiency type 1 (ORPHA:79279)
NAGLU	Mucopolysaccharidosis type IIIB (OMIM:252920); Charcot-Marie-Tooth disease, axonal, type 2V (OMIM:616491); ORPHA:447964; ORPHA:79270
NAGPA	—
NAGS	N-acetylglutamate synthase deficiency (OMIM:237310); Hyperammonemia due to N-acetylglutamate synthase deficiency (ORPHA:927)
NAIP	ORPHA:83418; ORPHA:83419; ORPHA:83330
NAMA	—
NAPRT	—
NARS2	Combined oxidative phosphorylation deficiency 24 (OMIM:616239); Deafness, autosomal recessive 94 (OMIM:618434); ORPHA:90636; ORPHA:444458; DEND syndrome (ORPHA:79134)
NAT1	—
NAT2	Acetylation, slow (OMIM:243400)
NAT8L	—
NAV2	—
NBAS	Infantile liver failure syndrome 2 (OMIM:616483); Short stature, optic nerve atrophy, and pelger-huet anomaly (OMIM:614800); ORPHA:464724; ORPHA:391677
NBEA	Neurodevelopmental disorder with or without early-onset generalized epilepsy (OMIM:619157); ORPHA:178469
NBEAL2	Gray platelet syndrome (OMIM:139090); Gray platelet syndrome (ORPHA:721)
NBN	Aplastic anemia (OMIM:609135); Leukemia, acute lymphocytic, susceptibility to, 1 (OMIM:613065); Nijmegen breakage syndrome (OMIM:251260); Hereditary breast and/or ovarian cancer syndrome (ORPHA:145); ORPHA:1331; Nijmegen breakage syndrome (ORPHA:647)
NBPF1	—
NCALD	—
NCAPD2	Microcephaly 21, primary, autosomal recessive (OMIM:617983)
NCAPH2	—
NCF2	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type II (OMIM:233710); Chronic granulomatous disease (ORPHA:379)
NCF4	Chronic granulomatous disease 3, autosomal recessive (OMIM:613960); Chronic granulomatous disease (ORPHA:379)
NCK1	—
NCKAP1	—
NCOA1	—
NCOA2	—
NCOA4	ORPHA:146
NCOR1	—
NCR3	OMIM:609148
NCS1	—
NCSTN	Acne inversa, familial (OMIM:142690); ORPHA:289478
NDE1	Lissencephaly 4 (OMIM:614019); MICROHYDRANENCEPHALY (OMIM:605013); Lissencephaly syndrome, Norman-Roberts type (ORPHA:89844); ORPHA:443162; Hydranencephaly (ORPHA:2177)
NDN	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2 (ORPHA:177904); Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1 (ORPHA:177901); Prader-Willi syndrome due to imprinting mutation (ORPHA:177910); Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15 (ORPHA:98754)
NDOR1	—
NDP	Norrie disease (OMIM:310600); Exudative vitreoretinopathy 2, X-linked (OMIM:305390); Coats disease (ORPHA:190); Familial exudative vitreoretinopathy (ORPHA:891); Retinopathy of prematurity (ORPHA:90050); Persistent hyperplastic primary vitreous (ORPHA:91495); Norrie disease (ORPHA:649)
NDRG1	Charcot-Marie-Tooth disease, type 4D (OMIM:601455); Charcot-Marie-Tooth disease type 4D (ORPHA:99950)
NDST1	Intellectual developmental disorder, autosomal recessive 46 (OMIM:616116); ORPHA:88616
NDST2	—
NDST3	—
NDST4	—
NDUFA1	Mitochondrial complex I deficiency, nuclear type 12 (OMIM:301020); Isolated complex I deficiency (ORPHA:2609)
NDUFA10	Mitochondrial complex I deficiency, nuclear type 22 (OMIM:618243)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
NDUFA11	Mitochondrial complex I deficiency, nuclear type 14 (OMIM:618236); Isolated complex I deficiency (ORPHA:2609)
NDUFA12	Mitochondrial complex I deficiency, nuclear type 23 (OMIM:618244)
NDUFA13	Mitochondrial complex I deficiency, nuclear type 28 (OMIM:618249); OMIM:607464; ORPHA:146
NDUFA2	Mitochondrial complex I deficiency, nuclear type 13 (OMIM:618235); ORPHA:85136
NDUFA4	—
NDUFA6	Mitochondrial complex I deficiency, nuclear type 33 (OMIM:618253); Isolated complex I deficiency (ORPHA:2609)
NDUFA8	Mitochondrial complex I deficiency, nuclear type 37 (OMIM:619272)
NDUFA9	Mitochondrial complex I deficiency, nuclear type 26 (OMIM:618247)
NDUFAF1	Mitochondrial complex I deficiency, nuclear type 11 (OMIM:618234); Isolated complex I deficiency (ORPHA:2609)
NDUFAF3	Mitochondrial complex I deficiency, nuclear type 18 (OMIM:618240); Isolated complex I deficiency (ORPHA:2609)
NDUFAF4	Mitochondrial complex I deficiency, nuclear type 15 (OMIM:618237); Isolated complex I deficiency (ORPHA:2609)
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16 (OMIM:618238); Isolated complex I deficiency (ORPHA:2609)
NDUFAF6	Fanconi renotubular syndrome 5 (OMIM:618913); Mitochondrial complex I deficiency, nuclear type 17 (OMIM:618239); Primary Fanconi renotubular syndrome (ORPHA:3337)
NDUFAF7	—
NDUFB1	—
NDUFB11	Linear skin defects with multiple congenital anomalies 3 (OMIM:300952); Mitochondrial complex I deficiency, nuclear type 30 (OMIM:301021); Linear skin defects with multiple congenital anomalies 1 (OMIM:309801); Microphthalmia with linear skin defects syndrome (ORPHA:2556); Isolated complex I deficiency (ORPHA:2609)
NDUFB3	Mitochondrial complex I deficiency, nuclear type 25 (OMIM:618246); Isolated complex I deficiency (ORPHA:2609)
NDUFB9	Mitochondrial complex I deficiency, nuclear type 24 (OMIM:618245); Isolated complex I deficiency (ORPHA:2609)
NDUFS1	Mitochondrial complex I deficiency, nuclear type 5 (OMIM:618226); Isolated complex I deficiency (ORPHA:2609)
NDUFS2	Mitochondrial complex I deficiency, nuclear type 6 (OMIM:618228); Leber-like hereditary optic neuropathy, autosomal recessive 2 (OMIM:620569); Leber hereditary optic neuropathy (ORPHA:104); Isolated complex I deficiency (ORPHA:2609)
NDUFS3	Mitochondrial complex I deficiency, nuclear type 8 (OMIM:618230); Isolated complex I deficiency (ORPHA:2609)
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1 (OMIM:252010); Isolated complex I deficiency (ORPHA:2609)
NDUFS5	—
NDUFS6	Mitochondrial complex I deficiency, nuclear type 9 (OMIM:618232); Isolated complex I deficiency (ORPHA:2609)
NDUFS7	Mitochondrial complex I deficiency, nuclear type 3 (OMIM:618224); Isolated complex I deficiency (ORPHA:2609)
NDUFS8	Mitochondrial complex I deficiency, nuclear type 2 (OMIM:618222); Isolated complex I deficiency (ORPHA:2609)
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4 (OMIM:618225); Isolated complex I deficiency (ORPHA:2609)
NDUFV2	Mitochondrial complex I deficiency, nuclear type 7 (OMIM:618229); ORPHA:139447; Isolated complex I deficiency (ORPHA:2609)
NDUFV3	—
NEB	Arthrogryposis multiplex congenita 6 (OMIM:619334); Nemaline myopathy 2, autosomal recessive (OMIM:256030); Autosomal recessive distal nebulin myopathy (ORPHA:399103); ORPHA:708123; Typical nemaline myopathy (ORPHA:171436); Intermediate nemaline myopathy (ORPHA:171433); Severe congenital nemaline myopathy (ORPHA:171430); Childhood-onset nemaline myopathy (ORPHA:171439); ORPHA:33108
NEBL	—
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome (OMIM:225060); Isolated cleft lip (ORPHA:199302); Cleft lip/palate (ORPHA:199306); Cleft lip/palate-ectodermal dysplasia syndrome (ORPHA:3253); Cleft lip and alveolus (ORPHA:141291)
NECTIN4	Ectodermal dysplasia-syndactyly syndrome 1 (OMIM:613573); ORPHA:247820
NEDD4	—
NEDD4L	Periventricular nodular heterotopia 7 (OMIM:617201); Periventricular nodular heterotopia (ORPHA:98892)
NEFH	Amyotrophic lateral sclerosis 1 (OMIM:105400); Charcot-Marie-Tooth disease, axonal, type 2CC (OMIM:616924); Amyotrophic lateral sclerosis (ORPHA:803)
NEFL	Charcot-Marie-Tooth disease, dominant intermediate G (OMIM:617882); Charcot-Marie-Tooth disease, axonal, type 2E (OMIM:607684); Charcot-Marie-Tooth disease, demyelinating, type 1F (OMIM:607734); Charcot-Marie-Tooth disease type 1F (ORPHA:101085); ORPHA:228374; Autosomal dominant Charcot-Marie-Tooth disease type 2E (ORPHA:99939)
NEFM	—
NEGR1	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
NEIL1	—
NEIL2	—
NEK1	Amyotrophic lateral sclerosis, susceptibility to, 24 (OMIM:617892); Short-Rib thoracic dysplasia 6 with or without polydactyly (OMIM:263520); Mohr syndrome (OMIM:252100); Orofaciodigital syndrome type 2 (ORPHA:2751); ORPHA:93269; Amyotrophic lateral sclerosis (ORPHA:803)
NEK8	Renal-Hepatic-Pancreatic dysplasia 2 (OMIM:615415); Nephronophthisis 9 (OMIM:613824); Polycystic kidney disease 8 (OMIM:620903); Renal-hepatic-pancreatic dysplasia (ORPHA:294415); Autosomal dominant polycystic kidney disease (ORPHA:730); ORPHA:93591
NEU2	—
NEUROD1	Diabetes mellitus, noninsulin-dependent (OMIM:125853); Maturity-onset diabetes of the young 6 (OMIM:606394); MODY (ORPHA:552)
NEUROG3	Diarrhea 4, malabsorptive, congenital (OMIM:610370); Enteric anendocrinosis (ORPHA:83620)
NEXMIF	Intellectual developmental disorder, X-linked 98 (OMIM:300912); X-linked intellectual disability, Cantagrel type (ORPHA:85277); Epilepsy with myoclonic-atonic seizures (ORPHA:1942)
NEXN	Cardiomyopathy, dilated, 2M, autosomal recessive (OMIM:621261); Cardiomyopathy, dilated, 1cc (OMIM:613122); Cardiomyopathy, familial hypertrophic, 20 (OMIM:613876); Familial isolated dilated cardiomyopathy (ORPHA:154)
NF1	Neurofibromatosis-Noonan syndrome (OMIM:601321); Neurofibromatosis, familial spinal (OMIM:162210); Neurofibromatosis, type 1 (OMIM:162200); Watson syndrome (OMIM:193520); Juvenile myelomonocytic leukemia (OMIM:607785); 17q11.2 microduplication syndrome (ORPHA:139474); ORPHA:86834; 17q11 microdeletion syndrome (ORPHA:97685); Neurofibromatosis-Noonan syndrome (ORPHA:638); ORPHA:293199; Neurofibromatosis type 1 due to NF1 mutation or intragenic deletion (ORPHA:363700); Hereditary pheochromocytoma-paraganglioma (ORPHA:29072); ORPHA:99757; ORPHA:99756
NF2	Meningioma, familial, susceptibility to (OMIM:607174); Neurofibromatosis, type II (OMIM:101000); Full schwannomatosis (ORPHA:93921); ORPHA:634475; Full NF2-related schwannomatosis (ORPHA:637); Meningioma (ORPHA:2495)
NFATC2	Joint contracture, osteochondromas, and B-cell lymphoma (OMIM:620232)
NFATC3	—
NFATC4	—
NFE2L1	—
NFE2L2	Immunodeficiency, developmental delay, and hypohomocysteinemia (OMIM:617744); ORPHA:619979
NFIA	Brain malformations with or without urinary tract defects (OMIM:613735); 1p31p32 microdeletion syndrome (ORPHA:401986)
NFIB	Macrocephaly, acquired, with impaired intellectual development (OMIM:618286); ORPHA:714407; ORPHA:714413
NFIX	Malan syndrome (OMIM:614753); Marshall-Smith syndrome (OMIM:602535); 19p13.3 microduplication syndrome (ORPHA:447980); Malan overgrowth syndrome (ORPHA:420179); Marshall-Smith syndrome (ORPHA:561)
NFKB2	Immunodeficiency, common variable, 10 (OMIM:615577); Deficiency in anterior pituitary function-variable immunodeficiency syndrome (ORPHA:293978)
NFKBIA	Ectodermal dysplasia and immunodeficiency 2 (OMIM:612132); ORPHA:251579; ORPHA:251576; ORPHA:150; Hypohidrotic ectodermal dysplasia with immunodeficiency (ORPHA:98813)
NFKBIE	—
NFKBIZ	—
NFU1	Multiple mitochondrial dysfunctions syndrome 1 (OMIM:605711); Spastic paraplegia 93, autosomal recessive (OMIM:620938); ORPHA:401869
NGF	Neuropathy, hereditary sensory and autonomic, type V (OMIM:608654); Hereditary sensory and autonomic neuropathy type 5 (ORPHA:64752)
NGLY1	Congenital disorder of deglycosylation 1 (OMIM:615273); Alacrimia-choreoathetosis-liver dysfunction syndrome (ORPHA:404454)
NHEJ1	Immunodeficiency 124, severe combined (OMIM:611291); Microphthalmia/coloboma 13 (OMIM:620968); Cernunnos-XLF deficiency (ORPHA:169079)
NHERF1	Nephrolithiasis/osteoporosis, hypophosphatemic, 2 (OMIM:612287); ORPHA:244305
NHLRC1	Myoclonic epilepsy of Lafora 2 (OMIM:620681); Lafora disease (ORPHA:501)
NHS	Cataract, congenital total, with posterior sutural opacities in heterozygotes (OMIM:302200); Nance-Horan syndrome (OMIM:302350); ORPHA:98994; ORPHA:98991; Nance-Horan syndrome (ORPHA:627)
NICN1	—
NIN	Seckel syndrome 7 (OMIM:614851); Seckel syndrome (ORPHA:808); Microcephalic primordial dwarfism, Dauber type (ORPHA:319675)
NINJ1	—
NIP7	—
NIPA1	Spastic paraplegia 6, autosomal dominant (OMIM:600363); Autosomal dominant spastic paraplegia type 6 (ORPHA:100988); 15q11.2 microdeletion syndrome (ORPHA:261183)
NIPAL1	—
NIPAL4	Ichthyosis, congenital, autosomal recessive 6 (OMIM:612281); Lamellar ichthyosis (ORPHA:313); Congenital ichthyosiform erythroderma (ORPHA:79394)
NIPBL	Cornelia de Lange syndrome 1 (OMIM:122470); Cornelia de Lange syndrome (ORPHA:199); ORPHA:329802
NIPSNAP1	—
NIPSNAP3A	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
NKAIN2	—
NKAIN4	—
NKX2	—
NKX3	—
NLGN1	Autism, susceptibility to, 20 (OMIM:618830)
NLGN2	—
NLGN3	Autism susceptibility, X-linked 1 (OMIM:300425); Autism susceptibility 1 (OMIM:209850)
NLGN4X	Autism, susceptibility to, X-linked 2 (OMIM:300495); Autism susceptibility 1 (OMIM:209850)
NLGN4Y	—
NLRP1	Palmoplantar carcinoma, multiple self-healing (OMIM:615225); Respiratory papillomatosis, juvenile recurrent, congenital (OMIM:618803); Autoinflammation with arthritis and dyskeratosis (OMIM:617388); Vitiligo-Associated multiple autoimmune disease susceptibility 1 (OMIM:606579); ORPHA:352662
NLRP12	Familial cold autoinflammatory syndrome 2 (OMIM:611762); ORPHA:247868
NLRP14	—
NLRP2	Oocyte/zygote/embryo maturation arrest 18 (OMIM:620332)
NLRP3	Deafness, autosomal dominant 34, with or without inflammation (OMIM:617772); Muckle-Wells syndrome (OMIM:191900); Cinca syndrome (OMIM:607115); Familial cold inflammatory syndrome 1 (OMIM:120100); Keratoendotheliitis fugax hereditaria (OMIM:148200); CINCA syndrome (ORPHA:1451); Familial cold urticaria (ORPHA:47045); ORPHA:647815; Muckle-Wells syndrome (ORPHA:575)
NLRP7	Hydatidiform mole, recurrent, 1 (OMIM:231090); Ooxyte/zygote/embryo maturation arrest 25 (OMIM:621471); ORPHA:254693; ORPHA:254688
NLRX1	—
NME1	—
NME2	—
NME5	Ciliary dyskinesia, primary, 48, without situs inversus (OMIM:620032); Primary ciliary dyskinesia (ORPHA:244)
NME7	ORPHA:101063
NME8	Ciliary dyskinesia, primary, 6 (OMIM:610852); Primary ciliary dyskinesia (ORPHA:244)
NMNAT1	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis (OMIM:619260); Leber congenital amaurosis 9 (OMIM:608553); Leber congenital amaurosis (ORPHA:65); Cone rod dystrophy (ORPHA:1872)
NMT2	—
NMU	—
NNT	Glucocorticoid deficiency 4 with or without mineralocorticoid deficiency (OMIM:614736); Familial glucocorticoid deficiency (ORPHA:361)
NOBOX	Premature ovarian failure 5 (OMIM:611548)
NOD1	—
NOD2	Inflammatory bowel disease 1, Crohn disease (OMIM:266600); Blau syndrome (OMIM:186580); Yao syndrome (OMIM:617321); Blau syndrome (ORPHA:90340)
NODAL	Heterotaxy, visceral, 5 (OMIM:270100); ORPHA:101063; Septopreoptic holoprosencephaly (ORPHA:280195); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925); ORPHA:157769
NOG	Symphalangism, proximal, 1A (OMIM:185800); Brachydactyly, type B2 (OMIM:611377); Stapes ankylosis with broad thumb and toes (OMIM:184460); Tarsal-Carpal coalition syndrome (OMIM:186570); Multiple synostoses syndrome 1 (OMIM:186500); Proximal symphalangism (ORPHA:3250); Multiple synostoses syndrome (ORPHA:3237); Tarsal-carpal coalition syndrome (ORPHA:1412); ORPHA:140917; Brachydactyly type B2 (ORPHA:140908)
NOL3	Myoclonus, familial cortical (OMIM:614937); ORPHA:319189
NOP10	Dyskeratosis congenita, autosomal recessive 1 (OMIM:224230); Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9 (OMIM:620400); Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2 (OMIM:620425); Dyskeratosis congenita (ORPHA:1775)
NOP16	—
NOP56	Spinocerebellar ataxia 36 (OMIM:614153); Spinocerebellar ataxia type 36 (ORPHA:276198)
NOS1	Idiopathic achalasia (ORPHA:930)
NOS1AP	Nephrotic syndrome, type 22 (OMIM:619155); Romano-Ward syndrome (ORPHA:101016)
NOS2	OMIM:611162
NOS3	Moyamoya disease 8 (OMIM:621469)
NOTCH1	Aortic valve disease 1 (OMIM:109730); Adams-Oliver syndrome 5 (OMIM:616028); Adams-Oliver syndrome (ORPHA:974); Familial bicuspid aortic valve (ORPHA:402075)
NOTCH2	Alagille syndrome 2 (OMIM:610205); Hajdu-Cheney syndrome (OMIM:102500); ORPHA:261629; Hajdu-Cheney syndrome (ORPHA:955)
NOTCH3	Myofibromatosis, infantile 2 (OMIM:615293); Lateral meningocele syndrome (OMIM:130720); Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 1 (OMIM:621295); Cerebral arteriopathy, autosomal dominant, with subcortical infarctsand leukoencephalopathy (OMIM:125310); Lipodystrophy, familial partial, type 1 (OMIM:608600); Lateral meningocele syndrome (ORPHA:2789); Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy (ORPHA:136); Infantile myofibromatosis (ORPHA:2591)
NOVA1	—
NOX3	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
NPAS2	—
NPAS3	—
NPAT	Familial colorectal cancer Type X (ORPHA:440437)
NPC1	Niemann-pick disease, type C1 (OMIM:257220); ORPHA:216972; ORPHA:216975; ORPHA:216978; ORPHA:216986; ORPHA:216981
NPC1L1	—
NPC2	Niemann-pick disease, type C2 (OMIM:607625); ORPHA:216972; ORPHA:216975; ORPHA:216978; ORPHA:216986; ORPHA:216981
NPEPL1	—
NPFFR2	—
NPHP1	Joubert syndrome 4 (OMIM:609583); Senior-Loken syndrome 1 (OMIM:266900); Nephronophthisis 1 (OMIM:256100); Joubert syndrome with renal defect (ORPHA:220497); Senior-Loken syndrome (ORPHA:3156); Bardet-Biedl syndrome (ORPHA:110); ORPHA:93592
NPHP3	Meckel syndrome, type 7 (OMIM:267010); Nephronophthisis 3 (OMIM:604387); Renal-hepatic-pancreatic dysplasia 1 (OMIM:208540); Senior-Loken syndrome (ORPHA:3156); NPHP3-related Meckel-like syndrome (ORPHA:3032); Renal-hepatic-pancreatic dysplasia (ORPHA:294415); ORPHA:93591; ORPHA:93589
NPHP4	Senior-Loken syndrome 4 (OMIM:606996); Nephronophthisis 4 (OMIM:606966); Senior-Loken syndrome (ORPHA:3156); ORPHA:93592
NPHS1	Nephrotic syndrome, type 1 (OMIM:256300); Congenital nephrotic syndrome, Finnish type (ORPHA:839); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
NPHS2	Nephrotic syndrome, type 2 (OMIM:600995); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
NPL	—
NPM1	Leukemia, acute myeloid (OMIM:601626); ORPHA:98842; ORPHA:98833; ORPHA:98834; ORPHA:402026; ORPHA:300865; Acute promyelocytic leukemia (ORPHA:520); Dyskeratosis congenita (ORPHA:1775)
NPPA	Atrial standstill 2 (OMIM:615745); Atrial fibrillation, familial, 6 (OMIM:612201); Hereditary atrial fibrillation (ORPHA:334); Isolated atrial standstill (ORPHA:1344)
NPPC	—
NPR2	Short stature with nonspecific skeletal abnormalities (OMIM:616255); Epiphyseal chondrodysplasia, Miura type (OMIM:615923); Acromesomelic dysplasia, Maroteaux type (OMIM:602875); Acromesomelic dysplasia, Maroteaux type (ORPHA:40); ORPHA:329191
NPRL3	Epilepsy, familial focal, with variable foci 3 (OMIM:617118); Familial focal epilepsy with variable foci (ORPHA:98820)
NPSR1	Short sleep, familial natural, 3 (OMIM:621336)
NPY	—
NPY1R	—
NPY2R	—
NQO1	—
NR0B1	Dosage-Sensitive sex reversal (OMIM:300018); Adrenal hypoplasia, congenital (OMIM:300200); 46,XX testicular difference of sex development (ORPHA:393); 46,XY partial gonadal dysgenesis (ORPHA:251510); 46,XY complete gonadal dysgenesis (ORPHA:242); X-linked adrenal hypoplasia congenita (ORPHA:95702)
NR0B2	OBESITY (OMIM:601665)
NR1D1	—
NR1H2	—
NR1H3	—
NR1H4	Cholestasis, progressive familial intrahepatic, 5 (OMIM:617049); Intrahepatic cholestasis of pregnancy (ORPHA:69665); ORPHA:480476
NR1I2	—
NR1I3	—
NR2C2AP	—
NR2E1	—
NR2E3	Retinitis pigmentosa 37 (OMIM:611131); Enhanced S-cone syndrome (OMIM:268100); Retinitis pigmentosa (ORPHA:791); ORPHA:53540
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome (OMIM:615722); Optic atrophy-intellectual disability syndrome (ORPHA:401777)
NR2F2	46,XX sex reversal 5 (OMIM:618901); Congenital heart defects, multiple types, 4 (OMIM:615779); ORPHA:99067; ORPHA:99068
NR3C1	Glucocorticoid resistance (OMIM:615962); Cushing disease (ORPHA:96253); Generalized glucocorticoid resistance syndrome (ORPHA:786)
NR3C2	Pseudohypoaldosteronism, type I, autosomal dominant (OMIM:177735); Hypertension, early-onset, autosomal dominant, with severe exacerbationin pregnancy (OMIM:605115); ORPHA:171871
NR4A1	—
NR4A1AS	—
NR4A2	Parkinson disease, late-onset (OMIM:168600); Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism (OMIM:619911); Autosomal dominant dopa-responsive dystonia (ORPHA:98808); ORPHA:660012; Developmental delay-language impairment-dopa responsive dystonia-parkinsonism syndrome due to 2q24 microdeletion (ORPHA:1617)
NR4A3	Chondrosarcoma, extraskeletal myxoid (OMIM:612237); ORPHA:209916
NR5A1	Premature ovarian failure 7 (OMIM:612964); 46XY sex reversal 3 (OMIM:612965); 46,xx sex reversal 4 (OMIM:617480); Spermatogenic failure 8 (OMIM:613957); 46,XX testicular difference of sex development

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	(ORPHA:393); 46,XY partial gonadal dysgenesis (ORPHA:251510); Male infertility with azoospermia or oligozoospermia due to single gene mutation (ORPHA:399805); 46,XX gonadal dysgenesis (ORPHA:243); 46,XY complete gonadal dysgenesis (ORPHA:242); 46,XX ovotesticular difference of sex development (ORPHA:2138)
NRAS	Epidermal nevus, somatic (OMIM:162900); Neurocutaneous melanosis, somatic (OMIM:249400); Thyroid cancer, nonmedullary, 2 (OMIM:188470); Schimmelpenning-Feuerstein-Mims syndrome (OMIM:163200); Noonan syndrome 6 (OMIM:613224); Colorectal cancer (OMIM:114500); RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic (OMIM:614470); Melanocytic nevus syndrome, congenital (OMIM:137550); Langerhans cell histiocytosis (ORPHA:389); ORPHA:146; Linear nevus sebaceus syndrome (ORPHA:2612); ORPHA:86834; ORPHA:268114; Large/giant congenital melanocytic nevus (ORPHA:626); Noonan syndrome (ORPHA:648)
NRBF2P6	—
NRCAM	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities (OMIM:619833); ORPHA:528084
NRG1	—
NRIP3	—
NRL	Enhanced S-cone syndrome 2 (OMIM:621371); Retinitis pigmentosa 27 (OMIM:613750); Retinitis pigmentosa (ORPHA:791)
NRP2	—
NRTN	Hirschsprung disease (ORPHA:388)
NRXN1	Pitt-Hopkins-Like syndrome 2 (OMIM:614325); Schizophrenia, susceptibility to, 17 (OMIM:621407); ORPHA:600663
NRXN2	—
NRXN3	—
NSD1	Sotos syndrome (OMIM:117550); Sotos syndrome (ORPHA:821); Deletion 5q35 syndrome (ORPHA:1627); Weaver syndrome (ORPHA:3447); 5q35 microduplication syndrome (ORPHA:228415)
NSD2	Wolf-Hirschhorn syndrome (OMIM:194190); Rauch-Steindl syndrome (OMIM:619695); Wolf-Hirschhorn syndrome (ORPHA:280)
NSDHL	Congenital hemidysplasia with ichthyosiform erythroderma and limb defects (OMIM:308050); CK syndrome (OMIM:300831); CHILD syndrome (ORPHA:139); CK syndrome (ORPHA:251383)
NSMF	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Hypogonadotropic hypogonadism 9 with or without anosmia (OMIM:614838); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432)
NSUN2	Intellectual developmental disorder, autosomal recessive 5 (OMIM:611091); Dubowitz syndrome (ORPHA:235); ORPHA:88616
NSUN7	—
NT5C3A	Uridine 5-prime monophosphate hydrolase deficiency, hemolytic anemiadue to (OMIM:266120); ORPHA:35120
NT5DC1	—
NT5E	Calcification of joints and arteries (OMIM:211800); Hereditary arterial and articular multiple calcification syndrome (ORPHA:289601)
NTF3	—
NTF4	Glaucoma 1, open angle, O (OMIM:613100)
NTHL1	Familial adenomatous polyposis 3 (OMIM:616415); NTHL1-related polyposis (ORPHA:454840)
NTNG1	Atypical Rett syndrome (ORPHA:3095); ORPHA:528084
NTRK1	Insensitivity to pain, congenital, with anhidrosis (OMIM:256800); Thyroid carcinoma, familial medullary (OMIM:155240); ORPHA:146; ORPHA:99361; Hereditary sensory and autonomic neuropathy type 5 (ORPHA:64752); Hereditary sensory and autonomic neuropathy type 4 (ORPHA:642)
NTRK2	Developmental and epileptic encephalopathy 58 (OMIM:617830); Obesity, hyperphagia, and developmental delay (OMIM:613886); Infantile epileptic spasms syndrome (ORPHA:697160); Non-specific early-onset epileptic encephalopathy (ORPHA:442835); ORPHA:251615; ORPHA:99704
NTRK3	ORPHA:2030; ORPHA:146; ORPHA:2665
NUAK1	—
NUBPL	Mitochondrial complex I deficiency, nuclear type 21 (OMIM:618242); Isolated complex I deficiency (ORPHA:2609)
NUDC	—
NUDT1	—
NUDT15	THIOPURINES, POOR METABOLISM OF, 2 (OMIM:616903)
NUDT6	—
NUMA1	Acute promyelocytic leukemia (ORPHA:520)
NUP155	Atrial fibrillation, familial, 15 (OMIM:615770); Hereditary atrial fibrillation (ORPHA:334)
NUP214	Leukemia, acute lymphocytic, susceptibility to, 1 (OMIM:613065); Encephalopathy, acute, infection-induced, susceptibility to, 9 (OMIM:618426); Leukemia, acute myeloid (OMIM:601626); ORPHA:99861; ORPHA:402014
NUP62	Striatonigral degeneration, infantile (OMIM:271930); Familial infantile bilateral striatal necrosis (ORPHA:225154)
NUP98	—
NXF5	—
NXNL1	—
NYX	Night blindness, congenital stationary, type 1A (OMIM:310500)
OAS1	Pulmonary alveolar proteinosis with hypogammaglobulinemia (OMIM:618042); ORPHA:572428
OAS2	Autoinflammation and autoimmunity, systemic, with immune dysregulation 2 (OMIM:621409)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia (OMIM:258870); Gyrate atrophy of choroid and retina (ORPHA:414)
OBI1	—
OBSCN	Rhabdomyolysis, susceptibility to, 1 (OMIM:620235); Genetic recurrent myoglobinuria (ORPHA:99845)
OBSL1	3-M syndrome 2 (OMIM:612921); 3M syndrome (ORPHA:2616)
OCA2	OMIM:227220; Albinism, oculocutaneous, type II (OMIM:203200); Oculocutaneous albinism type 2 (ORPHA:79432); Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2 (ORPHA:177904); Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1 (ORPHA:177901); Angelman syndrome due to maternal 15q11q13 deletion (ORPHA:98794); Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15 (ORPHA:98754)
OCLN	Band-Like calcification with simplified gyration and polymicrogyria (OMIM:251290); Pseudo-TORCH syndrome type 1 (ORPHA:1229)
OCRL	Dent disease 2 (OMIM:300555); Lowe syndrome (OMIM:309000); Oculocerebrorenal syndrome of Lowe (ORPHA:534); ORPHA:93623
ODAPH	Amelogenesis imperfecta, hypomaturation type, iia4 (OMIM:614832); ORPHA:100033
ODC1	Neurodevelopmental disorder with alopecia and brain abnormalities (OMIM:619075); Global developmental delay-alopecia-macrocephaly-facial dysmorphism-structural brain anomalies syndrome (ORPHA:544488)
OFCC1	—
OFD1	Retinitis pigmentosa 23 (OMIM:300424); Simpson-Golabi-Behmel syndrome, type 2 (OMIM:300209); Joubert syndrome 10 (OMIM:300804); Orofaciodigital syndrome I (OMIM:311200); Orofaciodigital syndrome type 1 (ORPHA:2750); Orofaciodigital syndrome type 6 (ORPHA:2754); Primary ciliary dyskinesia (ORPHA:244); Retinitis pigmentosa (ORPHA:791); Isolated Joubert syndrome (ORPHA:475)
OGG1	Renal cell carcinoma, nonpapillary (OMIM:144700); ORPHA:422526
OGT	Intellectual developmental disorder, X-linked 106 (OMIM:300997)
OLFM2	—
OLIG2	—
OLR1	OMIM:608446
OMG	—
OPA1	Mitochondrial DNA depletion syndrome 14A (encephalomyopathic type) (OMIM:621481); Optic atrophy 1 (OMIM:165500); Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type) (OMIM:616896); Behr syndrome (OMIM:210000); Optic atrophy with or without deafness, ophthalmoplegia, myopathy, ataxia, and neuropathy (OMIM:125250); OMIM:606657; Autosomal dominant optic atrophy, classic form (ORPHA:98673); ORPHA:1239; Autosomal dominant optic atrophy plus syndrome (ORPHA:1215)
OPA3	Optic atrophy 3, autosomal dominant (OMIM:165300); 3-methylglutaconic aciduria, type III (OMIM:258501); Autosomal dominant optic atrophy and cataract (ORPHA:67036); 3-methylglutaconic aciduria type 3 (ORPHA:67047)
OPCML	Ovarian cancer (OMIM:167000)
OPHN1	Intellectual developmental disorder, X-linked syndromic, Billuart type (OMIM:300486); X-linked intellectual disability-cerebellar hypoplasia syndrome (ORPHA:137831)
OPLAH	5-@oxoprolinase deficiency (OMIM:260005); 5-oxoprolinase deficiency (ORPHA:33572)
OPN1SW	TRITANOPIA (OMIM:190900); Tritanopia (ORPHA:88629)
OPRL1	—
OPRM1	—
OPTC	—
OPTN	Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia (OMIM:613435); OMIM:606657; Glaucoma, primary open angle (OMIM:137760); Amyotrophic lateral sclerosis (ORPHA:803)
OR10X1	—
OR10Z1	—
OR13G1	—
OR1B1	—
OR51G1	—
OR52H1	—
OR5AC2	—
OR8K3	—
OR9H1P	—
ORAI1	Immunodeficiency 9 (OMIM:612782); Myopathy, tubular aggregate, 2 (OMIM:615883); Stormorken-Sjaastad-Langslet syndrome (ORPHA:3204); Tubular aggregate myopathy (ORPHA:2593); ORPHA:317428
ORC1	Meier-Gorlin syndrome 1 (OMIM:224690); Ear-patella-short stature syndrome (ORPHA:2554)
ORC4	Meier-Gorlin syndrome 2 (OMIM:613800); Ear-patella-short stature syndrome (ORPHA:2554)
ORC6	Meier-Gorlin syndrome 3 (OMIM:613803); Ear-patella-short stature syndrome (ORPHA:2554)
OSMR	Amyloidosis, primary localized cutaneous, 1 (OMIM:105250); ORPHA:353220
OSTM1	Osteopetrosis, autosomal recessive 5 (OMIM:259720); Infantile osteopetrosis with neuroaxonal dysplasia (ORPHA:85179)
OT1	—
OTC	Ornithine transcarbamylase deficiency, hyperammonemia due to (OMIM:311250); Ornithine transcarbamylase deficiency (ORPHA:664)
OTOA	Deafness, autosomal recessive 22 (OMIM:607039); ORPHA:90636
OTOF	Deafness, autosomal recessive 9 (OMIM:601071); ORPHA:90636
OTOG	Deafness, autosomal recessive 18B (OMIM:614945); ORPHA:90636

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
OTOGL	Deafness, autosomal recessive 84B (OMIM:614944); ORPHA:90636
OTOP2	—
OTULIN	Autoinflammation, panniculitis, and dermatosis syndrome, autosomal dominant (OMIM:621030); Immunodeficiency 107, susceptibility to invasive staphylococcus aureus infection (OMIM:619986); Autoinflammation, panniculitis, and dermatosis syndrome (OMIM:617099); ORPHA:500062
OTX2	Microphthalmia, syndromic 5 (OMIM:610125); Pituitary hormone deficiency, combined, 6 (OMIM:613986); ORPHA:99001; Septo-optic dysplasia spectrum (ORPHA:3157); Combined pituitary hormone deficiencies, genetic forms (ORPHA:95494); ORPHA:178364; Agnathia-holoprosencephaly-situs inversus syndrome (ORPHA:990); ORPHA:98938; Nanophthalmos (ORPHA:35612)
OVCH2	—
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency (OMIM:245050); Succinyl-CoA:3-oxoacid CoA transferase deficiency (ORPHA:832)
OXSM	—
P2RX1	—
P2RX5	—
P2RX7	ORPHA:67038
P2RY12	Bleeding disorder, platelet-type, 8 (OMIM:609821); ORPHA:36355
P2RY4	—
P3H1	Osteogenesis imperfecta, type VIII (OMIM:610915); ORPHA:216812; ORPHA:216804
P3H2	Myopia, high, with cataract and vitreoretinal degeneration (OMIM:614292); ORPHA:98619
P4HA2	Myopia 25, autosomal dominant (OMIM:617238); Giant cell arteritis (ORPHA:397)
PABPC4L	—
PABPN1	Oculopharyngeal muscular dystrophy (OMIM:164300); Oculopharyngeal muscular dystrophy (ORPHA:270)
PACERR	—
PACRG	—
PACRGL	—
PACS1	Schuurs-Hoeijmakers syndrome (OMIM:615009); Schuurs-Hoeijmakers syndrome (ORPHA:329224)
PACS2	Epileptic encephalopathy, early infantile, 66 (OMIM:618067); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
PADI4	—
PAFAH1B1	Lissencephaly 1 (OMIM:607432); 17p13.3 microduplication syndrome (ORPHA:217385); Miller-Dieker syndrome (ORPHA:531); Lissencephaly due to LIS1 mutation (ORPHA:95232); ORPHA:99796
PAFAH1B3	—
PAG1	—
PAH	Phenylketonuria (OMIM:261600); ORPHA:708895; ORPHA:293284; Maternal phenylketonuria syndrome (ORPHA:2209)
PAK3	Intellectual developmental disorder, X-linked 30 (OMIM:300558); ORPHA:528084
PAK6	—
PALB2	Fanconi anemia, complementation group N (OMIM:610832); Pancreatic cancer, susceptibility to, 3 (OMIM:613348); Breast-ovarian cancer, familial, susceptibility to, 5 (OMIM:620442); Fanconi anemia (ORPHA:84); Hereditary breast and/or ovarian cancer syndrome (ORPHA:145); ORPHA:178; ORPHA:227535; Familial pancreatic carcinoma (ORPHA:1333)
PALD1	—
PALLD	Pancreatic cancer, susceptibility to, 1 (OMIM:606856); Familial pancreatic carcinoma (ORPHA:1333)
PALM2AKAP2	—
PALS2	—
PAM16	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melki type (OMIM:613320); ORPHA:401979
PAMR1	—
PANK2	Neurodegeneration with brain iron accumulation 1 (OMIM:234200); Atypical pantothenate kinase-associated neurodegeneration (ORPHA:216873); Classic pantothenate kinase-associated neurodegeneration (ORPHA:216866)
PAPSS2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes (OMIM:612847); ORPHA:93282; ORPHA:448242
PARD3B	—
PARK7	Parkinson disease 7, autosomal recessive early-onset (OMIM:606324); Young-onset Parkinson disease (ORPHA:2828); ORPHA:90020
PARL	—
PARP1	—
PASK	—
PAX1	Otofaciocervical syndrome 2 (OMIM:615560); Otofaciocervical syndrome (ORPHA:2792)
PAX2	Papillorenal syndrome (OMIM:120330); Focal segmental glomerulosclerosis 7 (OMIM:616002); Renal coloboma syndrome (ORPHA:1475); Renal hypoplasia, bilateral (ORPHA:97362); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
PAX3	Waardenburg syndrome, type 3 (OMIM:148820); Craniofacial-deafness-hand syndrome (OMIM:122880); Waardenburg syndrome, type 1 (OMIM:193500); Rhabdomyosarcoma 2, alveolar (OMIM:268220); Craniofacial-deafness-hand syndrome (ORPHA:1529); Waardenburg syndrome type 3 (ORPHA:896); Waardenburg syndrome type 1 (ORPHA:894); ORPHA:99756
PAX4	Diabetes mellitus, noninsulin-dependent (OMIM:125853); Maturity-onset diabetes of the young, type IX (OMIM:612225); Diabetes mellitus, ketosis-prone (OMIM:612227); MODY (ORPHA:552)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
PAX5	OMIM:615545; ORPHA:641372
PAX6	Foveal hypoplasia and presenile cataract syndromefoveal hypoplasia, isolated, included (OMIM:136520); Anterior segment dysgenesis 5, multiple subtypes (OMIM:604229); Optic nerve hypoplasia (OMIM:165550); Wilms tumor, aniridia, genitourinary anomalies and impaired intellectual development syndrome (OMIM:194072); Coloboma, ocular (OMIM:120200); Coloboma of optic nerveoptic nerve head pits, bilateral congenital, included (OMIM:120430); Aniridia (OMIM:106210); Keratitis, hereditary (OMIM:148190); Isolated aniridia (ORPHA:250923); WAGR syndrome (ORPHA:893); ORPHA:98947; Aniridia-cerebellar ataxia-intellectual disability syndrome (ORPHA:1065); ORPHA:98943; ORPHA:98944; ORPHA:98945; ORPHA:98946; ORPHA:98942; Autosomal dominant keratitis (ORPHA:2334); Peters anomaly (ORPHA:708); Morning glory disc anomaly (ORPHA:35737); Foveal hypoplasia-presenile cataract syndrome (ORPHA:2253)
PAX7	Myopathy, congenital, progressive, with scoliosis (OMIM:618578); Rhabdomyosarcoma 2, alveolar (OMIM:268220); ORPHA:99756
PAX8	Hypothyroidism, congenital, nongoitrous, 2 (OMIM:218700); ORPHA:146; Thyroid hypoplasia (ORPHA:95720); Athyreosis (ORPHA:95713); Thyroid ectopia (ORPHA:95712)
PAX9	Tooth agenesis, selective, 3 (OMIM:604625); Oligodontia (ORPHA:99798)
PBX1	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay (OMIM:617641); Renal hypoplasia, bilateral (ORPHA:97362); ORPHA:585956; ORPHA:656130
PC	Pyruvate carboxylase deficiency (OMIM:266150); ORPHA:353320; ORPHA:353314; ORPHA:353308
PCARE	Retinitis pigmentosa 54 (OMIM:613428); Retinitis pigmentosa (ORPHA:791)
PCAT7	—
PCBD1	Hyperphenylalaninemia, BH4-deficient, D (OMIM:264070); Pterin-4 alpha-carbinolamine dehydratase deficiency (ORPHA:1578)
PCBD2	—
PCBP3	—
PCCA	Propionic acidemia (OMIM:606054); Propionic acidemia (ORPHA:35)
PCCB	Propionic acidemia (OMIM:606054); Propionic acidemia (ORPHA:35)
PCDH11X	—
PCDH15	Usher syndrome, type ID (OMIM:601067); Deafness, autosomal recessive 23 (OMIM:609533); Usher syndrome, type IF (OMIM:602083); ORPHA:90636; Usher syndrome type 1 (ORPHA:231169)
PCDH18	—
PCDH19	Epileptic encephalopathy, early infantile, 9 (OMIM:300088); PCDH19 clustering epilepsy (ORPHA:714652); Dravet syndrome (ORPHA:33069)
PCDHA1	—
PCDHA10	—
PCDHA3	—
PCDHB4	—
PCK2	Phosphoenolpyruvate carboxykinase 2, mitochondrial (OMIM:261650); Phosphoenolpyruvate carboxykinase deficiency (ORPHA:2880)
PCLO	Pontocerebellar hypoplasia, type 3 (OMIM:608027); ORPHA:97249
PCM1	ORPHA:146
PCMT1	—
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II (OMIM:210720); Microcephalic osteodysplastic primordial dwarfism type II (ORPHA:2637); Seckel syndrome (ORPHA:808)
PCOLCE	—
PCP4	—
PCSK1	Proprotein convertase 1/3 deficiency (OMIM:600955); OMIM:612362; Obesity due to prohormone convertase I deficiency (ORPHA:71528)
PCSK5	—
PCSK9	Hypercholesterolemia, familial, 3 (OMIM:603776); Homozygous familial hypercholesterolemia (ORPHA:391665)
PDCD1	Autoimmune disease with susceptibility to mycobacterium tuberculosis (OMIM:621004); Systemic lupus erythematosus (ORPHA:536)
PDCD10	Cerebral cavernous malformations 3 (OMIM:603285); Cerebral cavernous malformations 1 (OMIM:116860); Familial cerebral cavernous malformation (ORPHA:221061)
PDCD6	—
PDE11A	Pigmented nodular adrenocortical disease, primary, 2 (OMIM:610475); ORPHA:647782; ORPHA:647772; Carney complex (ORPHA:1359)
PDE12	—
PDE4B	—
PDE4D	Acrodysostosis 2 with or without hormone resistance (OMIM:614613); PDE4D haploinsufficiency syndrome (ORPHA:439822); Acrodysostosis (ORPHA:950)
PDE6A	Retinitis pigmentosa 43 (OMIM:613810); Retinitis pigmentosa (ORPHA:791)
PDE6B	Night blindness, congenital stationary, autosomal dominant 2 (OMIM:163500); Retinitis pigmentosa 40 (OMIM:613801); ORPHA:714096; Retinitis pigmentosa (ORPHA:791)
PDE6C	Cone dystrophy 4 (OMIM:613093); Progressive cone dystrophy (ORPHA:1871); Achromatopsia (ORPHA:49382)
PDE6G	Retinitis pigmentosa (OMIM:268000); Retinitis pigmentosa 57 (OMIM:613582); Retinitis pigmentosa (ORPHA:791)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
PDE6H	Retinal cone dystrophy 3A (OMIM:610024); Achromatopsia (ORPHA:49382)
PDE8B	Striatal degeneration, autosomal dominant (OMIM:609161); Pigmented nodular adrenocortical disease, primary, 3 (OMIM:614190); Autosomal dominant striatal neurodegeneration (ORPHA:228169); ORPHA:647782
PDGFB	Meningioma, familial, susceptibility to (OMIM:607174); Basal ganglia calcification, idiopathic, 1 (OMIM:213600); Basal ganglia calcification, idiopathic, 5 (OMIM:615483); Dermatofibrosarcoma protuberans (ORPHA:31112); Bilateral striopallidodentate calcinosis (ORPHA:1980); Meningioma (ORPHA:2495); ORPHA:263662
PDGFRA	Hypereosinophilic syndrome, idiopathic (OMIM:607685); Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial (OMIM:175510); Cleft lip/palate (ORPHA:199306); ORPHA:168947; ORPHA:168940; ORPHA:314950; Gastrointestinal stromal tumor (ORPHA:44890); ORPHA:585877
PDGFRB	Kosaki overgrowth syndrome (OMIM:616592); Ocular pterygium-digital keloid dysplasia syndrome (OMIM:621091); Basal ganglia calcification, idiopathic, 4 (OMIM:615007); Basal ganglia calcification, idiopathic, 1 (OMIM:213600); Myofibromatosis, infantile, 1 (OMIM:228550); Premature aging syndrome, Penttinen type (OMIM:601812); ORPHA:477831; ORPHA:168950; ORPHA:314950; Infantile myofibromatosis (ORPHA:2591); ORPHA:86830; Bilateral striopallidodentate calcinosis (ORPHA:1980); ORPHA:363665
PDGFRL	Colorectal cancer (OMIM:114500); Hepatocellular carcinoma (OMIM:114550); ORPHA:210159
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency (OMIM:312170); Pyruvate dehydrogenase E1-alpha deficiency (ORPHA:79243)
PDHB	Pyruvate dehydrogenase E1-beta deficiency (OMIM:614111); Pyruvate dehydrogenase E1-beta deficiency (ORPHA:255138)
PDHX	Pyruvate dehydrogenase e3-binding protein deficiency (OMIM:245349); Pyruvate dehydrogenase E3-binding protein deficiency (ORPHA:255182)
PDK1	—
PDLIM1P4	—
PDLIM3	—
PDLIM4	OSTEOPOROSIS (OMIM:166710)
PDP1	Pyruvate dehydrogenase phosphatase deficiency (OMIM:608782); Pyruvate dehydrogenase phosphatase deficiency (ORPHA:79246)
PDPK1	—
PDPR2P	—
PDSS1	Coenzyme Q10 deficiency, primary, 2 (OMIM:614651); ORPHA:254898
PDSS2	Coenzyme Q10 deficiency, primary, 3 (OMIM:614652)
PDX1	Diabetes mellitus, noninsulin-dependent (OMIM:125853); Pancreatic agenesis, congenital (OMIM:260370); Maturity-onset diabetes of the young, type 4 (OMIM:606392); Isolated permanent neonatal diabetes mellitus (ORPHA:99885); ORPHA:2805; MODY (ORPHA:552)
PDYN	Spinocerebellar ataxia 23 (OMIM:610245); Spinocerebellar ataxia type 23 (ORPHA:101108)
PDZD7	Usher syndrome, type IIA (OMIM:276901); Deafness, autosomal recessive 57 (OMIM:618003); Usher syndrome, type IIC (OMIM:605472); Usher syndrome type 2 (ORPHA:231178)
PECR	—
PELO	—
PEPD	Prolidase deficiency (OMIM:170100); Prolidase deficiency (ORPHA:742)
PER1	—
PER2	Advanced sleep phase syndrome, familial, 1 (OMIM:604348); ORPHA:164736
PEX1	Peroxisome biogenesis disorder 1A (Zellweger) (OMIM:214100); Heimler syndrome 1 (OMIM:234580); Peroxisome biogenesis disorder 1B (OMIM:601539); Deafness-enamel hypoplasia-nail defects syndrome (ORPHA:3220); Neonatal adrenoleukodystrophy (ORPHA:44); Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772)
PEX10	Peroxisome biogenesis disorder 6A (Zellweger) (OMIM:614870); Peroxisome biogenesis disorder 6B (OMIM:614871); Neonatal adrenoleukodystrophy (ORPHA:44); Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772); Autosomal recessive ataxia due to PEX10 deficiency (ORPHA:247815)
PEX11B	Peroxisome biogenesis disorder 14B (OMIM:614920); Neonatal adrenoleukodystrophy (ORPHA:44); Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772)
PEX12	Peroxisome biogenesis disorder 3B (OMIM:266510); Peroxisome biogenesis disorder 3A (Zellweger) (OMIM:614859); Neonatal adrenoleukodystrophy (ORPHA:44); Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772)
PEX13	Peroxisome biogenesis disorder 11B (OMIM:614885); Peroxisome biogenesis disorder 11A (Zellweger) (OMIM:614883); Neonatal adrenoleukodystrophy (ORPHA:44); Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772)
PEX14	Peroxisome biogenesis disorder 13A (Zellweger) (OMIM:614887); Neonatal adrenoleukodystrophy (ORPHA:44); Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772)
PEX16	Peroxisome biogenesis disorder 8B (OMIM:614877); Peroxisome biogenesis disorder 8A (Zellweger) (OMIM:614876); Neonatal adrenoleukodystrophy (ORPHA:44); ORPHA:642954; Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772)
PEX19	Peroxisome biogenesis disorder 12A (Zellweger) (OMIM:614886); Neonatal adrenoleukodystrophy (ORPHA:44); Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772)
PEX2	Peroxisome biogenesis disorder 5A (Zellweger) (OMIM:614866); Peroxisome biogenesis disorder 5B (OMIM:614867); Neonatal adrenoleukodystrophy (ORPHA:44); ORPHA:642965; Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772)
PEX26	Peroxisome biogenesis disorder 7B (OMIM:614873); Peroxisome biogenesis disorder 7A (zellweger)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	(OMIM:614872); Neonatal adrenoleukodystrophy (ORPHA:44); Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772)
PEX3	Peroxisome biogenesis disorder 10B (OMIM:617370); Peroxisome biogenesis disorder 10A (Zellweger) (OMIM:614882); Neonatal adrenoleukodystrophy (ORPHA:44); Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772)
PEX5	Rhizomelic chondrodysplasia punctata, type 5 (OMIM:616716); Peroxisome biogenesis disorder 2A (Zellweger) (OMIM:214110); Peroxisome biogenesis disorder 2B (OMIM:202370); Neonatal adrenoleukodystrophy (ORPHA:44); Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772); ORPHA:468717
PEX6	Heimler syndrome 2 (OMIM:616617); Peroxisome biogenesis disorder 4A (Zellweger) (OMIM:614862); Peroxisome biogenesis disorder 4B (OMIM:614863); Autosomal recessive spinocerebellar ataxia-blindness-deafness syndrome (ORPHA:95433); Deafness-enamel hypoplasia-nail defects syndrome (ORPHA:3220); Neonatal adrenoleukodystrophy (ORPHA:44); Zellweger syndrome (ORPHA:912); Infantile Refsum disease (ORPHA:772)
PEX7	Rhizomelic chondrodysplasia punctata, type 1 (OMIM:215100); Refsum disease (OMIM:266500); Peroxisome biogenesis disorder 9B (OMIM:614879); ORPHA:309789; Adult Refsum disease (ORPHA:773)
PFAS	—
PFKM	Glycogen storage disease VII (OMIM:232800); Glycogen storage disease due to muscle phosphofructokinase deficiency (ORPHA:371)
PFN1	Amyotrophic lateral sclerosis 18 (OMIM:614808); Paget disease of bone 7, early-onset (OMIM:621600); Amyotrophic lateral sclerosis (ORPHA:803)
PGAM2	Phosphoglycerate mutase, muscle, deficiency of (OMIM:261670); ORPHA:97234
PGAM5	—
PGAP6	—
PGK1	Phosphoglycerate kinase 1 deficiency (OMIM:300653); Glycogen storage disease due to phosphoglycerate kinase 1 deficiency (ORPHA:713)
PGLS	—
PGM1	Congenital disorder of glycosylation, type It (OMIM:614921); ORPHA:319646
PGR	Progesterone resistance (OMIM:264080)
PGRMC1	ORPHA:98994
PHB1	Breast cancer (OMIM:114480)
PHEX	Hypophosphatemic rickets, X-linked dominant (OMIM:307800); X-linked hypophosphatemia (ORPHA:89936)
PHF11	—
PHF2	—
PHF3	—
PHF6	Borjeson-Forssman-Lehmann syndrome (OMIM:301900); Borjeson-Forssman-Lehmann syndrome (ORPHA:127)
PHF8	Intellectual developmental disorder, X-linked syndromic, Siderius type (OMIM:300263); X-linked intellectual disability, Siderius type (ORPHA:85287)
PHGDH	Neu-Laxova syndrome 1 (OMIM:256520); Phosphoglycerate dehydrogenase deficiency (OMIM:601815); 3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form (ORPHA:79351); ORPHA:583607
PHIP	Developmental delay, intellectual disability, obesity, and dysmorphic features (OMIM:617991); PHIP-related behavioral problems-intellectual disability-obesity-dysmorphic features syndrome (ORPHA:589905)
PHKA1	Muscle glycogenosis, X-linked (OMIM:300559); Glycogen storage disease due to muscle phosphorylase kinase deficiency (ORPHA:715)
PHKA2	Glycogen storage disease, type IXa1 (OMIM:306000); Glycogen storage disease due to liver phosphorylase kinase deficiency (ORPHA:264580)
PHKB	Glycogen storage disease type IXb (OMIM:261750); Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency (ORPHA:79240)
PHKG2	Glycogen storage disease IXc (OMIM:613027); Glycogen storage disease due to liver phosphorylase kinase deficiency (ORPHA:264580)
PHLPP2	—
PHOX2A	Fibrosis of extraocular muscles, congenital, 2 (OMIM:602078); Congenital fibrosis of extraocular muscles (ORPHA:45358)
PHOX2B	Neuroblastoma, susceptibility to, 2 (OMIM:613013); Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease (OMIM:209880); Haddad syndrome (ORPHA:99803); Congenital central hypoventilation syndrome (ORPHA:661); Neuroblastoma (ORPHA:635); Hirschsprung disease-ganglioneuroblastoma syndrome (ORPHA:2151)
PHYH	Refsum disease (OMIM:266500); Adult Refsum disease (ORPHA:773)
PI4KA	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis (OMIM:616531); Spastic paraplegia 84, autosomal recessive (OMIM:619621); Gastrointestinal defects and immunodeficiency syndrome 2 (OMIM:619708); Bilateral perisylvian polymicrogyria (ORPHA:98889); ORPHA:631079; Combined immunodeficiency-multiple intestinal atresia (ORPHA:436252)
PICALM	Leukemia, acute myeloid (OMIM:601626); ORPHA:99861
PICK1	ORPHA:171709
PIF1	—
PIGA	Neurodevelopmental disorder with epilepsy and hemochromatosis (OMIM:301072); Multiple congenital anomalies-hypotonia-seizures syndrome 2 (OMIM:300868); Paroxysmal nocturnal hemoglobinuria (OMIM:300818); ORPHA:300496; Infantile epileptic spasms syndrome (ORPHA:697160); Epilepsy of infancy

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	with migrating focal seizures (ORPHA:293181); ORPHA:397922; Paroxysmal nocturnal hemoglobinuria (ORPHA:447)
PIGL	CHIME syndrome (OMIM:280000); Hyperphosphatasia-intellectual disability syndrome (ORPHA:247262); CHIME syndrome (ORPHA:3474)
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1 (OMIM:614080); Fryns syndrome (ORPHA:2059); Multiple congenital anomalies-hypotonia-seizures syndrome (ORPHA:280633)
PIGO	Hyperphosphatasia with impaired intellectual development syndrome 2 (OMIM:614749); Hyperphosphatasia-intellectual disability syndrome (ORPHA:247262)
PIGR	—
PIGV	Hyperphosphatasia with impaired intellectual development syndrome 1 (OMIM:239300); Hyperphosphatasia-intellectual disability syndrome (ORPHA:247262)
PIGZ	—
PIH1D1	—
PIK3CA	Epidermal nevus, somatic (OMIM:162900); Congenital lipomatous overgrowth, vascular malformations, and epidermal nevi (OMIM:612918); Cowden syndrome 5 (OMIM:615108); Capillary malformation of the lower lip, lymphatic malformation of face and neck, asymmetry of face and limbs, and partial/generalized overgrowth (OMIM:613089); Keratosis, seborrheic, somatic (OMIM:182000); Breast cancer (OMIM:114480); Cerebral cavernous malformations 4 (OMIM:619538); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic (OMIM:602501); Colorectal cancer (OMIM:114500); Ovarian cancer (OMIM:167000); Hepatocellular carcinoma (OMIM:114550); Hemifacial myohyperplasia, somatic (OMIM:606773); Lung cancer, susceptibility to (OMIM:211980); Gastric cancer, somatic (OMIM:613659); Macrodactyly, somatic (OMIM:155500); ORPHA:295243; ORPHA:295239; Hemimegalencephaly (ORPHA:99802); ORPHA:210159; CLAPO syndrome (ORPHA:168984); Cowden syndrome (ORPHA:201); Megalencephaly-capillary malformation-polymicrogyria syndrome (ORPHA:60040); Hemihyperplasia-multiple lipomatosis syndrome (ORPHA:276280); Lynch syndrome (ORPHA:144); Capillary-lymphatic-venous malformation with segmental distribution (ORPHA:90308); CLOVES syndrome (ORPHA:140944); Meningioma (ORPHA:2495); ORPHA:714737; Segmental progressive overgrowth syndrome with fibroadipose hyperplasia (ORPHA:314662); Familial cerebral cavernous malformation (ORPHA:221061)
PIK3CB	—
PIK3CD	Immunodeficiency 14 (OMIM:615513); Roifman-Chitayat syndrome (OMIM:613328); Immunodeficiency 14B, autosomal recessive (OMIM:619281); Activated PI3K-delta syndrome 1 (ORPHA:693661); Combined immunodeficiency with facio-oculo-skeletal anomalies (ORPHA:221139); Autosomal non-syndromic agammaglobulinemia (ORPHA:33110)
PIK3R1	Agammaglobulinemia 7, autosomal recessive (OMIM:615214); Short syndrome (OMIM:269880); Immunodeficiency 36 (OMIM:616005); SHORT syndrome (ORPHA:3163); Activated PI3K-delta syndrome 2 (ORPHA:693681); Autosomal non-syndromic agammaglobulinemia (ORPHA:33110)
PIK3R2	Megalencephaly-Polymicrogyria-Polydactyly-Hydrocephalus syndrome (OMIM:603387); Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome (ORPHA:83473)
PIK3R5	Ataxia-Oculomotor apraxia 3 (OMIM:615217); Spinocerebellar ataxia with axonal neuropathy type 2 (ORPHA:64753)
PIKFYVE	Corneal fleck dystrophy (OMIM:121850); ORPHA:98970
PIM1	—
PIN1	—
PINK1	Parkinson disease 6, autosomal recessive early-onset (OMIM:605909); Young-onset Parkinson disease (ORPHA:2828)
PINX1	—
PIP5K1C	Lethal congenital contracture syndrome 3 (OMIM:611369); ORPHA:137783
PIPOX	—
PITPNA	—
PITPNM3	Cone-Rod dystrophy 5 (OMIM:600977); Cone rod dystrophy (ORPHA:1872)
PITX1	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly (OMIM:119800); Brachydactyly-elbow wrist dysplasia syndrome (ORPHA:1275); ORPHA:293144; ORPHA:293150; ORPHA:498494
PITX2	Ring dermoid of cornea (OMIM:180550); Axenfeld-Rieger syndrome, type 1 (OMIM:180500); Iridogoniodysgenesis, type 2 (OMIM:137600); Hereditary atrial fibrillation (ORPHA:334); Axenfeld-Rieger syndrome (ORPHA:782); ORPHA:98978; ORPHA:91481; ORPHA:91483; Peters anomaly (ORPHA:708)
PITX3	Cataract 11, multiple types (OMIM:610623); Anterior segment dysgenesis 1 (OMIM:107250); ORPHA:162; ORPHA:98993
PJVK	Deafness, autosomal recessive 59 (OMIM:610220); ORPHA:90636
PKD1	Polycystic kidneys (OMIM:173900); Autosomal dominant polycystic kidney disease (ORPHA:730); ORPHA:88924
PKD2	Polycystic kidney disease 2 (OMIM:613095); Autosomal dominant polycystic kidney disease (ORPHA:730)
PKD2L2	—
PKHD1	Polycystic kidney disease 4 with or without polycystic liver disease (OMIM:263200); Caroli disease (ORPHA:53035); Autosomal recessive polycystic kidney disease (ORPHA:731)
PKIG	—
PKLR	Pyruvate kinase deficiency of red cells (OMIM:266200); Adenosine triphosphate, elevated, of erythrocytes (OMIM:102900); Hemolytic anemia due to red cell pyruvate kinase deficiency (ORPHA:766)
PKM	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
PKN3	—
PKP1	Ectodermal dysplasia/skin fragility syndrome (OMIM:604536); Ectodermal dysplasia-skin fragility syndrome (ORPHA:158668)
PKP2	Arrhythmogenic right ventricular dysplasia, familial, 9 (OMIM:609040); ORPHA:54260; Brugada syndrome (ORPHA:130); ORPHA:293888; ORPHA:293899; ORPHA:293910
PLA2G10	—
PLA2G2A	Colorectal cancer (OMIM:114500)
PLA2G4A	Gastrointestinal ulceration, recurrent, with dysfunctional platelets (OMIM:618372); ORPHA:477787; ORPHA:468635
PLA2G5	Fleck retina, familial benign (OMIM:228980); ORPHA:363989
PLA2G6	Parkinson disease 14, autosomal recessive (OMIM:612953); Neurodegeneration with brain iron accumulation 2B (OMIM:610217); Neurodegeneration with brain iron accumulation 2A (OMIM:256600); Adult-onset dystonia-parkinsonism (ORPHA:199351); Infantile neuroaxonal dystrophy (ORPHA:35069)
PLA2G7	Platelet-Activating factor acetylhydrolase deficiency (OMIM:614278)
PLAG1	Silver-Russell syndrome 4 (OMIM:618907); Salivary gland adenoma, pleomorphic (OMIM:181030); ORPHA:276148; Silver-Russell syndrome due to a point mutation (ORPHA:397590)
PLAGL1	Diabetes mellitus, transient neonatal, 1 (OMIM:601410); Transient neonatal diabetes mellitus (ORPHA:99886); Paternal uniparental disomy of chromosome 6 syndrome (ORPHA:96191)
PLAU	Alzheimer disease (OMIM:104300); Quebec platelet disorder (OMIM:601709); ORPHA:220436
PLBD1	—
PLCB1	Developmental and epileptic encephalopathy 12 (OMIM:613722); Infantile epileptic spasms syndrome (ORPHA:697160); Epilepsy of infancy with migrating focal seizures (ORPHA:293181)
PLCB4	Auriculocondylar syndrome 2 (OMIM:614669); Auriculocondylar syndrome 2B (OMIM:620458); Auriculocondylar syndrome (ORPHA:137888)
PLCD1	Nail disorder, nonsyndromic congenital, 3 (OMIM:151600); Leukonychia totalis (ORPHA:2387)
PLCE1	Nephrotic syndrome, type 3 (OMIM:610725); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
PLCG1	Immune dysregulation, autoimmunity, and autoinflammation (OMIM:620514)
PLCG2	Familial cold autoinflammatory syndrome 3 (OMIM:614468); Autoinflammation, antibody deficiency, and immune dysregulation, plcg2-associated (OMIM:614878); ORPHA:300359; ORPHA:324530
PLCZ1	Spermatogenic failure 17 (OMIM:617214)
PLD2	—
PLEC	Epidermolysis bullosa simplex with muscular dystrophy (OMIM:226670); Epidermolysis bullosa simplex with nail dystrophy (OMIM:616487); Epidermolysis bullosa, junctional, with pyloric atresia (OMIM:226730); Epidermolysis bullosa simplex, Ogna type (OMIM:131950); Epidermolysis bullosa simplex with pyloric atresia (OMIM:612138); Muscular dystrophy, limb-girdle, type 2Q (OMIM:613723); PLEC-related intermediate epidermolysis bullosa simplex without extracutaneous involvement (ORPHA:79401); Epidermolysis bullosa simplex with muscular dystrophy (ORPHA:257); Epidermolysis bullosa simplex with pyloric atresia (ORPHA:158684); Plectin-related limb-girdle muscular dystrophy R17 (ORPHA:254361); Aplasia cutis congenita (ORPHA:1114)
PLEKHG3	—
PLEKHG4	Spinocerebellar ataxia type 4 (ORPHA:98765)
PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate C (OMIM:615376); Spinal muscular atrophy, distal, autosomal recessive, 4 (OMIM:611067); ORPHA:206580; ORPHA:369867
PLG	Plasminogen deficiency, type I (OMIM:217090); Angioedema, hereditary, 4 (OMIM:619360); ORPHA:537072; Hypoplasminogenemia (ORPHA:722)
PLIN1	Lipodystrophy, familial partial, type 4 (OMIM:613877); PLIN1-related familial partial lipodystrophy (ORPHA:280356)
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1 (OMIM:225400); Kyphoscoliotic Ehlers-Danlos syndrome due to lysyl hydroxylase 1 deficiency (ORPHA:1900)
PLOD2	Bruck syndrome 2 (OMIM:609220); Bruck syndrome (ORPHA:2771)
PLOD3	Bone fragility with contractures, arterial rupture, and deafness (OMIM:612394); ORPHA:300284
PLP1	Spastic paraplegia 2, X-linked (OMIM:312920); Pelizaeus-Merzbacher disease (OMIM:312080); Spastic paraplegia type 2 (ORPHA:99015); Pelizaeus-Merzbacher disease, connatal form (ORPHA:280210); Pelizaeus-Merzbacher disease, transitional form (ORPHA:280224); Pelizaeus-Merzbacher disease, classic form (ORPHA:280219); Null syndrome (ORPHA:280234); Pelizaeus-Merzbacher disease in female carriers (ORPHA:280229); ORPHA:599376
PLP2	—
PLSCR3	—
PLTP	—
PLXNB3	—
PML	Acute promyelocytic leukemia (ORPHA:520)
PMM2	Congenital disorder of glycosylation, type Ia (OMIM:212065); PMM2-CDG (ORPHA:79318)
PMP22	Roussy-Levy hereditary areflexic dystasia (OMIM:180800); Neuropathy, inflammatory demyelinating (OMIM:139393); Charcot-Marie-Tooth disease, demyelinating, type 1A (OMIM:118220); Neuropathy, hereditary, with liability to pressure palsies (OMIM:162500); Charcot-Marie-Tooth disease and deafness (OMIM:118300); Hypertrophic neuropathy of dejerine-sottas (OMIM:145900); Charcot-Marie-Tooth disease type 1E (ORPHA:90658); Charcot-Marie-Tooth disease type 1A (ORPHA:101081); Roussy-Lévy syndrome (ORPHA:3115); ORPHA:64748; Acute inflammatory demyelinating polyradiculoneuropathy (ORPHA:98916); Hereditary neuropathy with liability to pressure palsies (ORPHA:640)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
PMS1	Lynch syndrome I (OMIM:120435); Lynch syndrome (ORPHA:144)
PMS2	Colorectal cancer, hereditary nonpolyposis, type 4 (OMIM:614337); Mismatch repair cancer syndrome 4 (OMIM:619101); Lynch syndrome (ORPHA:144); ORPHA:252202
PNCK	—
PNKD	Paroxysmal nonkinesigenic dyskinesia 1 (OMIM:118800); Paroxysmal non-kinesigenic dyskinesia (ORPHA:98810)
PNKP	Ataxia-oculomotor apraxia 4 (OMIM:616267); Charcot-Marie-Tooth disease, axonal, type 2B2 (OMIM:605589); Microcephaly, seizures, and developmental delay (OMIM:613402); Early infantile developmental and epileptic encephalopathy (ORPHA:1934); Ataxia-oculomotor apraxia type 4 (ORPHA:459033); ORPHA:101101
PNLIP	Pancreatic lipase deficiency (OMIM:614338)
PNMT	—
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency (OMIM:613179); Purine nucleoside phosphorylase deficiency (ORPHA:760)
PNPLA1	Ichthyosis, congenital, autosomal recessive 10 (OMIM:615024); Congenital ichthyosiform erythroderma (ORPHA:79394)
PNPLA2	Neutral lipid storage disease with myopathy (OMIM:610717); Primary triglyceride deposit cardiomyovasculopathy (ORPHA:565612); Neutral lipid storage disease with myopathy (ORPHA:98908)
PNPLA3	—
PNPLA6	Laurence-Moon syndrome (OMIM:245800); Spastic paraplegia 39, autosomal recessive (OMIM:612020); Boucher-Neuhauser syndrome (OMIM:215470); Oliver-Mcfarlane syndrome (OMIM:275400); Trichomegaly-retina pigmentary degeneration-dwarfism syndrome (ORPHA:3363); Autosomal recessive spastic paraplegia type 39 (ORPHA:139480); Laurence-Moon syndrome (ORPHA:2377); Ataxia-hypogonadism-choroidal dystrophy syndrome (ORPHA:1180); Cerebellar ataxia-hypogonadism syndrome (ORPHA:1173)
PNPO	Pyridoxamine 5-prime-phosphate oxidase deficiency (OMIM:610090); Pyridoxamine-5-phosphate deficiency-developmental and epileptic encephalopathy (ORPHA:79096)
PNPT1	Spinocerebellar ataxia 25 (OMIM:608703); Combined oxidative phosphorylation deficiency 13 (OMIM:614932); Deafness, autosomal recessive 70 (OMIM:614934); ORPHA:90636; Combined oxidative phosphorylation defect type 13 (ORPHA:319514); Spinocerebellar ataxia type 25 (ORPHA:101111)
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis (OMIM:614813); ORPHA:314394
POF1B	Premature ovarian failure 2B (OMIM:300604)
POFUT2	—
POGZ	White-Sutton syndrome (OMIM:616364); White-Sutton syndrome (ORPHA:468678)
POLB	—
POLD1	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome (OMIM:615381); Colorectal cancer, susceptibility to, 10 (OMIM:612591); Immunodeficiency 120 (OMIM:620836); ORPHA:363649; Familial colorectal cancer Type X (ORPHA:440437); Polymerase proofreading-related polyposis (ORPHA:447877)
POLE2	—
POLG	Mitochondrial DNA depletion syndrome 1 (MNGIE type) (OMIM:603041); Progressive external ophthalmoplegia with mitochondrial DNA deletions,autosomal recessive (OMIM:258450); Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis (OMIM:607459); Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 1 (OMIM:157640); Mitochondrial DNA depletion syndrome 4A (Alpers type) (OMIM:203700); Mitochondrial DNA depletion syndrome 4B (mngie type) (OMIM:613662); Recessive mitochondrial ataxia syndrome (ORPHA:94125); Mitochondrial neurogastrointestinal encephalomyopathy (ORPHA:298); Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome (ORPHA:70595); Alpers-Huttenlocher syndrome (ORPHA:726); ORPHA:402082; Autosomal dominant progressive external ophthalmoplegia (ORPHA:254892); Spinocerebellar ataxia with epilepsy (ORPHA:254881); Autosomal recessive progressive external ophthalmoplegia (ORPHA:254886)
POLG2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4 (OMIM:610131); Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type) (OMIM:619425); Mitochondrial DNA depletion syndrome 16 (hepatic type) (OMIM:618528); Autosomal dominant progressive external ophthalmoplegia (ORPHA:254892)
POLGARF	—
POLH	Xeroderma pigmentosum, variant type (OMIM:278750); Xeroderma pigmentosum variant (ORPHA:90342)
POLL	—
POLR1C	Leukodystrophy, hypomyelinating, 11 (OMIM:616494); Treacher Collins syndrome 3 (OMIM:248390); Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome (ORPHA:88637); Treacher-Collins syndrome (ORPHA:861)
POLR1D	Treacher collins syndrome 2 (OMIM:613717); Treacher-Collins syndrome (ORPHA:861)
POLR1G	—
POLR1HASP	—
POLR2F	—
POLR2L	—
POLR3A	Wiedemann-Rautenstrauch syndrome (OMIM:264090); Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/orhypogonadotropic hypogonadism (OMIM:607694); ORPHA:137639; Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome (ORPHA:88637); ORPHA:77295; Wiedemann-Rautenstrauch syndrome (ORPHA:3455); ORPHA:447893; Tremor-ataxia-central hypomyelination syndrome (ORPHA:447896)
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism (OMIM:614381); Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/orhypogonadotropic

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	hypogonadism (OMIM:607694); Charcot-Marie-Tooth disease, demyelinating, type 1I (OMIM:619742); Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome (ORPHA:88637); ORPHA:85186
POLR3H	46,XX gonadal dysgenesis (ORPHA:243)
POLRMT	Combined oxidative phosphorylation deficiency 55 (OMIM:619743)
POMC	Obesity, early-onset, with adrenal insufficiency and red hair (OMIM:609734); OBESITY (OMIM:601665); Obesity due to pro-opiomelanocortin deficiency (ORPHA:71526)
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3 (OMIM:253280); Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3 (OMIM:613151); Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3 (OMIM:613157); Retinitis pigmentosa 76 (OMIM:617123); Congenital muscular dystrophy with cerebellar involvement (ORPHA:370959); Walker-Warburg syndrome (ORPHA:899); Retinitis pigmentosa (ORPHA:791); ORPHA:206564; Muscle-eye-brain disease (ORPHA:588)
POMGNT2	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 8 (OMIM:618135); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8 (OMIM:614830); Walker-Warburg syndrome (ORPHA:899)
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12 (OMIM:615249); Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12 (OMIM:616094); Congenital muscular dystrophy with cerebellar involvement (ORPHA:370959); Walker-Warburg syndrome (ORPHA:899); ORPHA:445110
POMP	Proteasome-Associated autoinflammatory syndrome 2 (OMIM:618048); Keratosis linearis with ichthyosis congenita and sclerosing keratoderma (OMIM:601952); Keratosis linearis-ichthyosis congenita-sclerosing keratoderma syndrome (ORPHA:281201)
POMT1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1 (OMIM:609308); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1 (OMIM:236670); Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1 (OMIM:613155); Congenital muscular dystrophy with intellectual disability (ORPHA:370968); Congenital muscular dystrophy without intellectual disability (ORPHA:370980); Congenital muscular dystrophy with cerebellar involvement (ORPHA:370959); Walker-Warburg syndrome (ORPHA:899); POMT1-related limb-girdle muscular dystrophy R11 (ORPHA:86812); Muscle-eye-brain disease (ORPHA:588)
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1 (OMIM:236670); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2 (OMIM:613150); Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2 (OMIM:613156); Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2 (OMIM:613158); Congenital muscular dystrophy with intellectual disability (ORPHA:370968); Congenital muscular dystrophy with cerebellar involvement (ORPHA:370959); Walker-Warburg syndrome (ORPHA:899); POMT2-related limb-girdle muscular dystrophy R14 (ORPHA:206559); Muscle-eye-brain disease (ORPHA:588)
PON1	Amyotrophic lateral sclerosis (ORPHA:803)
PON2	Amyotrophic lateral sclerosis (ORPHA:803)
PON3	Amyotrophic lateral sclerosis (ORPHA:803)
POP1	Anauxetic dysplasia 2 (OMIM:617396); ORPHA:93347
POR	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis (OMIM:207410); Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis (OMIM:201750); Disordered steroidogenesis due to cytochrome P450 oxidoreductase (OMIM:613571); ORPHA:63269; Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency (ORPHA:95699)
PORCN	Focal dermal hypoplasia (OMIM:305600); ORPHA:98938; Focal dermal hypoplasia (ORPHA:2092)
POSTN	—
POT1	Melanoma, cutaneous malignant, susceptibility to, 10 (OMIM:615848); Cerebroretinal microangiopathy with calcifications and cysts 3 (OMIM:620368); Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8 (OMIM:620367); Familial melanoma (ORPHA:618); ORPHA:67038; ORPHA:251627; ORPHA:251630
POU1F1	Pituitary hormone deficiency, combined, 1 (OMIM:613038); Combined pituitary hormone deficiencies, genetic forms (ORPHA:95494); Hypothyroidism due to deficient transcription factors involved in pituitary development or function (ORPHA:226307); ORPHA:231679
POU3F4	Deafness, X-linked 2 (OMIM:304400); ORPHA:90641; Xq21 microdeletion syndrome (ORPHA:1435)
POU4F3	Deafness, autosomal dominant 15 (OMIM:602459); ORPHA:90635
POU5F1	—
POU6F2	Wilms tumor 5 (OMIM:601583); Nephroblastoma (ORPHA:654)
PPARA	—
PPARG	Diabetes mellitus, noninsulin-dependent (OMIM:125853); Lipodystrophy, familial partial, type 3 (OMIM:604367); OBESITY (OMIM:601665); ORPHA:251579; ORPHA:251576; ORPHA:146; PPARG-related familial partial lipodystrophy (ORPHA:79083); ORPHA:696242
PPARGC1B	—
PPDPF	—
PPIB	Osteogenesis imperfecta, type IX (OMIM:259440); ORPHA:216812; ORPHA:216804; ORPHA:216820
PPM1B	2p21 microdeletion syndrome (ORPHA:163693)
PPM1D	Breast cancer (OMIM:114480); Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold (OMIM:617450); ORPHA:653767
PPM1E	—
PPM1G	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
PPM1K	Maple syrup urine disease, mild variant (OMIM:615135); ORPHA:268162
PPM1N	—
PPOX	Porphyria variegata (OMIM:176200); Variegate porphyria, childhood-onset (OMIM:620483); Variegate porphyria (ORPHA:79473)
PPP1R12B	—
PPP1R17	Hypercholesterolemia, familial, 1 (OMIM:143890)
PPP1R1A	—
PPP1R3A	Diabetes mellitus, noninsulin-dependent (OMIM:125853)
PPP1R3C	—
PPP2R1B	Lung cancer, susceptibility to (OMIM:211980)
PPP2R2A	—
PPP2R2B	Spinocerebellar ataxia 12 (OMIM:604326); Spinocerebellar ataxia type 12 (ORPHA:98762)
PPP2R2C	—
PPP3CA	Developmental and epileptic encephalopathy 91 (OMIM:617711); Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development (OMIM:618265); ORPHA:178469; Non-specific early-onset epileptic encephalopathy (ORPHA:442835); ORPHA:565858
PPP3R1	—
PPT1	Ceroid lipofuscinosis, neuronal, 1 (OMIM:256730); ORPHA:699745; ORPHA:699739; ORPHA:699734; ORPHA:699718
PQBP1	Renpenning syndrome (OMIM:309500); X-linked intellectual disability, Sutherland-Haan type (ORPHA:93950); X-linked intellectual disability, Porteous type (ORPHA:93945); Hamel cerebro-palato-cardiac syndrome (ORPHA:93946); X-linked intellectual disability, Golabi-Ito-Hall type (ORPHA:93947)
PRAF2	—
PRB1	—
PRB3	—
PRB4	—
PRCC	Renal cell carcinoma, papillary, 1, familial and somatic (OMIM:605074); ORPHA:319308
PRCD	Retinitis pigmentosa 36 (OMIM:610599); Retinitis pigmentosa (ORPHA:791)
PRCP	—
PRDM16	Left ventricular noncompaction 8 (OMIM:615373); ORPHA:54260; Familial isolated dilated cardiomyopathy (ORPHA:154); 1p36 deletion syndrome (ORPHA:1606)
PRDM2	—
PRDM5	Brittle cornea syndrome 2 (OMIM:614170); Brittle cornea syndrome (ORPHA:90354)
PRDM9	—
PRELID2	—
PREPL	Myasthenic syndrome, congenital, 22 (OMIM:616224); ORPHA:238523; Hypotonia-cystinuria syndrome (ORPHA:163690); 2p21 microdeletion syndrome (ORPHA:163693); ORPHA:369881
PRF1	Aplastic anemia (OMIM:609135); Hemophagocytic lymphohistiocytosis, familial, 2 (OMIM:603553); Lymphoma, non-hodgkin, familial (OMIM:605027); Idiopathic aplastic anemia (ORPHA:88); ORPHA:391343; Familial hemophagocytic lymphohistiocytosis (ORPHA:540)
PRG4	Camptodactyly-Arthropathy-Coxa vara-pericarditis syndrome (OMIM:208250); Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome (ORPHA:2848)
PRH1	—
PRICKLE1	Epilepsy, progressive myoclonic 1B (OMIM:612437); Progressive myoclonic epilepsy type 1 (ORPHA:308)
PRICKLE2	ORPHA:178469; ORPHA:402082
PRKACA	Cardioacrofacial dysplasia 1 (OMIM:619142); Pigmented nodular adrenocortical disease, primary, 4 (OMIM:615830); Ellis Van Creveld syndrome (ORPHA:289); ORPHA:401920
PRKAG2	Cardiomyopathy, familial hypertrophic, 6 (OMIM:600858); Wolff-Parkinson-White syndrome (OMIM:194200); Glycogen storage disease of heart, lethal congenital (OMIM:261740); ORPHA:439854
PRKAG3	OMIM:619030
PRKAR1A	Acrodysostosis 1, with or without hormone resistance (OMIM:101800); Carney complex, type 1 (OMIM:160980); Pigmented nodular adrenocortical disease, primary, 1 (OMIM:610489); Myxoma, intracardiac (OMIM:255960); Adrenocortical carcinoma (ORPHA:1501); ORPHA:647772; Acrodysostosis (ORPHA:950); Carney complex (ORPHA:1359); Familial atrial myxoma (ORPHA:615); Acute promyelocytic leukemia (ORPHA:520)
PRKAR1B	Marbach-Schaaf neurodevelopmental syndrome (OMIM:619680); ORPHA:692173; PRKAR1B-related neurodegenerative dementia with intermediate filaments (ORPHA:412066)
PRKAR2A	—
PRKCA	—
PRKCB	—
PRKCG	Spinocerebellar ataxia 14 (OMIM:605361); Spinocerebellar ataxia type 14 (ORPHA:98763)
PRKCH	—
PRKCSH	Polycystic liver disease 1 with or without kidney cysts (OMIM:174050); Isolated polycystic liver disease (ORPHA:2924)
PRKDC	Immunodeficiency 26 with or without neurologic abnormalities (OMIM:615966); ORPHA:317425
PRKG1	Aortic aneurysm, familial thoracic 8 (OMIM:615436); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387)
PRKG2	Spondylometaphyseal dysplasia, Pagnamenta type (OMIM:619638); Acromesomelic dysplasia 4 (OMIM:619636)

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PRKN	Parkinson disease, juvenile, type 2 (OMIM:600116); Ovarian cancer (OMIM:167000); Lung cancer, susceptibility to (OMIM:211980); Young-onset Parkinson disease (ORPHA:2828)
PRLH	—
PRLR	Hyperprolactinemia (OMIM:615555); Multiple fibroadenomas of the breast (OMIM:615554); Familial hyperprolactinemia (ORPHA:397685)
PRM1	—
PRM2	—
PRMT3	—
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures (OMIM:617157); Short stature-brachydactyly-obesity-global developmental delay syndrome (ORPHA:464288)
PRMT9	—
PRND	—
PRNP	Huntington disease-like 1 (OMIM:603218); Creutzfeldt-Jakob disease (OMIM:123400); Fatal familial insomnia (OMIM:600072); KURU, SUSCEPTIBILITY TO (OMIM:245300); Spongiform encephalopathy with neuropsychiatric features (OMIM:606688); Gerstmann-Straussler disease (OMIM:137440); Gerstmann-Straussler-Scheinker syndrome (ORPHA:356); Familial Alzheimer-like prion disease (ORPHA:280397); Inherited Creutzfeldt-Jakob disease (ORPHA:282166); ORPHA:454745; Huntington disease-like 1 (ORPHA:157941); ORPHA:397606; Fatal familial insomnia (ORPHA:466)
PROC	Thrombophilia, hereditary, due to protein C deficiency, autosomaldominant (OMIM:176860); Thrombophilia due to protein C deficiency, autosomal recessive (OMIM:612304); Severe hereditary thrombophilia due to congenital protein C deficiency (ORPHA:745)
PROCR	—
PRODH	Schizophrenia 4 (OMIM:600850); Hyperprolinemia, type I (OMIM:239500); Hyperprolinemia type 1 (ORPHA:419)
PROK1	—
PROK2	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Hypogonadotropic hypogonadism 4 with or without anosmia (OMIM:610628); Kallmann syndrome (ORPHA:478); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432)
PROKR1	—
PROKR2	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Hypogonadotropic hypogonadism 3 with or without anosmia (OMIM:244200); Septo-optic dysplasia spectrum (ORPHA:3157); Pituitary stalk interruption syndrome (ORPHA:95496); Kallmann syndrome (ORPHA:478); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432)
PROM1	Macular dystrophy, retinal, 2 (OMIM:608051); Cone-rod dystrophy 12 (OMIM:612657); Retinitis pigmentosa 41 (OMIM:612095); Stargardt disease 4 (OMIM:603786); Cone rod dystrophy (ORPHA:1872); Retinitis pigmentosa (ORPHA:791); Stargardt disease (ORPHA:827); ORPHA:319640
PROP1	Pituitary hormone deficiency, combined, 2 (OMIM:262600); Non-acquired panhypopituitarism (ORPHA:90695); Combined pituitary hormone deficiencies, genetic forms (ORPHA:95494); Hypothyroidism due to deficient transcription factors involved in pituitary development or function (ORPHA:226307)
PROS1	Thrombophilia due to protein S deficiency, autosomal recessive (OMIM:614514); Thrombophilia, hereditary, due to protein S deficiency, autosomal (OMIM:612336); Severe hereditary thrombophilia due to congenital protein S deficiency (ORPHA:743)
PROX1	—
PROZ	OMIM:614024; ORPHA:329217
PRPF3	Retinitis pigmentosa 18 (OMIM:601414); Retinitis pigmentosa (ORPHA:791)
PRPF31	Retinitis pigmentosa 11 (OMIM:600138); Retinitis pigmentosa (ORPHA:791)
PRPF6	Retinitis pigmentosa 60 (OMIM:613983); Retinitis pigmentosa (ORPHA:791)
PRPF8	Retinitis pigmentosa 13 (OMIM:600059); Retinitis pigmentosa (ORPHA:791); ORPHA:528084
PRPH	Amyotrophic lateral sclerosis 1 (OMIM:105400); Amyotrophic lateral sclerosis (ORPHA:803)
PRPH2	Retinitis pigmentosa 7 (OMIM:608133); Fundus albipunctatus (OMIM:136880); Macular dystrophy, patterned, 1 (OMIM:169150); Macular dystrophy, vitelliform, 3 (OMIM:608161); Choroidal dystrophy, central areolar 2 (OMIM:613105); ORPHA:99001; ORPHA:99003; Adult-onset foveomacular vitelliform dystrophy (ORPHA:99000); ORPHA:227796; Retinitis punctata albescens (ORPHA:52427); Central areolar choroidal dystrophy (ORPHA:75377); Cone rod dystrophy (ORPHA:1872); Retinitis pigmentosa (ORPHA:791); Stargardt disease (ORPHA:827)
PRPS1	Deafness, X-linked 1 (OMIM:304500); Charcot-Marie-Tooth disease, X-linked recessive, 5 (OMIM:311070); Arts syndrome (OMIM:301835); Phosphoribosylpyrophosphate synthetase superactivity (OMIM:300661); X-linked Charcot-Marie-Tooth disease type 5 (ORPHA:99014); ORPHA:90625; X-linked intellectual disability-limb spasticity-retinal dystrophy-arginine vasopressin deficiency (ORPHA:423479); Severe phosphoribosylpyrophosphate synthetase superactivity (ORPHA:411543); Mild phosphoribosylpyrophosphate synthetase superactivity (ORPHA:411536); Lethal ataxia with deafness and optic atrophy (ORPHA:1187)
PRR19	—
PRRT2	Episodic kinesigenic dyskinesia 1 (OMIM:128200); Seizures, benign familial infantile, 2 (OMIM:605751); Convulsions, familial infantile, with paroxysmal choreoathetosis (OMIM:602066); Self-limited infantile epilepsy (ORPHA:306); Genetic epilepsy with febrile seizure plus (ORPHA:36387); Paroxysmal exertion-induced dyskinesia (ORPHA:98811); Paroxysmal non-kinesigenic dyskinesia (ORPHA:98810); Paroxysmal kinesigenic dyskinesia (ORPHA:98809); Familial or sporadic hemiplegic migraine (ORPHA:569)
PRRX1	Agnathia-Otocephaly complex (OMIM:202650); Agnathia-holoprosencephaly-situs inversus syndrome (ORPHA:990)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
PRSS1	Pancreatitis, hereditary (OMIM:167800); Autosomal dominant hereditary chronic pancreatitis (ORPHA:676)
PRSS12	Intellectual developmental disorder, autosomal recessive 1 (OMIM:249500); ORPHA:88616
PRSS23	—
PRSS56	Microphthalmia, isolated 6 (OMIM:613517); Nanophthalmos (ORPHA:35612)
PRSS8	—
PRTG	—
PRX	Hypertrophic neuropathy of dejerine-sottas (OMIM:145900); Charcot-Marie-Tooth disease, demyelinating, type 4F (OMIM:614895); ORPHA:99952; ORPHA:64748
PRXL2B	—
PSAP	Combined saposin deficiency (OMIM:611721); Krabbe disease, atypical, due to saposin A deficiency (OMIM:611722); Metachromatic leukodystrophy due to saposin B deficiency (OMIM:249900); Gaucher disease, atypical (OMIM:610539); Parkinson disease 24, autosomal dominant, susceptibility to (OMIM:619491); Encephalopathy due to prosaposin deficiency (ORPHA:139406); Infantile Krabbe disease (ORPHA:206436); Metachromatic leukodystrophy, adult form (ORPHA:309271); Metachromatic leukodystrophy, juvenile form (ORPHA:309263); Atypical Gaucher disease due to saposin C deficiency (ORPHA:309252); Metachromatic leukodystrophy, late infantile form (ORPHA:309256)
PSAT1	Phosphoserine aminotransferase deficiency (OMIM:610992); Neu-Laxova syndrome 2 (OMIM:616038); Phosphoserine aminotransferase deficiency, infantile/juvenile form (ORPHA:284417); ORPHA:583602
PSEN1	Pick disease of brain (OMIM:172700); Acne inversa, familial, 3 (OMIM:613737); Alzheimer disease 3 (OMIM:607822); Cardiomyopathy, dilated, 1U (OMIM:613694); Frontotemporal dementia (OMIM:600274); Familial isolated dilated cardiomyopathy (ORPHA:154); Behavioral variant of frontotemporal dementia (ORPHA:275864); Early-onset autosomal dominant Alzheimer disease (ORPHA:1020); Progressive non-fluent aphasia (ORPHA:100070); Semantic dementia (ORPHA:100069)
PSEN2	Alzheimer disease 4 (OMIM:606889); Cardiomyopathy, dilated, 1V (OMIM:613697); Familial isolated dilated cardiomyopathy (ORPHA:154); Early-onset autosomal dominant Alzheimer disease (ORPHA:1020)
PSENEN	Acne inversa, familial, 2, with or without dowling-degos disease (OMIM:613736); Dowling-Degos disease (ORPHA:79145)
PSMA4	—
PSMA6	OMIM:608446
PSMB5	—
PSMB8	Proteasome-associated autoinflammatory syndrome 1 and digenic forms (OMIM:256040); ORPHA:324977
PSMB9	Proteasome-associated autoinflammatory syndrome 6 (OMIM:620796)
PSMC2	—
PSMC3IP	Ovarian dysgenesis 3 (OMIM:614324); 46,XX gonadal dysgenesis (ORPHA:243)
PSMD1	—
PSMD7	—
PSPH	Phosphoserine phosphatase deficiency (OMIM:614023); 3-phosphoserine phosphatase deficiency, infantile/juvenile form (ORPHA:79350); ORPHA:583612
PSPN	—
PSTPIP1	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne (OMIM:604416); Hyperzincemia with functional zinc depletion (OMIM:601979); ORPHA:251523; PAPA syndrome (ORPHA:69126)
PSTPIP2	—
PTAFR	—
PTCD1	—
PTCH1	Holoprosencephaly 7 (OMIM:610828); Basal cell nevus syndrome (OMIM:109400); Basal cell carcinoma, susceptibility to, 1 (OMIM:605462); Gorlin syndrome (ORPHA:377); Septopreoptic holoprosencephaly (ORPHA:280195); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925); ORPHA:2353; Monosomy 9q22.3 syndrome (ORPHA:77301)
PTCH2	Basal cell carcinoma, susceptibility to, 1 (OMIM:605462); MEDULLOBLASTOMA (OMIM:155255); Gorlin syndrome (ORPHA:377); ORPHA:141276
PTCHD1	Autism, susceptibility to, X-linked 4 (OMIM:300830); ORPHA:777
PTCHD3	—
PTEN	Meningioma, familial, susceptibility to (OMIM:607174); Glioma susceptibility 2 (OMIM:613028); Cowden syndrome 1 (OMIM:158350); Prostate cancer (OMIM:176807); Macrocephaly/autism syndrome (OMIM:605309); Cowden syndrome (ORPHA:201); Hereditary breast and/or ovarian cancer syndrome (ORPHA:145); Bannayan-Riley-Ruvalcaba syndrome (ORPHA:109); ORPHA:494547; ORPHA:494550; Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome (ORPHA:137608); Lhermitte-Duclos disease (ORPHA:65285); Macrocephaly-intellectual disability-autism syndrome (ORPHA:210548); Juvenile polyposis of infancy (ORPHA:79076); Proteus-like syndrome (ORPHA:2969); ORPHA:502363; ORPHA:502366; ORPHA:500478; ORPHA:500464
PTF1A	Pancreatic and cerebellar agenesis (OMIM:609069); Pancreatic agenesis 2 (OMIM:615935); ORPHA:2805; Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome (ORPHA:65288)
PTGDR	OMIM:607277
PTGER2	Asthma, nasal polyps, and aspirin intolerance (OMIM:208550)
PTGER4	—
PTGIR	—
PTGIS	Hypertension, essential (OMIM:145500)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
PTGS1	—
PTGS2	—
PTH	Hypoparathyroidism, familial isolated (OMIM:146200); ORPHA:189466
PTH1R	Chondrodysplasia, Blomstrand type (OMIM:215045); Eiken syndrome (OMIM:600002); Metaphyseal chondrodysplasia, Jansen type (OMIM:156400); Dental noneruption (OMIM:125350); Ollier disease (ORPHA:296); Blomstrand lethal chondrodysplasia (ORPHA:50945); ORPHA:412206; Eiken syndrome (ORPHA:79106); Metaphyseal chondrodysplasia, Jansen type (ORPHA:33067)
PTHLH	Brachydactyly, type E2 (OMIM:613382); Brachydactyly type E (ORPHA:93387)
PTK7	—
PTPN1	Diabetes mellitus, noninsulin-dependent (OMIM:125853)
PTPN11	Leopard syndrome 1 (OMIM:151100); Metachondromatosis (OMIM:156250); Juvenile myelomonocytic leukemia (OMIM:607785); Noonan syndrome 1 (OMIM:163950); ORPHA:86834; Noonan syndrome (ORPHA:648); Metachondromatosis (ORPHA:2499); Noonan syndrome with multiple lentigines (ORPHA:500)
PTPN12	Colorectal cancer (OMIM:114500)
PTPN14	Choanal atresia and lymphedema (OMIM:613611); ORPHA:99141
PTPN2	Oligoarticular juvenile idiopathic arthritis (ORPHA:85410); Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis (ORPHA:85408)
PTPN22	Giant cell arteritis (ORPHA:397); Oligoarticular juvenile idiopathic arthritis (ORPHA:85410); Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis (ORPHA:85408); Granulomatosis with polyangiitis (ORPHA:900); Systemic lupus erythematosus (ORPHA:536); Vogt-Koyanagi-Harada disease (ORPHA:3437)
PTPN6	Sweet syndrome (ORPHA:3243); ORPHA:538869; ORPHA:538866; ORPHA:538863; ORPHA:538872
PTPRC	Immunodeficiency 105, severe combined (OMIM:619924); ORPHA:169157
PTPRJ	Thrombocytopenia 10 (OMIM:620484); Colorectal cancer (OMIM:114500)
PTPRK	—
PTPRN2	—
PTPRO	Nephrotic syndrome, type 6 (OMIM:614196); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
PTPRQ	Deafness, autosomal dominant 73 (OMIM:617663); Deafness, autosomal recessive 84 (OMIM:613391); ORPHA:90635; ORPHA:90636
PTPRT	—
PTRH1	—
PTS	Hyperphenylalaninemia, BH4-deficient, A (OMIM:261640); 6-pyruvoyl-tetrahydropterin synthase deficiency (ORPHA:13)
PUM3	—
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1 (OMIM:600462); Mitochondrial myopathy and sideroblastic anemia (ORPHA:2598)
PUS10	—
PUS3	Neurodevelopmental disorder with microcephaly and gray sclerae (OMIM:617051); Severe growth deficiency-strabismus-extensive dermal melanocytosis-intellectual disability syndrome (ORPHA:488627)
PXDN	Corneal opacification with other ocular anomalies (OMIM:269400); ORPHA:289499; ORPHA:699850
PYCR1	Cutis laxa, autosomal recessive, type IIB (OMIM:612940); Cutis laxa, autosomal recessive, type IIIB (OMIM:614438); ORPHA:293633; Geroderma osteodysplastica (ORPHA:2078); ORPHA:357064
PYGL	Glycogen storage disease VI (OMIM:232700); Glycogen storage disease due to liver glycogen phosphorylase deficiency (ORPHA:369)
PYGM	Glycogen storage disease V (OMIM:232600); Glycogen storage disease due to muscle glycogen phosphorylase deficiency (ORPHA:368)
PYY	—
PZP	—
QDPR	Hyperphenylalaninemia, bh4-deficient, C (OMIM:261630); Dihydropteridine reductase deficiency (ORPHA:226)
QKI	ORPHA:251671
QKILA	—
RAB11B	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter (OMIM:617807)
RAB11FIP5	—
RAB18	Warburg micro syndrome 3 (OMIM:614222); Micro syndrome (ORPHA:2510)
RAB23	Carpenter syndrome 1 (OMIM:201000); Carpenter syndrome (ORPHA:65759)
RAB25	—
RAB27A	Griscelli syndrome, type 2 (OMIM:607624); Griscelli syndrome type 2 (ORPHA:79477)
RAB27B	—
RAB29	—
RAB2A	—
RAB39B	Intellectual developmental disorder, X-linked 72 (OMIM:300271); Waisman syndrome (OMIM:311510); OMIM:300815; ORPHA:777; Early-onset parkinsonism-intellectual disability syndrome (ORPHA:2379)
RAB3GAP1	Warburg micro syndrome 1 (OMIM:600118); Martsolf syndrome 2 (OMIM:619420); Micro syndrome (ORPHA:2510); Cataract-intellectual disability-hypogonadism syndrome (ORPHA:1387)
RAB3GAP2	Warburg micro syndrome 2 (OMIM:614225); Martsolf syndrome 1 (OMIM:212720); Autosomal recessive spastic paraplegia type 69 (ORPHA:401830); Micro syndrome (ORPHA:2510); Cataract-intellectual disability-hypogonadism syndrome (ORPHA:1387)
RAB7A	Charcot-Marie-Tooth disease, axonal, type 2B (OMIM:600882); ORPHA:99936
RABEP2	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
RABGGTA	—
RABL6	—
RAC1	Intellectual developmental disorder, autosomal dominant 48 (OMIM:617751); Microcephaly-corpus callosum and cerebellar vermis hypoplasia-facial dysmorphism-intellectual disability syndrom (ORPHA:500159)
RAC2	Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia (OMIM:618986); Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia (OMIM:618987); Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis (OMIM:608203); Infantile LAD-like disease due to RAC2 deficiency (ORPHA:183707); ORPHA:688543; ORPHA:692812
RAD21	Mungan syndrome (OMIM:611376); Cornelia de Lange syndrome 4 (OMIM:614701); Cornelia de Lange syndrome (ORPHA:199); Trichorhinophalangeal syndrome type 2 (ORPHA:502)
RAD21L1	—
RAD50	Nijmegen breakage syndrome-like disorder (OMIM:613078); ORPHA:240760; Hereditary breast and/or ovarian cancer syndrome (ORPHA:145)
RAD51	Breast cancer (OMIM:114480); Mirror movements 2 (OMIM:614508); Mirror movements 1 and/or agenesis of the corpus callosum (OMIM:157600); Fanconi anemia, complementation group R (OMIM:617244); Fanconi anemia (ORPHA:84); Hereditary breast and/or ovarian cancer syndrome (ORPHA:145); Familial congenital mirror movements (ORPHA:238722)
RAD51B	—
RAD51C	Fanconi anemia, complementation group O (OMIM:613390); Familial breast-ovarian cancer-1 (OMIM:613399); Fanconi anemia (ORPHA:84); Hereditary breast and/or ovarian cancer syndrome (ORPHA:145)
RAD51D	OMIM:614291; Hereditary breast and/or ovarian cancer syndrome (ORPHA:145); ORPHA:1331
RAD52	—
RAD54B	Lymphoma, non-hodgkin, familial (OMIM:605027); Colorectal cancer (OMIM:114500)
RAD54L	Lymphoma, non-hodgkin, familial (OMIM:605027); Breast cancer (OMIM:114480); ORPHA:227535
RAD9A	—
RAF1	Leopard syndrome 2 (OMIM:611554); Noonan syndrome 5 (OMIM:611553); Cardiomyopathy, dilated, 1nn (OMIM:615916); Familial isolated dilated cardiomyopathy (ORPHA:154); Large/giant congenital melanocytic nevus (ORPHA:626); Noonan syndrome (ORPHA:648); Noonan syndrome with multiple lentigines (ORPHA:500); ORPHA:251615
RAG1	Combined cellular and humoral immune defects with granulomas (OMIM:233650); Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity (OMIM:609889); Omenn syndrome (OMIM:603554); Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-positive (OMIM:601457); ORPHA:157949; Omenn syndrome (ORPHA:39041); Severe combined immunodeficiency due to complete RAG1/2 deficiency (ORPHA:331206); Combined immunodeficiency due to partial RAG1 deficiency (ORPHA:231154)
RAG2	Combined cellular and humoral immune defects with granulomas (OMIM:233650); Omenn syndrome (OMIM:603554); Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-positive (OMIM:601457); ORPHA:157949; Omenn syndrome (ORPHA:39041); Severe combined immunodeficiency due to complete RAG1/2 deficiency (ORPHA:331206)
RAI1	Potocki-Lupski syndrome (OMIM:610883); Smith-Magenis syndrome (OMIM:182290); PMP22-RAI1 contiguous gene duplication syndrome (ORPHA:477817); Smith-Magenis syndrome (ORPHA:819); 17p11.2 microduplication syndrome (ORPHA:1713)
RALGAPA1	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation (OMIM:618797)
RALGDS	—
RANBP2	Encephalopathy, acute, infection-induced, susceptibility to, 3 (OMIM:608033); ORPHA:263524; ORPHA:178342; Familial acute necrotizing encephalopathy (ORPHA:88619)
RANBP3	—
RANGRF	Brugada syndrome (ORPHA:130)
RAP1GDS1	Alfadhel syndrome (OMIM:620655)
RAPSN	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency (OMIM:616326); Fetal akinesia deformation sequence 2 (OMIM:618388); Fetal akinesia deformation sequence (ORPHA:994); Postsynaptic congenital myasthenic syndrome (ORPHA:98913); ORPHA:33108
RARA	Acute promyelocytic leukemia (OMIM:612376); Acute promyelocytic leukemia (ORPHA:520)
RARS2	Pontocerebellar hypoplasia, type 6 (OMIM:611523); ORPHA:166073
RASA1	Capillary malformation-arteriovenous malformation 1 (OMIM:608354); Basal cell carcinoma, susceptibility to, 1 (OMIM:605462); ORPHA:693907; Parkes Weber syndrome (ORPHA:90307)
RASGEF1B	—
RASGRP1	Immunodeficiency 64 (OMIM:618534); ORPHA:664699
RASGRP2	Bleeding disorder, platelet-type, 18 (OMIM:615888); ORPHA:420566
RASSF5	—
RAX	Microphthalmia, isolated 3 (OMIM:611038); ORPHA:98938; Nanophthalmos (ORPHA:35612)
RAX2	Cone-Rod dystrophy 11 (OMIM:610381); Retinitis pigmentosa 95 (OMIM:620102); Macular degeneration, age-related, 6 (OMIM:613757); Cone rod dystrophy (ORPHA:1872)
RB1	Small cell cancer of the lung (OMIM:182280); Osteosarcoma (OMIM:259500); Bladder cancer (OMIM:109800); RETINOBLASTOMA (OMIM:180200); Monosomy 13q14 syndrome (ORPHA:1587); ORPHA:70573; Osteosarcoma (ORPHA:668); ORPHA:357034; ORPHA:357027
RB1CC1	Breast cancer (OMIM:114480)
RBBP6	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
RBBP8	Seckel syndrome 2 (OMIM:606744); Jawad syndrome (OMIM:251255); ORPHA:313795; Seckel syndrome (ORPHA:808)
RBFOX1	—
RBL1	—
RBL2	Brunet-Wagner neurodevelopmental syndrome (OMIM:619690)
RBM10	TARP syndrome (OMIM:311900); TARP syndrome (ORPHA:2886)
RBM15	ORPHA:402023
RBM20	Cardiomyopathy, dilated, 1DD (OMIM:613172); Familial isolated dilated cardiomyopathy (ORPHA:154)
RBM28	Alopecia, neurologic defects, and endocrinopathy syndrome (OMIM:612079); ANE syndrome (ORPHA:157954)
RBMXL2	—
RBP3	Retinitis pigmentosa 66 (OMIM:615233); Retinitis pigmentosa (OMIM:268000); Retinitis pigmentosa (ORPHA:791)
RBP4	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome (OMIM:615147); Microphthalmia, isolated, with coloboma 10 (OMIM:616428); ORPHA:98938; ORPHA:352718
RBPJ	Adams-Oliver syndrome 3 (OMIM:614814); Adams-Oliver syndrome (ORPHA:974)
RC3H1	Immune dysregulation and systemic hyperinflammation syndrome (OMIM:618998)
RCBTB1	Retinal dystrophy with or without extraocular anomalies (OMIM:617175); ORPHA:99002
RCC2P5	—
RCHY1	—
RD3	Leber congenital amaurosis 12 (OMIM:610612); Leber congenital amaurosis (ORPHA:65)
RDH12	Leber congenital amaurosis 13 (OMIM:612712); Leber congenital amaurosis (ORPHA:65); Retinitis pigmentosa (ORPHA:791)
RDH5	Fundus albipunctatus (OMIM:136880); ORPHA:227796; Retinitis punctata albescens (ORPHA:52427)
RDH8	Stargardt disease 5 (OMIM:621259)
RDX	Deafness, autosomal recessive, 24 (OMIM:611022); ORPHA:90636
RECQL4	Rothmund-Thomson syndrome (OMIM:268400); Baller-Gerold syndrome (OMIM:218600); RAPADILINO syndrome (OMIM:266280); RAPADILINO syndrome (ORPHA:3021); Baller-Gerold syndrome (ORPHA:1225); Rothmund-Thomson syndrome type 2 (ORPHA:221016)
REEP1	Spastic paraplegia 31, autosomal dominant (OMIM:610250); Spinal muscular atrophy, distal, autosomal recessive, 6 (OMIM:620011); Neuronopathy, distal hereditary motor, type VB (OMIM:614751); Autosomal dominant spastic paraplegia type 31 (ORPHA:101011); Distal hereditary motor neuropathy type 5 (ORPHA:139536)
REL	Immunodeficiency 92 (OMIM:619652); ORPHA:697394
RELL1	—
RELN	Epilepsy, familial temporal lobe, 7 (OMIM:616436); Lissencephaly 2 (OMIM:257320); Epilepsy, familial temporal lobe, 1 (OMIM:600512); Epilepsy with auditory features (ORPHA:101046); Lissencephaly syndrome, Norman-Roberts type (ORPHA:89844)
REN	Tubulointerstitial kidney disease, autosomal dominant, 4 (OMIM:613092); Renal tubular dysgenesis (OMIM:267430); ORPHA:97369; ORPHA:217330
REPS2	—
RERG	—
RET	Hirschsprung disease, susceptibility to, 1 (OMIM:142623); Multiple endocrine neoplasia, type IIA (OMIM:171400); PHEOCHROMOCYTOMA (OMIM:171300); Multiple endocrine neoplasia, type IIB (OMIM:162300); Thyroid carcinoma, familial medullary (OMIM:155240); Hirschsprung disease (ORPHA:388); Haddad syndrome (ORPHA:99803); ORPHA:146; ORPHA:99361; ORPHA:93100; Renal agenesis, bilateral (ORPHA:1848); Sporadic pheochromocytoma/secreting paraganglioma (ORPHA:276621); ORPHA:247709; ORPHA:247698; Hereditary pheochromocytoma-paraganglioma (ORPHA:29072)
RETN	Diabetes mellitus, noninsulin-dependent (OMIM:125853)
RETREG1	Neuropathy, hereditary sensory and autonomic, type II (OMIM:201300); Neuropathy, hereditary sensory and autonomic, type IIB (OMIM:613115); Hereditary sensory and autonomic neuropathy type 2 (ORPHA:970)
RFC2	Williams syndrome (ORPHA:904)
RFT1	Congenital disorder of glycosylation, type In (OMIM:612015); RFT1-CDG (ORPHA:244310)
RFX2	—
RFX5	MHC class II deficiency 5 (OMIM:620818); MHC class II deficiency 3 (OMIM:620816); Immunodeficiency by defective expression of MHC class II (ORPHA:572)
RFX6	Mitchell-Riley syndrome (OMIM:615710); ORPHA:293864
RFX8	—
RFXANK	MHC class II deficiency 2 (OMIM:620815); Immunodeficiency by defective expression of MHC class II (ORPHA:572)
RFXAP	MHC class II deficiency 4 (OMIM:620817); Immunodeficiency by defective expression of MHC class II (ORPHA:572)
RGL1	—
RGMA	—
RGR	Retinitis pigmentosa 44 (OMIM:613769); Retinitis pigmentosa (ORPHA:791)
RGS2	—
RGS5	Hypertension, essential (OMIM:145500)
RGS7	—
RGS9	Prolonged electroretinal response suppression 1 (OMIM:608415); Bradyopsia (ORPHA:75374)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
RGS9BP	Prolonged electroretinal response suppression 2 (OMIM:620344); Bradyopsia (ORPHA:75374)
RHAG	Overhydrated hereditary stomatocytosis (OMIM:185000); Anemia, hemolytic, Rh-null, Regulator type (OMIM:268150); Overhydrated hereditary stomatocytosis (ORPHA:3203); Rh deficiency syndrome (ORPHA:71275)
RHBDF2	Tylosis with esophageal cancer (OMIM:148500); Palmoplantar keratoderma-esophageal carcinoma syndrome (ORPHA:2198)
RHCE	Rh-null disease, amorph type (OMIM:617970); Rh deficiency syndrome (ORPHA:71275)
RHD	OMIM:111690; Hemolytic disease of fetus and newborn, RH-induced (OMIM:619462); Rh deficiency syndrome (ORPHA:71275)
RHO	Night blindness, congenital stationary, autosomal dominant 1 (OMIM:610445); Fundus albipunctatus (OMIM:136880); Retinitis pigmentosa 4 (OMIM:613731); Congenital stationary night blindness (ORPHA:215); Retinitis punctata albescens (ORPHA:52427); Retinitis pigmentosa (ORPHA:791)
RHOG	—
RHOH	Epidermodysplasia verruciformis, susceptibility to, 4 (OMIM:618307); ORPHA:324294
RHPN2	—
RIC1	Catifa syndrome (OMIM:618761); Cleft lip/palate (ORPHA:199306)
RIF1	—
RIGI	Singleton-Merten syndrome 2 (OMIM:616298); ORPHA:85191
RIMS1	Cone rod dystrophy (ORPHA:1872)
RIMS2	Cone-rod synaptic disorder syndrome, congenital nonprogressive (OMIM:618970)
RIMS3	—
RIN2	Macrocephaly, alopecia, cutis laxa, and scoliosis (OMIM:613075); RIN2 syndrome (ORPHA:217335)
RIOK2	—
RIPK4	Popliteal pterygium syndrome, Bartsocas-Papas type 1 (OMIM:263650); CHANDS (OMIM:214350); CHAND syndrome (ORPHA:1401); Bartsocas-Papas syndrome (ORPHA:1234)
RIT2	—
RLBP1	Fundus albipunctatus (OMIM:136880); Newfoundland rod-cone dystrophy (OMIM:607476); Bothnia retinal dystrophy (OMIM:607475); ORPHA:227796; Retinitis punctata albescens (ORPHA:52427); Retinitis pigmentosa (ORPHA:791); Bothnia retinal dystrophy (ORPHA:85128)
RLIG1	—
RMC1	—
RMDN2	—
RMI2	ORPHA:508512
RMND1	Combined oxidative phosphorylation deficiency 11 (OMIM:614922); ORPHA:642976; ORPHA:642945; ORPHA:324535
RN7SKP23	—
RNASEH2B	Aicardi-Goutieres syndrome 2 (OMIM:610181); Aicardi-Goutières syndrome (ORPHA:51); ORPHA:689234
RNASEH2C	Aicardi-Goutieres syndrome 3 (OMIM:610329); Aicardi-Goutières syndrome (ORPHA:51)
RNASEL	Prostate cancer, hereditary, 1 (OMIM:601518); ORPHA:1331
RNASET2	Leukoencephalopathy, cystic, without megalencephaly (OMIM:612951); ORPHA:85136
RNF113A	Trichothiodystrophy 5, nonphotosensitive (OMIM:300953); Trichothiodystrophy (ORPHA:33364)
RNF135	Overgrowth-macrocephaly-facial dysmorphism syndrome (ORPHA:137634)
RNF139	Renal cell carcinoma, nonpapillary (OMIM:144700); ORPHA:422526
RNF157	—
RNF168	Riddle syndrome (OMIM:611943); RIDDLE syndrome (ORPHA:420741)
RNF170	Spastic paraplegia 85, autosomal recessive (OMIM:619686); Ataxia, sensory, autosomal dominant (OMIM:608984); ORPHA:631082
RNF212	Spermatogenic failure 62 (OMIM:619673); OMIM:612042; Male infertility with azoospermia or oligozoospermia due to single gene mutation (ORPHA:399805)
RNF213	Moyamoya disease 2, susceptibility to (OMIM:607151); Moyamoya disease (ORPHA:2573)
RNF217	—
RNF6	Esophageal cancer, somatic (OMIM:133239); Squamous cell carcinoma of the esophagus (ORPHA:99977)
RO60	—
ROBO1	Neurooculorenal syndrome (OMIM:620305); Pituitary hormone deficiency, combined or isolated, 8 (OMIM:620303); Nystagmus, congenital motor, autosomal recessive (OMIM:257400); Pituitary stalk interruption syndrome (ORPHA:95496)
ROBO2	Vesicoureteral reflux 2 (OMIM:610878); ORPHA:289365
ROBO3	Gaze palsy, familial horizontal, with progressive scoliosis, 1 (OMIM:607313); Horizontal gaze palsy with progressive scoliosis (ORPHA:2744)
ROCK1	—
ROGDI	Kohlschutter-Tonz syndrome (OMIM:226750); Amelocerebrohypohidrotic syndrome (ORPHA:1946)
ROM1	Retinitis pigmentosa 7 (OMIM:608133); Retinitis pigmentosa (OMIM:268000); Retinitis pigmentosa (ORPHA:791)
ROR2	Robinow syndrome, autosomal recessive (OMIM:268310); Brachydactyly, type B1 (OMIM:113000); ORPHA:572385; Autosomal recessive Robinow syndrome (ORPHA:1507)
RORA	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia (OMIM:618060); ORPHA:528084
ROS1	ORPHA:251579; Cholangiocarcinoma (ORPHA:70567)
RP1	Retinitis pigmentosa 1 (OMIM:180100); Retinitis pigmentosa (ORPHA:791)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
RP1L1	Retinitis pigmentosa 88 (OMIM:618826); Occult macular dystrophy (OMIM:613587); Retinitis pigmentosa (ORPHA:791); ORPHA:247834
RP2	Retinitis pigmentosa 2, X-linked (OMIM:312600); Retinitis pigmentosa (ORPHA:791)
RP9	Retinitis pigmentosa 9 (OMIM:180104); Retinitis pigmentosa (ORPHA:791)
RPA1	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 6 (OMIM:619767)
RPA4	—
RPE65	Leber congenital amaurosis, type II (OMIM:204100); Retinitis pigmentosa 87 with choroidal involvement (OMIM:618697); Retinitis pigmentosa 20 (OMIM:613794); Leber congenital amaurosis (ORPHA:65); Retinitis pigmentosa (ORPHA:791); Severe early-childhood-onset retinal dystrophy (ORPHA:364055)
RPGR	Retinitis pigmentosa 3 (OMIM:300029); Retinitis pigmentosa, X-linked, and sinorespiratory infections, withor without deafness (OMIM:300455); Macular degeneration, X-linked atrophic (OMIM:300834); Cone-rod dystrophy, X-linked, 1 (OMIM:304020); Primary ciliary dyskinesia (ORPHA:244); Cone rod dystrophy (ORPHA:1872); Retinitis pigmentosa (ORPHA:791); Achromatopsia (ORPHA:49382); ORPHA:247522
RPGRIP1	Cone-rod dystrophy 13 (OMIM:608194); Leber congenital amaurosis 6 (OMIM:613826); Leber congenital amaurosis (ORPHA:65); Cone rod dystrophy (ORPHA:1872); Meckel syndrome (ORPHA:564)
RPGRIP1L	Meckel syndrome, type 5 (OMIM:611561); Joubert syndrome 7 (OMIM:611560); COACH syndrome 3 (OMIM:619113); Joubert syndrome with renal defect (ORPHA:220497); Joubert syndrome with hepatic defect (ORPHA:1454); Meckel syndrome (ORPHA:564)
RPIA	Ribose 5-phosphate isomerase deficiency (OMIM:608611); ORPHA:440706
RPL10	Intellectual developmental disorder, X-linked syndromic 35 (OMIM:300998); OMIM:300847; X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome (ORPHA:435938); X-linked intellectual disability-cerebellar hypoplasia-spondylo-epiphyseal dysplasia syndrome (ORPHA:459070)
RPL11	Diamond-Blackfan anemia 7 (OMIM:612562); Diamond-Blackfan anemia (ORPHA:124)
RPL21P126	—
RPL22	—
RPL24	—
RPL27A	—
RPL35A	Diamond-Blackfan anemia 5 (OMIM:612528); Diamond-Blackfan anemia (ORPHA:124)
RPL36	—
RPL36A	—
RPL37A	—
RPL38	—
RPL5	Diamond-Blackfan anemia 6 (OMIM:612561); Diamond-Blackfan anemia (ORPHA:124)
RPL6	—
RPN2	—
RPP38	—
RPS11	—
RPS15	ORPHA:67038
RPS15AP28	—
RPS17	Diamond-Blackfan anemia 4 (OMIM:612527); Diamond-Blackfan anemia (ORPHA:124)
RPS19	Diamond-Blackfan anemia 1 (OMIM:105650); Diamond-Blackfan anemia (ORPHA:124)
RPS21P1	—
RPS24	Diamond-Blackfan anemia 3 (OMIM:610629); Diamond-Blackfan anemia (ORPHA:124)
RPS26	Diamond-blackfan anemia 10 (OMIM:613309); Diamond-Blackfan anemia (ORPHA:124)
RPS5	—
RPS6KA3	Coffin-Lowry syndrome (OMIM:303600); Intellectual developmental disorder, X-linked 19 (OMIM:300844); Coffin-Lowry syndrome (ORPHA:192); ORPHA:777; Symptomatic form of Coffin-Lowry syndrome in female carriers (ORPHA:276630)
RPS6KL1	—
RPSAP52	—
RPTOR	—
RRAS2	Noonan syndrome 12 (OMIM:618624); Noonan syndrome (ORPHA:648)
RRAS2P1	—
RRH	—
RRM1	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6 (OMIM:620647); Autosomal recessive progressive external ophthalmoplegia (ORPHA:254886)
RRM2	—
RRM2B	Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction (OMIM:268315); Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5 (OMIM:613077); Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy) (OMIM:612075); Mitochondrial neurogastrointestinal encephalomyopathy (ORPHA:298); Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy (ORPHA:329336); Autosomal dominant progressive external ophthalmoplegia (ORPHA:254892); ORPHA:255235; Kearns-Sayre syndrome (ORPHA:480)
RS1	Retinoschisis 1, X-linked, juvenile (OMIM:312700); X-linked retinoschisis (ORPHA:792)
RSC1A1	—
RSKR	—
RSPH4A	Ciliary dyskinesia, primary, 11 (OMIM:612649); Primary ciliary dyskinesia (ORPHA:244)
RSPH9	Ciliary dyskinesia, primary, 12 (OMIM:612650); Primary ciliary dyskinesia (ORPHA:244)
RSPO1	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and 46,xx sex reversal (OMIM:610644);

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	Palmoplantar keratoderma-XX sex reversal-predisposition to squamous cell carcinoma syndrome (ORPHA:85112)
RSPO4	Anonychia congenita (OMIM:206800); ORPHA:94150
RSRC1	Intellectual developmental disorder, autosomal recessive 70 (OMIM:618402); ORPHA:88616
RSRP1	—
RTN2	Spastic paraplegia 12, autosomal dominant (OMIM:604805); Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity (OMIM:620854); Autosomal dominant spastic paraplegia type 12 (ORPHA:100993)
RTN4R	Schizophrenia (OMIM:181500)
RTTN	Microcephaly, short stature, and polymicrogyria with or without seizures (OMIM:614833); Microcephalic cortical malformations-short stature due to RTTN deficiency (ORPHA:468631)
RUBCN	Spinocerebellar ataxia, autosomal recessive 15 (OMIM:615705); Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to RUBCN deficiency (ORPHA:404499)
RUFY2	—
RUNX1	Platelet disorder, familial, with associated myeloid malignancy (OMIM:601399); Leukemia, acute myeloid (OMIM:601626); Aggressive systemic mastocytosis (ORPHA:98850); ORPHA:102724; Chronic myeloid leukemia (ORPHA:521); ORPHA:71290
RUNX1T1	ORPHA:102724
RUNX2	Metaphyseal dysplasia with maxillary hypoplasia and brachydactyly (OMIM:156510); Cleidocranial dysplasia (OMIM:119600); Cleidocranial dysplasia (ORPHA:1452); ORPHA:2504
RUNX3	—
RUSF1	—
RUVBL1	—
RXFP2	—
RXRA	—
RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10 (OMIM:615041); Walker-Warburg syndrome (ORPHA:899)
RYK	—
RYR1	Minicore myopathy with external ophthalmoplegia (OMIM:255320); King-Denborough syndrome (OMIM:619542); Malignant hyperthermia, susceptibility to, 1 (OMIM:145600); Central core disease (OMIM:117000); ORPHA:700188; Benign Samaritan congenital myopathy (ORPHA:324581); Moderate multiminicore disease with hand involvement (ORPHA:178145); Autosomal recessive centronuclear myopathy (ORPHA:169186); Autosomal dominant centronuclear myopathy (ORPHA:169189); Exercise-induced malignant hyperthermia (ORPHA:466650); Congenital multicore myopathy with external ophthalmoplegia (ORPHA:98905); Congenital myopathy with myasthenic-like onset (ORPHA:424107); Central core disease (ORPHA:597); ORPHA:33108; Malignant hyperthermia of anesthesia (ORPHA:423); ORPHA:99741
RYR2	Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome (OMIM:115000); Ventricular tachycardia, catecholaminergic polymorphic, 1, with or without atrial dysfunction and/or dilated cardiomyopathy (OMIM:604772); Catecholaminergic polymorphic ventricular tachycardia (ORPHA:3286); ORPHA:293888; ORPHA:293899; ORPHA:293910
RYR3	Congenital myopathy 20 (OMIM:620310)
S100PBP	—
SAA1	AA amyloidosis (ORPHA:85445)
SACS	Spastic ataxia, Charlevoix-Saguenay type (OMIM:270550); Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ORPHA:98)
SAG	Retinitis pigmentosa 96, autosomal dominant (OMIM:620228); Oguchi disease 1 (OMIM:258100); Retinitis pigmentosa 47 (OMIM:613758); Oguchi disease (ORPHA:75382); Retinitis pigmentosa (ORPHA:791)
SAGE1	—
SALL1	Townes-Brocks syndrome 1 (OMIM:107480); Townes-Brocks syndrome (ORPHA:857)
SALL4	IVIC syndrome (OMIM:147750); Duane-Radial ray syndrome (OMIM:607323); Duane retraction syndrome (ORPHA:233); ORPHA:261647; ORPHA:261638; Acro-renal-ocular syndrome (ORPHA:959); IVIC syndrome (ORPHA:2307)
SAMD9	Tumoral calcinosis, normophosphatemic, familial (OMIM:610455); Monosomy 7 myelodysplasia and leukemia syndrome 2 (OMIM:619041); Mirage syndrome (OMIM:617053); ORPHA:494433; ORPHA:306658
SAMHD1	Aicardi-Goutieres syndrome 5 (OMIM:612952); Chilblain lupus 2 (OMIM:614415); Aicardi-Goutières syndrome (ORPHA:51); ORPHA:481662
SAR1B	Chylomicron retention disease (OMIM:246700); Chylomicron retention disease (ORPHA:71)
SARDH	SARCOSINEMIA (OMIM:268900); Sarcosinemia (ORPHA:3129)
SARM1	—
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis syndrome (OMIM:613845); ORPHA:363694
SART3	—
SAT1	Keratosis follicularis spinulosa decalvans (ORPHA:2340); Pediatric systemic lupus erythematosus (ORPHA:93552)
SATB2	Glass syndrome (OMIM:612313); SATB2-associated syndrome due to a pathogenic variant (ORPHA:576283); SATB2-associated syndrome due to a chromosomal rearrangement (ORPHA:251028); 2q32q33 deletion syndrome (ORPHA:251019)
SATL1	—
SAV1	—

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SBDS	Aplastic anemia (OMIM:609135); Shwachman-Diamond syndrome 1 (OMIM:260400); Idiopathic aplastic anemia (ORPHA:88); Shwachman-Diamond syndrome (ORPHA:811); ORPHA:622934
SBF2	Charcot-Marie-Tooth disease, type 4B2 (OMIM:604563); Charcot-Marie-Tooth disease type 4B2 (ORPHA:99956)
SC5D	Lathosterolosis (OMIM:607330); Lathosterolosis (ORPHA:46059)
SCARB1	OMIM:610762; ORPHA:181428
SCARB2	Epilepsy, progressive myoclonic, 4, with or without renal failure (OMIM:254900); Progressive myoclonic epilepsy type 1 (ORPHA:308); Gaucher disease type 1 (ORPHA:77259); ORPHA:163696
SCARF2	Van den Ende-Gupta syndrome (OMIM:600920); Van den Ende-Gupta syndrome (ORPHA:2460)
SCAT8	—
SCGB1A1	—
SCGB3A2	Asthma, susceptibility to (OMIM:600807)
SCLT1	Bardet-Biedl syndrome (ORPHA:110)
SCN10A	Episodic pain syndrome, familial, 2 (OMIM:615551); Romano-Ward syndrome (ORPHA:101016); ORPHA:306577; Brugada syndrome (ORPHA:130); ORPHA:88642; Primary erythromelalgia (ORPHA:90026); Paroxysmal extreme pain disorder (ORPHA:46348)
SCN11A	Neuropathy, hereditary sensory and autonomic, type VII (OMIM:615548); Episodic pain syndrome, familial, 3 (OMIM:615552); ORPHA:306577; ORPHA:391397; ORPHA:391392; ORPHA:88642; Primary erythromelalgia (ORPHA:90026); Paroxysmal extreme pain disorder (ORPHA:46348)
SCN1A	Generalized epilepsy with febrile seizures plus, type 2 (OMIM:604403); Epileptic encephalopathy, early infantile, 6 (Dravet syndrome) (OMIM:607208); Migraine, familial hemiplegic, 3 (OMIM:609634); Developmental and epileptic encephalopathy 6B, non-Dravet (OMIM:619317); Genetic epilepsy with febrile seizure plus (ORPHA:36387); Lennox-Gastaut syndrome (ORPHA:2382); Non-specific early-onset epileptic encephalopathy (ORPHA:442835); Epilepsy with myoclonic-atonic seizures (ORPHA:1942); Epilepsy of infancy with migrating focal seizures (ORPHA:293181); Familial or sporadic hemiplegic migraine (ORPHA:569); Dravet syndrome (ORPHA:33069)
SCN1B	Brugada syndrome 5 (OMIM:612838); Generalized epilepsy with febrile seizures plus, type 1 (OMIM:604233); Atrial fibrillation, familial, 13 (OMIM:615377); Epileptic encephalopathy, early infantile, 52 (OMIM:617350); Hereditary atrial fibrillation (ORPHA:334); Genetic epilepsy with febrile seizure plus (ORPHA:36387); Brugada syndrome (ORPHA:130); Hereditary progressive cardiac conduction defect (ORPHA:871); Early infantile developmental and epileptic encephalopathy (ORPHA:1934); Dravet syndrome (ORPHA:33069)
SCN2A	Episodic ataxia, type 9 (OMIM:618924); Epileptic encephalopathy, early infantile, 11 (OMIM:613721); Seizures, benign familial infantile, 3 (OMIM:607745); Self-limited infantile epilepsy (ORPHA:306); Genetic epilepsy with febrile seizure plus (ORPHA:36387); Infantile epileptic spasms syndrome (ORPHA:697160); Early infantile developmental and epileptic encephalopathy (ORPHA:1934); Epilepsy of infancy with migrating focal seizures (ORPHA:293181); Self-limited neonatal-infantile epilepsy (ORPHA:140927); Alternating hemiplegia of childhood (ORPHA:2131); Dravet syndrome (ORPHA:33069)
SCN2B	Atrial fibrillation, familial, 14 (OMIM:615378); Hereditary atrial fibrillation (ORPHA:334); Brugada syndrome (ORPHA:130)
SCN3A	Epilepsy, familial focal, with variable foci 4 (OMIM:617935); Developmental and epileptic encephalopathy 62 (OMIM:617938); Familial focal epilepsy with variable foci (ORPHA:98820); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
SCN3B	Brugada syndrome 7 (OMIM:613120); Hereditary atrial fibrillation (ORPHA:334); Brugada syndrome (ORPHA:130)
SCN4A	Myotonia, potassium-aggravated (OMIM:608390); Myasthenic syndrome, congenital, 16 (OMIM:614198); Hyperkalemic periodic paralysis (OMIM:170500); Hypokalemic periodic paralysis, type 1 (OMIM:170400); Hypokalemic periodic paralysis, type 2 (OMIM:613345); Paramyotonia congenita (OMIM:168300); Congenital myopathy 22A, classic (OMIM:620351); Congenital myopathy 22B, severe fetal (OMIM:620369); Postsynaptic congenital myasthenic syndrome (ORPHA:98913); Paramyotonia congenita of Von Eulenburg (ORPHA:684); Hyperkalemic periodic paralysis (ORPHA:682); Hypokalemic periodic paralysis (ORPHA:681); Myotonia permanens (ORPHA:99735); Acetazolamide-responsive myotonia (ORPHA:99736); Myotonia fluctuans (ORPHA:99734)
SCN4B	Long QT syndrome 10 (OMIM:611819); Romano-Ward syndrome (ORPHA:101016); Hereditary atrial fibrillation (ORPHA:334)
SCN5A	Progressive familial heart block, type IA (OMIM:113900); Sudden infant death syndrome (OMIM:272120); Sick sinus syndrome 1 (OMIM:608567); Atrial fibrillation, familial, 10 (OMIM:614022); Long QT syndrome 3 (OMIM:603830); Ventricular fibrillation, familial, 1 (OMIM:603829); Cardiomyopathy, dilated, 1E (OMIM:601154); Brugada syndrome 1 (OMIM:601144); Romano-Ward syndrome (ORPHA:101016); Hereditary atrial fibrillation (ORPHA:334); Familial isolated dilated cardiomyopathy (ORPHA:154); ORPHA:228140; Brugada syndrome (ORPHA:130); ORPHA:166282; Hereditary progressive cardiac conduction defect (ORPHA:871); Isolated atrial standstill (ORPHA:1344)
SCN7A	—
SCN8A	Cognitive impairment with or without cerebellar ataxia (OMIM:614306); Myoclonus, familial, 2 (OMIM:618364); Seizures, benign familial infantile, 5 (OMIM:617080); Epileptic encephalopathy, early infantile, 13 (OMIM:614558); Self-limited infantile epilepsy (ORPHA:306); ORPHA:178469; Familial infantile myoclonic epilepsy (ORPHA:352582); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
SCN9A	Neuropathy, hereditary sensory and autonomic, type II (OMIM:201300); Indifference to pain, congenital, autosomal recessive (OMIM:243000); Erythermalgia, primary (OMIM:133020); Paroxysmal extreme pain disorder (OMIM:167400); ORPHA:306577; Genetic epilepsy with febrile seizure plus (ORPHA:36387); ORPHA:88642; Hereditary sensory and autonomic neuropathy type 2 (ORPHA:970); Primary erythromelalgia

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	(ORPHA:90026); Paroxysmal extreme pain disorder (ORPHA:46348); Dravet syndrome (ORPHA:33069)
SCNN1A	Bronchiectasis with or without elevated sweat chloride 2 (OMIM:613021); Liddle syndrome 3 (OMIM:618126); Pseudohypoaldosteronism, type I, autosomal recessive (OMIM:264350); Generalized pseudohypoaldosteronism type 1 (ORPHA:171876); Idiopathic bronchiectasis (ORPHA:60033); Brugada syndrome (ORPHA:130); Liddle syndrome (ORPHA:526)
SCNN1B	Liddle syndrome 1 (OMIM:177200); Pseudohypoaldosteronism, type IB2, autosomal recessive (OMIM:620125); Bronchiectasis with or without elevated sweat chloride 1 (OMIM:211400); Generalized pseudohypoaldosteronism type 1 (ORPHA:171876); Idiopathic bronchiectasis (ORPHA:60033); Liddle syndrome (ORPHA:526)
SCNN1D	—
SCNN1G	Bronchiectasis with or without elevated sweat chloride 3 (OMIM:613071); Pseudohypoaldosteronism, type IB3, autosomal recessive (OMIM:620126); Liddle syndrome 2 (OMIM:618114); Generalized pseudohypoaldosteronism type 1 (ORPHA:171876); Idiopathic bronchiectasis (ORPHA:60033); Liddle syndrome (ORPHA:526)
SCO1	Mitochondrial complex IV deficiency, nuclear type 4 (OMIM:619048); Mitochondrial complex IV deficiency, nuclear type 1 (OMIM:220110); ORPHA:1561
SCO2	Mitochondrial complex IV deficiency, nuclear type 2 (OMIM:604377); Myopia 6 (OMIM:608908); Mitochondrial complex IV deficiency, nuclear type 1 (OMIM:220110); ORPHA:98619; ORPHA:1561; Autosomal recessive axonal Charcot-Marie-Tooth disease due to copper metabolism defect (ORPHA:521411)
SCP2	Leukoencephalopathy with dystonia and motor neuropathy (OMIM:613724); ORPHA:163684
SCRIB	—
SCUBE2	—
SDC3	OBESITY (OMIM:601665)
SDCBP2	—
SDCCAG8	Bardet-Biedl syndrome 16 (OMIM:615993); Senior-Loken syndrome 7 (OMIM:613615); Senior-Loken syndrome (ORPHA:3156); Bardet-Biedl syndrome (ORPHA:110)
SDHA	Neurodegeneration with ataxia and late-onset optic atrophy (OMIM:619259); Paragangliomas 5 (OMIM:614165); Mitochondrial complex II deficiency (OMIM:252011); Cardiomyopathy, dilated, 1GG (OMIM:613642); Carney-Stratakis syndrome (ORPHA:97286); Isolated succinate-CoQ reductase deficiency (ORPHA:3208); Gastrointestinal stromal tumor (ORPHA:44890); Familial isolated dilated cardiomyopathy (ORPHA:154); Carney triad (ORPHA:139411); Hereditary pheochromocytoma-paraganglioma (ORPHA:29072)
SDHAF1	Mitochondrial complex II deficiency, nuclear type 2 (OMIM:619166); Isolated succinate-CoQ reductase deficiency (ORPHA:3208)
SDHAF2	Paragangliomas 2 (OMIM:601650); Hereditary pheochromocytoma-paraganglioma (ORPHA:29072)
SDHB	Carney-Stratakis syndrome (OMIM:606864); Mitochondrial complex II deficiency, nuclear type 4 (OMIM:619224); Paragangliomas 4 (OMIM:115310); Gastrointestinal stromal tumor (OMIM:606764); Carney-Stratakis syndrome (ORPHA:97286); Isolated succinate-CoQ reductase deficiency (ORPHA:3208); Cowden syndrome (ORPHA:201); Gastrointestinal stromal tumor (ORPHA:44890); Carney triad (ORPHA:139411); Sporadic pheochromocytoma/secreting paraganglioma (ORPHA:276621); Hereditary pheochromocytoma-paraganglioma (ORPHA:29072)
SDHC	Carney-Stratakis syndrome (OMIM:606864); Gastrointestinal stromal tumor (OMIM:606764); Paragangliomas 3 (OMIM:605373); Carney-Stratakis syndrome (ORPHA:97286); Cowden syndrome (ORPHA:201); Gastrointestinal stromal tumor (ORPHA:44890); Carney triad (ORPHA:139411); Hereditary pheochromocytoma-paraganglioma (ORPHA:29072)
SDHD	Carney-Stratakis syndrome (OMIM:606864); Mitochondrial complex II deficiency, nuclear type 3 (OMIM:619167); Paragangliomas 1 (OMIM:168000); Carney-Stratakis syndrome (ORPHA:97286); Isolated succinate-CoQ reductase deficiency (ORPHA:3208); Cowden syndrome (ORPHA:201); Carcinoid syndrome (ORPHA:100093); Sporadic pheochromocytoma/secreting paraganglioma (ORPHA:276621); Hereditary pheochromocytoma-paraganglioma (ORPHA:29072)
SEC23A	Craniolenticulosutural dysplasia (OMIM:607812); Craniolenticulosutural dysplasia (ORPHA:50814)
SEC23B	Anemia, dyserythropoietic congenital, type II (OMIM:224100); Cowden syndrome 7 (OMIM:616858); Cowden syndrome (ORPHA:201); ORPHA:98873
SEC61A1	Neutropenia, severe congenital, 11, autosomal dominant (OMIM:620674); Immunodeficiency, common variable, 15 (OMIM:620670); Tubulointerstitial kidney disease, autosomal dominant, 5 (OMIM:617056); ORPHA:697417
SEC63	Polycystic liver disease 2 (OMIM:617004); Isolated polycystic liver disease (ORPHA:2924)
SECISBP2	Thyroid hormone metabolism, abnormal (OMIM:609698); Short stature-delayed bone age due to thyroid hormone metabolism deficiency (ORPHA:171706)
SEL1L2	—
SELE	—
SELENON	Rigid spine muscular dystrophy 1 (OMIM:602771); Rigid spine syndrome (ORPHA:97244); Congenital fiber-type disproportion myopathy (ORPHA:2020); ORPHA:84132; Classic multiminicore myopathy (ORPHA:324604)
SELL	—
SELP	—
SELPLG	—
SEM1	Isolated split hand-split foot malformation (ORPHA:2440)
SEMA3A	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Hypogonadotropic hypogonadism 16 with or without anosmia (OMIM:614897); Brugada syndrome (ORPHA:130); Kallmann syndrome (ORPHA:478)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
SEMA3E	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); CHARGE syndrome (ORPHA:138)
SEMA4A	Retinitis pigmentosa 35 (OMIM:610282); Cone-Rod dystrophy 10 (OMIM:610283); Cone rod dystrophy (ORPHA:1872); Retinitis pigmentosa (ORPHA:791); Familial colorectal cancer Type X (ORPHA:440437)
SEMA4C	—
SEMA4G	—
SEMA6D	—
SEMA7A	OMIM:614745; Cholestasis, progressive familial intrahepatic, 11 (OMIM:619874)
SEMG1	—
SENCR	—
SEPSECS	Pontocerebellar hypoplasia, type 2D (OMIM:613811); Pontocerebellar hypoplasia type 2 (ORPHA:2524); ORPHA:247198
SEPTIN12	Spermatogenic failure 10 (OMIM:614822); ORPHA:276234
SEPTIN4	ORPHA:171709
SEPTIN5	—
SEPTIN7	—
SERAC1	3-Methylglutaconic aciduria with deafness, encephalopathy, and leigh-like syndrome (OMIM:614739); ORPHA:352328
SERPINA1	Alpha-1-Antitrypsin deficiency (OMIM:613490); ORPHA:178396; Alpha-1-antitrypsin deficiency (ORPHA:60); Cystic fibrosis (ORPHA:586)
SERPINA10	—
SERPINA3	—
SERPINA6	Corticosteroid-binding globulin deficiency (OMIM:611489); ORPHA:199247
SERPINA7	OMIM:300932
SERPINB11	—
SERPINB5	—
SERPINB6	Deafness, autosomal recessive 91 (OMIM:613453); ORPHA:90636
SERPINC1	Antithrombin III deficiency (OMIM:613118); Hereditary thrombophilia due to congenital antithrombin deficiency (ORPHA:82)
SERPIND1	Heparin cofactor II deficiency (OMIM:612356)
SERPINE1	Plasminogen activator inhibitor-1 deficiency (OMIM:613329); Congenital plasminogen activator inhibitor type 1 deficiency (ORPHA:465); ORPHA:673556
SERPINF1	Osteogenesis imperfecta, type VI (OMIM:613982); ORPHA:216812; ORPHA:216820
SERPINF2	Plasmin inhibitor deficiency (OMIM:262850); Congenital alpha2-antiplasmin deficiency (ORPHA:79)
SERPING1	Angioedema, hereditary, 1 (OMIM:106100); Complement component 4, partial deficiency of (OMIM:120790); Hereditary angioedema type 1 (ORPHA:100050); ORPHA:100051
SERPINH1	Preterm premature rupture of the membranes (OMIM:610504); Osteogenesis imperfecta, type X (OMIM:613848); ORPHA:216812
SERPINI1	Encephalopathy, familial, with neuroserpin inclusion bodies (OMIM:604218); ORPHA:530303
SERPINI2	—
SESN2	—
SETBP1	Schinzel-Giedion midface-retraction syndrome (OMIM:269150); Intellectual developmental disorder, autosomal dominant 29 (OMIM:616078); Schinzel-Giedion syndrome (ORPHA:798); ORPHA:436151
SETD2	Intellectual developmental disorder, autosomal dominant 70 (OMIM:620157); Rabin-Pappas syndrome (OMIM:620155); Luscan-Lumish syndrome (OMIM:616831); ORPHA:597738; ORPHA:597743
SETD4	—
SETX	Amyotrophic lateral sclerosis 4, juvenile (OMIM:602433); Spinocerebellar ataxia, autosomal recessive 1 (OMIM:606002); Spinocerebellar ataxia with axonal neuropathy type 2 (ORPHA:64753); Amyotrophic lateral sclerosis type 4 (ORPHA:357043)
SEZ6	—
SEZ6L	—
SEZ6L2	—
SF1	—
SF3B1	Myelodysplastic syndrome (OMIM:614286); Acquired idiopathic sideroblastic anemia (ORPHA:75564); Uveal melanoma (ORPHA:39044)
SF3B4	Acrofacial dysostosis 1, Nager type (OMIM:154400); Nager syndrome (ORPHA:245); Acrofacial dysostosis, Rodríguez type (ORPHA:1788)
SFPQ	ORPHA:319308
SFRP1	—
SFTPA1	Interstitial lung disease 1 (OMIM:619611); Idiopathic pulmonary fibrosis (ORPHA:2032)
SFTPA2	Pulmonary fibrosis, idiopathic (OMIM:178500); Idiopathic pulmonary fibrosis (ORPHA:2032)
SFTPB	Surfactant metabolism dysfunction, pulmonary, 1 (OMIM:265120); Pediatric acute respiratory distress syndrome (ORPHA:685082); Neonatal acute respiratory distress syndrome (ORPHA:217563)
SFTPC	Pulmonary fibrosis, idiopathic (OMIM:178500); Surfactant metabolism dysfunction, pulmonary, 2 (OMIM:610913); Pediatric acute respiratory distress syndrome (ORPHA:685082); ORPHA:217566; Idiopathic pulmonary fibrosis (ORPHA:2032); ORPHA:440392
SFTPD	—
SFXN5	—
SGCA	Muscular dystrophy, limb-girdle, type 2D (OMIM:608099); Alpha-sarcoglycan-related limb-girdle muscular dystrophy R3 (ORPHA:62)

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SGCB	Muscular dystrophy, limb-girdle, type 2E (OMIM:604286); Beta-sarcoglycan-related limb-girdle muscular dystrophy R4 (ORPHA:119)
SGCD	Muscular dystrophy, limb-girdle, type 2F (OMIM:601287); Cardiomyopathy, dilated, 1L (OMIM:606685); Delta-sarcoglycan-related limb-girdle muscular dystrophy R6 (ORPHA:219); Familial isolated dilated cardiomyopathy (ORPHA:154)
SGCE	Dystonia-11, myoclonic (OMIM:159900); Myoclonus-dystonia syndrome (ORPHA:36899)
SGCG	Muscular dystrophy, limb-girdle, type 2C (OMIM:253700); Gamma-sarcoglycan-related limb-girdle muscular dystrophy R5 (ORPHA:353)
SGPL1	Nephrotic syndrome, type 14 (OMIM:617575); ORPHA:506334
SGSH	Mucopolysaccharidosis type IIIA (OMIM:252900); ORPHA:79269
SH2B1	Severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency (ORPHA:329249); Distal 16p11.2 microdeletion syndrome (ORPHA:261222); Proximal 16p11.2 microdeletion syndrome (ORPHA:261197)
SH2B3	Erythrocytosis, familial, 1 (OMIM:133100); Myelofibrosis with myeloid metaplasia, somatic (OMIM:254450); Thrombocythemia 1 (OMIM:187950); Essential thrombocythemia (ORPHA:3318); ORPHA:391366
SH2D1A	Lymphoproliferative syndrome, X-linked, 1 (OMIM:308240); ORPHA:538931
SH2D2A	—
SH3BP2	Cherubism (OMIM:118400); Cherubism (ORPHA:184)
SH3GL1	Leukemia, acute myeloid (OMIM:601626)
SH3PXD2B	Frank-ter Haar syndrome (OMIM:249420); Frank-Ter Haar syndrome (ORPHA:137834)
SH3TC2	Mononeuropathy of the median nerve, mild (OMIM:613353); Charcot-Marie-Tooth disease, type 4C (OMIM:601596); Charcot-Marie-Tooth disease type 4C (ORPHA:99949)
SH3YL1	—
SHANK2	OMIM:613436
SHANK3	Phelan-Mcdermid syndrome (OMIM:606232); Schizophrenia 15 (OMIM:613950); ORPHA:662169; ORPHA:662172
SHARPIN	Autoinflammation with episodic fever and immune dysregulation (OMIM:620795)
SHBG	—
SHFL	—
SHH	Microphthalmia, isolated, with coloboma 5 (OMIM:611638); Solitary median maxillary central incisor (OMIM:147250); Holoprosencephaly 3 (OMIM:142945); Septopreoptic holoprosencephaly (ORPHA:280195); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925); Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome (ORPHA:988); ORPHA:98938; ORPHA:476119; ORPHA:93336; Isolated radial hemimelia (ORPHA:93321); Syndactyly type 4 (ORPHA:93405); ORPHA:485275
SHMT1	—
SHOC2	Noonan syndrome-like with loose anagen hair 1 (OMIM:607721); Noonan syndrome-like disorder with loose anagen hair (ORPHA:2701)
SHOX	Langer mesomelic dysplasia (OMIM:249700); Leri-Weill dyschondrosteosis (OMIM:127300); Short stature, idiopathic familial (OMIM:300582); Léri-Weill dyschondrosteosis (ORPHA:240); Langer mesomelic dysplasia (ORPHA:2632); SHOX-related short stature (ORPHA:314795)
SHOX2	—
SHROOM2	—
SHROOM3	—
SHROOM4	X-linked intellectual disability, Stocco Dos Santos type (ORPHA:85288)
SI	Sucrase-isomaltase deficiency, congenital (OMIM:222900); Congenital sucrase-isomaltase deficiency (ORPHA:35122)
SIAE	OMIM:613551
SIGLEC12	—
SIGLEC14	—
SIGLEC5	—
SIGMAR1	Spinal muscular atrophy, distal, autosomal recessive, 2 (OMIM:605726); Amyotrophic lateral sclerosis 16, juvenile (OMIM:614373); ORPHA:139552; Juvenile amyotrophic lateral sclerosis (ORPHA:300605)
SIK2	—
SIK3	Spondyloepimetaphyseal dysplasia, Krakow type (OMIM:618162)
SIL1	Marinesco-Sjogren syndrome (OMIM:248800); Marinesco-Sjögren syndrome (ORPHA:559)
SIM1	6q16 microdeletion syndrome (ORPHA:171829); SIM1-related Prader-Willi-like syndrome (ORPHA:398079); Obesity due to SIM1 deficiency (ORPHA:369873)
SIM2	—
SIPA1	—
SIPA1L1	—
SIRT1	—
SIRT3	—
SIX1	Branchiootic syndrome 3 (OMIM:608389); Branchiootorenal syndrome 1 (OMIM:113650); Deafness, autosomal dominant 23 (OMIM:605192); ORPHA:90635; Branchiootic syndrome (ORPHA:52429); BOR syndrome (ORPHA:107)
SIX2	SIX2-related frontonasal dysplasia (ORPHA:488437)
SIX3	Holoprosencephaly 2 (OMIM:157170); SCHIZENCEPHALY (OMIM:269160); Septopreoptic holoprosencephaly

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	(ORPHA:280195); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925); ORPHA:485275
SIX5	Branchiootorenal syndrome 2 (OMIM:610896); BOR syndrome (ORPHA:107)
SIX6	Optic disc anomalies with retinal and/or macular dystrophy (OMIM:212550); Microphthalmia, syndromic 3 (OMIM:206900); ORPHA:435930; ORPHA:98938; Nanophthalmos (ORPHA:35612)
SKI	Shprintzen-Goldberg craniosynostosis syndrome (OMIM:182212); Shprintzen-Goldberg syndrome (ORPHA:2462); 1p36 deletion syndrome (ORPHA:1606)
SKIC3	Trichohepatoenteric syndrome 1 (OMIM:222470); Trichohepatoenteric syndrome (ORPHA:84064)
SLA	—
SLA2	—
SLC10A1	Hypercholanemia, familial 2 (OMIM:619256); ORPHA:238475
SLC10A2	Bile acid malabsorption, primary (OMIM:613291)
SLC11A1	OMIM:610446; OMIM:607948; ORPHA:3389; Cystic fibrosis (ORPHA:586)
SLC11A2	Anemia, hypochromic microcytic, with iron overload 1 (OMIM:206100); ORPHA:83642
SLC12A1	Bartter syndrome, type 1, antenatal (OMIM:601678)
SLC12A3	Gitelman syndrome (OMIM:263800); Gitelman syndrome (ORPHA:358)
SLC12A4	—
SLC12A5	Epilepsy, idiopathic generalized, susceptibility to, 14 (OMIM:616685); Epileptic encephalopathy, early infantile, 34 (OMIM:616645); Epilepsy of infancy with migrating focal seizures (ORPHA:293181)
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy (OMIM:218000); Charcot-Marie-Tooth disease, axonal, type 2II (OMIM:620068); Corpus callosum agenesis-neuronopathy syndrome (ORPHA:1496)
SLC13A2	—
SLC14A1	OMIM:111000
SLC15A1	—
SLC16A1	Hyperinsulinemic hypoglycemia, familial, 7 (OMIM:610021); Erythrocyte lactate transporter defect (OMIM:245340); Monocarboxylate transporter 1 deficiency (OMIM:616095); ORPHA:165991; ORPHA:171690; ORPHA:438075
SLC16A12	Cataract, juvenile, with microcornea and glucosuria (OMIM:612018); ORPHA:247794
SLC16A2	Allan-Herndon-Dudley syndrome (OMIM:300523); Allan-Herndon-Dudley syndrome (ORPHA:59)
SLC16A3	—
SLC17A3	Gout susceptibility 4 (OMIM:612671)
SLC17A5	Salla disease (OMIM:604369); Infantile sialic acid storage disease (OMIM:269920); ORPHA:309331; ORPHA:309334; ORPHA:309324
SLC17A8	Deafness, autosomal dominant 25 (OMIM:605583); ORPHA:90635
SLC19A1	Immunodeficiency 114, folate-responsive (OMIM:620603); Megaloblastic anemia, folate-responsive (OMIM:601775)
SLC19A2	Thiamine-Responsive megaloblastic anemia syndrome (OMIM:249270); Thiamine-responsive megaloblastic anemia syndrome (ORPHA:49827)
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2) (OMIM:607483); ORPHA:199348; Infantile spasms-psychomotor retardation-progressive brain atrophy-basal ganglia disease syndrome (ORPHA:263410); ORPHA:65284
SLC1A1	OMIM:615232; Dicarboxylicamino aciduria (OMIM:222730); ORPHA:166412; ORPHA:2195
SLC1A2	Epileptic encephalopathy, early infantile, 41 (OMIM:617105); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
SLC1A3	Episodic ataxia, type 6 (OMIM:612656); Episodic ataxia type 6 (ORPHA:209967); Alternating hemiplegia of childhood (ORPHA:2131)
SLC1A5	—
SLC20A2	Basal ganglia calcification, idiopathic, 1 (OMIM:213600); Bilateral striopallidodentate calcinosis (ORPHA:1980)
SLC22A1	—
SLC22A12	Hypouricemia, renal, 1 (OMIM:220150); Hereditary renal hypouricemia (ORPHA:94088)
SLC22A18	—
SLC22A18AS	—
SLC22A2	—
SLC22A3	—
SLC22A4	Rheumatoid arthritis (OMIM:180300)
SLC22A5	Carnitine deficiency, systemic primary (OMIM:212140); Systemic primary carnitine deficiency (ORPHA:158)
SLC22A6	—
SLC22A9	—
SLC24A1	Night blindness, congenital stationary, type 1D (OMIM:613830)
SLC24A2	—
SLC24A5	Albinism, oculocutaneous, type VI (OMIM:113750); Oculocutaneous albinism type 6 (ORPHA:370097)
SLC25A12	Epileptic encephalopathy, early infantile, 39 (OMIM:612949); ORPHA:353217
SLC25A13	Citrullinemia, type II, neonatal-onset (OMIM:605814); Citrullinemia, type II, adult-onset (OMIM:603471); Neonatal intrahepatic cholestasis due to citrin deficiency (ORPHA:247598); Citrullinemia type II (ORPHA:247585)
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (OMIM:238970); Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (ORPHA:415)
SLC25A19	Microcephaly, Amish type (OMIM:607196); Thiamine metabolism dysfunction syndrome 4 (bilateral striatal

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	degenerationand progressive polyneuropathy type) (OMIM:613710); ORPHA:217396; Amish lethal microcephaly (ORPHA:99742)
SLC25A20	Carnitine-acylcarnitine translocase deficiency (OMIM:212138); Carnitine-acylcarnitine translocase deficiency (ORPHA:159)
SLC25A22	Epileptic encephalopathy, early infantile, 3 (OMIM:609304); Early infantile developmental and epileptic encephalopathy (ORPHA:1934); Epilepsy of infancy with migrating focal seizures (ORPHA:293181)
SLC25A3	Mitochondrial phosphate carrier deficiency (OMIM:610773); Cardiomyopathy-hypotonia-lactic acidosis syndrome (ORPHA:91130)
SLC25A35	—
SLC25A38	Anemia, sideroblastic, 2, pyridoxine-refractory (OMIM:205950); ORPHA:260305
SLC25A39	—
SLC25A4	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2 (OMIM:609283); Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type) (OMIM:615418); Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type), autosomal dominant (OMIM:617184); Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome (ORPHA:1369); Autosomal dominant progressive external ophthalmoplegia (ORPHA:254892)
SLC26A1	Nephrolithiasis, calcium oxalate (OMIM:167030); Hypersulfaturia (OMIM:620372)
SLC26A11	—
SLC26A2	Achondrogenesis, type IB (OMIM:600972); Epiphyseal dysplasia, multiple, 4 (OMIM:226900); Atelosteogenesis, type II (OMIM:256050); Diastrophic dysplasia (OMIM:222600); Atelosteogenesis type II (ORPHA:56304); Achondrogenesis type 1B (ORPHA:93298); Diastrophic dysplasia (ORPHA:628); Multiple epiphyseal dysplasia type 4 (ORPHA:93307)
SLC26A3	Diarrhea 1, secretory chloride, congenital (OMIM:214700); ORPHA:53689
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct (OMIM:600791); Pendred syndrome (OMIM:274600); ORPHA:90636; Thyroid hypoplasia (ORPHA:95720); Athyreosis (ORPHA:95713); Pendred syndrome (ORPHA:705)
SLC26A5	Deafness, autosomal recessive 61 (OMIM:613865); ORPHA:90636
SLC26A6	—
SLC26A9	Cystic fibrosis (ORPHA:586)
SLC27A1	—
SLC27A4	Ichthyosis prematurity syndrome (OMIM:608649); Ichthyosis-prematurity syndrome (ORPHA:88621)
SLC27A5	—
SLC28A1	URIDINE-CYTIDINEURIA (OMIM:618477)
SLC28A2	—
SLC28A3	—
SLC29A1	—
SLC29A2	—
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome (OMIM:602782); H syndrome (ORPHA:168569); Dysosteosclerosis (ORPHA:1782)
SLC2A1	Dystonia 9 (OMIM:601042); Glut1 deficiency syndrome 2 (OMIM:612126); Stomatin-deficient cryohydrocytosis with neurologic defects (OMIM:608885); GLUT1 deficiency syndrome 1, infantile onset, severe (OMIM:606777); Epilepsy, idiopathic generalized, susceptibility to, 12 (OMIM:614847); Paroxysmal exertion-induced dyskinesia (ORPHA:98811); ORPHA:86911; Epilepsy with myoclonic-atonic seizures (ORPHA:1942); Hereditary cryohydrocytosis with reduced stomatin (ORPHA:168577); Alternating hemiplegia of childhood (ORPHA:2131); Classic glucose transporter type 1 deficiency syndrome (ORPHA:71277); Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity (ORPHA:53583)
SLC2A10	Arterial tortuosity syndrome (OMIM:208050); Arterial tortuosity syndrome (ORPHA:3342)
SLC2A2	Diabetes mellitus, noninsulin-dependent (OMIM:125853); Fanconi-Bickel syndrome (OMIM:227810); Fanconi-Bickel syndrome (ORPHA:2088)
SLC2A4	—
SLC2A9	Hypouricemia, renal, 2 (OMIM:612076); Hereditary renal hypouricemia (ORPHA:94088)
SLC30A10	Hypermanganesemia with dystonia 1 (OMIM:613280); Cirrhosis-dystonia-polycythemia-hypermanganesemia syndrome (ORPHA:309854)
SLC30A2	Zinc deficiency, transient neonatal (OMIM:608118)
SLC30A5	—
SLC30A8	Diabetes mellitus, noninsulin-dependent (OMIM:125853)
SLC31A1	Neurodegeneration and seizures due to copper transport defect (OMIM:620306)
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration (OMIM:614482); Spastic paraplegia 42, autosomal dominant (OMIM:612539); Autosomal dominant spastic paraplegia type 42 (ORPHA:171863); ORPHA:300313
SLC34A1	Hypercalcemia, infantile 2 (OMIM:616963); Fanconi renotubular syndrome 2 (OMIM:613388); Nephrolithiasis/osteoporosis, hypophosphatemic, 1 (OMIM:612286); ORPHA:244305; Primary Fanconi renotubular syndrome (ORPHA:3337); Hereditary hypophosphatemic rickets with hypercalciuria (ORPHA:157215); ORPHA:300547
SLC34A2	Pulmonary alveolar microlithiasis (OMIM:265100); Pulmonary alveolar microlithiasis (ORPHA:60025)
SLC34A3	Hypophosphatemic rickets with hypercalciuria, hereditary (OMIM:241530); Hereditary hypophosphatemic rickets with hypercalciuria (ORPHA:157215)
SLC35A1	Congenital disorder of glycosylation, type IIf (OMIM:603585); SLC35A1-CDG (ORPHA:238459)
SLC35C1	Congenital disorder of glycosylation, type IIc (OMIM:266265); Leukocyte adhesion deficiency type II (ORPHA:99843)

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SLC35D1	Schneckenbecken dysplasia (OMIM:269250); Schneckenbecken dysplasia (ORPHA:3144)
SLC35F3	—
SLC35G2	—
SLC36A2	IMINOGLYCINURIA (OMIM:242600); Glycinuria with or without oxalate urolithiasis (OMIM:138500); Iminoglycinuria (ORPHA:42062)
SLC37A4	Congenital disorder of glycosylation, type IIw (OMIM:619525); Glycogen storage disease Ib (OMIM:232220); Glycogen storage disease Ic (OMIM:232240); Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib (ORPHA:79259)
SLC39A12	—
SLC39A13	Ehlers-Danlos syndrome, spondylodysplastic type, 3 (OMIM:612350); SLC39A13-related spondylodysplastic Ehlers-Danlos syndrome (ORPHA:157965)
SLC39A4	Acrodermatitis enteropathica, Zinc-Deficiency type (OMIM:201100); Acrodermatitis enteropathica (ORPHA:37)
SLC3A1	Cystinuria (OMIM:220100); ORPHA:238523; Hypotonia-cystinuria syndrome (ORPHA:163690); 2p21 microdeletion syndrome (ORPHA:163693); ORPHA:93612
SLC40A1	Hemochromatosis, type 4 (OMIM:606069); ORPHA:647834; ORPHA:648562
SLC41A1	Nephronophthisis-like nephropathy 2 (OMIM:619468)
SLC44A2	—
SLC45A2	SKIN/HAIR/EYE PIGMENTATION, VARIATION IN, 5 (OMIM:227240); Albinism, oculocutaneous, type IV (OMIM:606574); Oculocutaneous albinism type 4 (ORPHA:79435)
SLC46A1	Folate malabsorption, hereditary (OMIM:229050); Hereditary folate malabsorption (ORPHA:90045)
SLC47A1	—
SLC47A2	—
SLC49A4	ORPHA:422526
SLC4A1	Renal tubular acidosis, distal, autosomal dominant (OMIM:179800); Renal tubular acidosis, distal, with hemolytic anemia (OMIM:611590); Spherocytosis, type 4 (OMIM:612653); OMIM:110500; Ovalocytosis, hereditary hemolytic (OMIM:166900); Cryohydrocytosis (OMIM:185020); OMIM:112010; OMIM:112050; OMIM:611162; OMIM:601550; OMIM:601551; Dehydrated hereditary stomatocytosis (ORPHA:3202); Southeast Asian ovalocytosis (ORPHA:98868); ORPHA:398088; Hereditary spherocytosis (ORPHA:822); ORPHA:93610; ORPHA:93608
SLC4A10	Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities (OMIM:620746); ORPHA:664430
SLC4A11	Corneal endothelial dystrophy and perceptive deafness (OMIM:217400); Corneal endothelial dystrophy, autosomal recessive (OMIM:217700); Corneal dystrophy, Fuchs endothelial, 4 (OMIM:613268); Corneal dystrophy-perceptive deafness syndrome (ORPHA:1490); Congenital hereditary endothelial dystrophy type II (ORPHA:293603); Fuchs endothelial corneal dystrophy (ORPHA:98974)
SLC4A3	Short QT syndrome 7 (OMIM:620231); Congenital short QT syndrome (ORPHA:51083)
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities and mentalretardation (OMIM:604278); ORPHA:93607
SLC4A7	—
SLC52A1	Riboflavin deficiency (OMIM:615026); ORPHA:411712
SLC52A2	Brown-Vialetto-Van laere syndrome 2 (OMIM:614707); ORPHA:572543
SLC52A3	Brown-Vialetto-Van Laere syndrome 1 (OMIM:211530); Bulbar palsy, progressive, of childhood (OMIM:211500); ORPHA:572550
SLC5A1	Glucose/galactose malabsorption (OMIM:606824); Glucose-galactose malabsorption (ORPHA:35710)
SLC5A11	—
SLC5A2	Renal glucosuria (OMIM:233100); Familial renal glucosuria (ORPHA:69076)
SLC5A5	Thyroid hormonogenesis, genetic defect in, 1 (OMIM:274400); Familial thyroid dyshormonogenesis (ORPHA:95716)
SLC5A7	Neuronopathy, distal hereditary motor, type VIIA (OMIM:158580); Myasthenic syndrome, congenital, 20, presynaptic (OMIM:617143); ORPHA:139589; Presynaptic congenital myasthenic syndromes (ORPHA:98914)
SLC6A1	Myoclonic-atonic epilepsy (OMIM:616421); ORPHA:178469; Epilepsy with myoclonic-atonic seizures (ORPHA:1942)
SLC6A11	—
SLC6A12	—
SLC6A13	—
SLC6A14	Cystic fibrosis (ORPHA:586)
SLC6A18	Iminoglycinuria (ORPHA:42062)
SLC6A19	Hartnup disorder (OMIM:234500); Hartnup disease (ORPHA:2116); Iminoglycinuria (ORPHA:42062)
SLC6A2	Orthostatic intolerance (OMIM:604715); ORPHA:443236
SLC6A20	Iminoglycinuria (ORPHA:42062)
SLC6A3	OMIM:188890; Parkinsonism-Dystonia, infantile, 1 (OMIM:613135); Infantile dystonia-parkinsonism (ORPHA:238455)
SLC6A4	Obsessive-Compulsive disorder 1 (OMIM:164230); OMIM:607834
SLC6A5	Hyperekplexia 3 (OMIM:614618); Hereditary hyperekplexia (ORPHA:3197)
SLC6A6	Hypotaurinemic retinal degeneration and cardiomyopathy (OMIM:145350)
SLC6A8	Cerebral creatine deficiency syndrome 1 (OMIM:300352); X-linked creatine transporter deficiency (ORPHA:52503)
SLC6A9	Scoliosis, isolated, susceptibility to, 6 (OMIM:621428); Glycine encephalopathy with normal serum glycine (OMIM:617301); ORPHA:289863; ORPHA:289860

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SLC7A10	—
SLC7A11	—
SLC7A2	—
SLC7A5	—
SLC7A7	Lysinuric protein intolerance (OMIM:222700); Lysinuric protein intolerance (ORPHA:470)
SLC7A9	Cystinuria (OMIM:220100); ORPHA:93613
SLC9A3	Diarrhea 8, secretory sodium, congenital (OMIM:616868); Cystic fibrosis (ORPHA:586); ORPHA:103908
SLC9A3R1	—
SLC9A6	Neurodegenerative disorder, X-linked, female-restricted, with parkinsonism and cognitive impairement (OMIM:301142); Intellectual developmental disorder, X-linked syndromic, Christianson type (OMIM:300243); Christianson syndrome (ORPHA:85278)
SLC9A9	OMIM:613410
SLC9B1	—
SLCO1A2	—
SLCO1B1	Hyperbilirubinemia, Rotor type, digenic (OMIM:237450); Rotor syndrome (ORPHA:3111)
SLCO1B3	Hyperbilirubinemia, Rotor type, digenic (OMIM:237450); Rotor syndrome (ORPHA:3111)
SLCO1C1	—
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal dominant (OMIM:167100); Hypertrophic osteoarthropathy, primary, autosomal recessive 2 (OMIM:614441); Pachydermoperiostosis (ORPHA:2796); ORPHA:468641
SLCO2B1	—
SLCO5A1	—
SLFN5	—
SLIT1	—
SLIT3	—
SLITRK1	TRICHOTILLOMANIA (OMIM:613229); Gilles de la tourette syndrome (OMIM:137580)
SLITRK5	—
SLITRK6	Deafness and myopia (OMIM:221200); ORPHA:90636; ORPHA:363396
SLURP1	Meleda disease (OMIM:248300); Mal de Meleda (ORPHA:87503)
SLX4	Fanconi anemia, complementation group P (OMIM:613951); Fanconi anemia (ORPHA:84)
SLX4IP	—
SMAD1	—
SMAD2	Loeys-Dietz syndrome 6 (OMIM:619656); Congenital heart defects, multiple types, 8, with or without heterotaxy (OMIM:619657); Loeys-Dietz syndrome (ORPHA:60030); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387)
SMAD3	Loeys-Dietz syndrome 3 (OMIM:613795); Loeys-Dietz syndrome (ORPHA:60030); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); Aneurysm-osteoarthritis syndrome (ORPHA:284984)
SMAD4	Juvenile polyposis syndrome (OMIM:174900); Myhre syndrome (OMIM:139210); Pancreatic cancer (OMIM:260350); Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome (OMIM:175050); Generalized juvenile polyposis/juvenile polyposis coli (ORPHA:329971); Myhre syndrome (ORPHA:2588); Familial pancreatic carcinoma (ORPHA:1333); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); Hereditary hemorrhagic telangiectasia (ORPHA:774)
SMAD5	—
SMAD6	Craniosynostosis 7 (OMIM:617439); Radioulnar synostosis (OMIM:179300); Aortic valve disease 2 (OMIM:614823); Familial bicuspid aortic valve (ORPHA:402075)
SMAD7	Colorectal cancer, susceptibility to, 3 (OMIM:612229)
SMAD9	Pulmonary hypertension, primary, 2 (OMIM:615342); ORPHA:275777
SMAP1	—
SMARCA2	Nicolaides-Baraitser syndrome (OMIM:601358); Blepharophimosis-impaired intellectual development syndrome (OMIM:619293); Nicolaides-Baraitser syndrome (ORPHA:3051); ORPHA:637013
SMARCA4	Coffin-Siris syndrome 4 (OMIM:614609); Rhabdoid tumor predisposition syndrome 2 (OMIM:613325); Otosclerosis 12 (OMIM:620792); ORPHA:231108; Coffin-Siris syndrome (ORPHA:1465); ORPHA:370396; ORPHA:466962
SMARCAD1	Fingerprints, absence of (OMIM:136000); Huriez syndrome (OMIM:181600); Basan syndrome (OMIM:129200); Huriez syndrome (ORPHA:384); ORPHA:289465; Absence of fingerprints-congenital milia syndrome (ORPHA:1658)
SMARCAL1	Immunoosseous dysplasia, Schimke type (OMIM:242900); Schimke immuno-osseous dysplasia (ORPHA:1830)
SMARCB1	Rhabdoid tumor predisposition syndrome 1 (OMIM:609322); Schwannomatosis (OMIM:162091); Coffin-Siris syndrome 3 (OMIM:614608); ORPHA:231108; Coffin-Siris syndrome (ORPHA:1465); Atypical teratoid rhabdoid tumor (ORPHA:99966); Full schwannomatosis (ORPHA:93921); Meningioma (ORPHA:2495); ORPHA:263662
SMARCE1	Meningioma, familial, susceptibility to (OMIM:607174); Coffin-Siris syndrome 5 (OMIM:616938); Coffin-Siris syndrome (ORPHA:1465); Meningioma (ORPHA:2495); ORPHA:263662
SMC1A	Developmental and epileptic encephalopathy 85, with or without midline brain defects (OMIM:301044); Cornelia de Lange syndrome 2 (OMIM:300590); Semilobar holoprosencephaly (ORPHA:220386); Cornelia de Lange syndrome (ORPHA:199); Atypical Rett syndrome (ORPHA:3095); ORPHA:708203
SMC3	Cornelia de Lange syndrome 3 (OMIM:610759); Cornelia de Lange syndrome (ORPHA:199)
SMCHD1	Bosma arhinia microphthalmia syndrome (OMIM:603457); Facioscapulohumeral muscular dystrophy 2 (OMIM:158901); Facioscapulohumeral dystrophy (ORPHA:269); Hyposmia-nasal and ocular hypoplasia-

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	hypogonadotropic hypogonadism syndrome (ORPHA:2250)
SMG1	—
SMG7	—
SMIM27	—
SMIM3	—
SMN2	Spinal muscular atrophy, type III (OMIM:253400); ORPHA:83420; ORPHA:83418; ORPHA:83419; ORPHA:83330
SMO	Pallister-Hall-like syndrome (OMIM:241800); Basal cell carcinoma, susceptibility to, 1 (OMIM:605462); Curry-Jones syndrome (OMIM:601707); Hirschsprung disease (ORPHA:388); Curry-Jones syndrome (ORPHA:1553); Meningioma (ORPHA:2495)
SMOC1	Microphthalmia with limb anomalies (OMIM:206920); Microphthalmia with limb anomalies (ORPHA:1106)
SMOC2	Dentin dysplasia, type I, with microdontia and misshapen teeth (OMIM:125400); ORPHA:314721
SMPD1	Niemann-pick disease, type B (OMIM:607616); Niemann-Pick disease, type A (OMIM:257200); Chronic visceral acid sphingomyelinase deficiency (ORPHA:77293); ORPHA:77292
SMPD3	—
SMPX	Deafness, X-linked 4 (OMIM:300066); Myopathy, distal, 7, adult-onset, X-linked (OMIM:301075); ORPHA:90625; ORPHA:700163
SMS	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type (OMIM:309583); X-linked intellectual disability, Snyder type (ORPHA:3063)
SMUG1	—
SMYD4	—
SNAI2	Piebaldism (ORPHA:2884); Waardenburg syndrome type 2 (ORPHA:895)
SNAP25	Myasthenic syndrome, congenital, 18 (OMIM:616330); Presynaptic congenital myasthenic syndromes (ORPHA:98914)
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome (OMIM:609528); CEDNIK syndrome (ORPHA:66631)
SNAPC5	—
SNCA	Parkinson disease 4 (OMIM:605543); Dementia, lewy body (OMIM:127750); Parkinson disease 1, autosomal dominant (OMIM:168601); Young-onset Parkinson disease (ORPHA:2828); Parkinsonian-pyramidal syndrome (ORPHA:171695); Hereditary late-onset Parkinson disease (ORPHA:411602)
SNCAIP	Parkinson disease, late-onset (OMIM:168600)
SNCB	Dementia, lewy body (OMIM:127750)
SND1	—
SNHG16	—
SNHG22	—
SNHG31	—
SNHG8	—
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism (OMIM:614501)
SNRK	—
SNRNP200	Retinitis pigmentosa 33 (OMIM:610359); Retinitis pigmentosa (ORPHA:791)
SNRPN	Autism susceptibility 1 (OMIM:209850); Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2 (ORPHA:177904); Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1 (ORPHA:177901); Prader-Willi syndrome due to imprinting mutation (ORPHA:177910); Prader-Willi syndrome due to translocation (ORPHA:177907); Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15 (ORPHA:98754); Angelman syndrome due to imprinting defect in 15q11-q13 (ORPHA:411515)
SNTA1	Long QT syndrome 12 (OMIM:612955); Romano-Ward syndrome (ORPHA:101016)
SNTB1	—
SNTG2	—
SNX1	—
SNX10	Osteopetrosis, autosomal recessive 8 (OMIM:615085); Autosomal recessive malignant osteopetrosis (ORPHA:667)
SNX22	—
SNX3	—
SOBP	Impaired intellectual development, anterior maxillary protrusion, and strabismus (OMIM:613671); ORPHA:562559
SOCS3	—
SOD1	Amyotrophic lateral sclerosis 1 (OMIM:105400); Spastic tetraplegia and axial hypotonia, progressive (OMIM:618598); Amyotrophic lateral sclerosis (ORPHA:803)
SOD2	OMIM:612634
SOD3	—
SOHLH1	Ovarian dysgenesis 5 (OMIM:617690); Spermatogenic failure 32 (OMIM:618115); Male infertility with azoospermia or oligozoospermia due to single gene mutation (ORPHA:399805)
SORCS1	—
SORD2P	—
SORL1	Early-onset autosomal dominant Alzheimer disease (ORPHA:1020)
SORT1	OMIM:613589
SOS1	Fibromatosis, gingival, 1 (OMIM:135300); Noonan syndrome 4 (OMIM:610733); Hereditary gingival fibromatosis (ORPHA:2024); Noonan syndrome (ORPHA:648)
SOST	van Buchem disease (OMIM:239100); Craniodiaphyseal dysplasia, autosomal dominant (OMIM:122860);

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	Sclerosteosis 1 (OMIM:269500); Sclerosteosis (ORPHA:3152); Craniodiaphyseal dysplasia (ORPHA:1513); Hyperostosis corticalis generalisata (ORPHA:3416)
SOX10	Waardenburg syndrome, type 2E (OMIM:611584); Peripheral demyelinating neuropathy, central dysmyelination, Waardenburg syndrome, and Hirschsprung disease (OMIM:609136); Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Waardenburg syndrome, type 4C (OMIM:613266); Waardenburg-Shah syndrome (ORPHA:897); Waardenburg syndrome type 2 (ORPHA:895); Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease (ORPHA:163746); Kallmann syndrome (ORPHA:478)
SOX17	Pulmonary hypertension, primary, 7 (OMIM:621248); Vesicoureteral reflux 3 (OMIM:613674); ORPHA:289365; ORPHA:275777
SOX18	Hypotrichosis-Lymphedema-Telangiectasia-Renal defect syndrome (OMIM:137940); Hypotrichosis-lymphedema-telangiectasia syndrome (OMIM:607823); Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome (ORPHA:69735)
SOX2	Microphthalmia, syndromic 3 (OMIM:206900); Septo-optic dysplasia spectrum (ORPHA:3157); ORPHA:98938; Anophthalmia/microphthalmia-esophageal atresia syndrome (ORPHA:77298); Nanophthalmos (ORPHA:35612)
SOX3	Panhypopituitarism, X-linked (OMIM:312000); Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency (OMIM:300123); 46,XX testicular difference of sex development (ORPHA:393); Non-acquired panhypopituitarism (ORPHA:90695); Septo-optic dysplasia spectrum (ORPHA:3157); ORPHA:79495; X-linked intellectual disability with isolated growth hormone deficiency (ORPHA:67045)
SOX5	Lamb-Shaffer syndrome (OMIM:616803); Developmental and speech delay due to SOX5 deficiency (ORPHA:313892); ORPHA:313884; Large/giant congenital melanocytic nevus (ORPHA:626)
SOX6	Tolchin-Le Caignec syndrome (OMIM:618971)
SOX7	—
SOX9	46,XY sex reversal 10 (OMIM:616425); Campomelic dysplasia (OMIM:114290); 46,XX sex reversal 2 (OMIM:278850); 46,XX testicular difference of sex development (ORPHA:393); 46,XY partial gonadal dysgenesis (ORPHA:251510); 46,XY complete gonadal dysgenesis (ORPHA:242); Campomelic dysplasia (ORPHA:140); Isolated Pierre Robin sequence (ORPHA:718); 46,XX ovotesticular difference of sex development (ORPHA:2138)
SP110	Hepatic venoocclusive disease with immunodeficiency (OMIM:235550); OMIM:607948; Hepatic veno-occlusive disease-immunodeficiency syndrome (ORPHA:79124)
SP140	—
SP7	Osteogenesis imperfecta, type XII (OMIM:613849); Craniodiaphyseal dysplasia (ORPHA:1513); ORPHA:216820
SP8	—
SPACA9	—
SPAG17	Spermatogenic failure 55 (OMIM:619380); Male infertility with azoospermia or oligozoospermia due to single gene mutation (ORPHA:399805); ORPHA:276234
SPAG5	—
SPAG8	—
SPANXN5	—
SPART	Spastic paraplegia 20, autosomal recessive (OMIM:275900); Autosomal recessive spastic paraplegia type 20 (ORPHA:101000)
SPAST	Spastic paraplegia 4, autosomal dominant (OMIM:182601); Autosomal dominant spastic paraplegia type 4 (ORPHA:100985)
SPATA13	—
SPATA16	Spermatogenic failure 6 (OMIM:102530); ORPHA:171709
SPATA21	—
SPATA22	Premature ovarian failure 25 (OMIM:621002); Spermatogenic failure 96 (OMIM:621001)
SPATA31C1	—
SPATA31F1	—
SPATA6L	—
SPATA7	Leber congenital amaurosis-3 (LCA3)/Retinitis pigmentosa, juvenile, autosomal recessive (OMIM:604232); Leber congenital amaurosis (ORPHA:65); Retinitis pigmentosa (ORPHA:791); Severe early-childhood-onset retinal dystrophy (ORPHA:364055)
SPC25	—
SPECC1	—
SPECC1L	Teebi hypertelorism syndrome 1 (OMIM:145420); Facial clefting, oblique, 1 (OMIM:600251); SPECC1L-related hypertelorism syndrome (ORPHA:1519); ORPHA:141258; ORPHA:141276
SPEF2	Spermatogenic failure 43 (OMIM:618751); Primary ciliary dyskinesia (ORPHA:244); ORPHA:276234
SPG11	Spastic paraplegia 11, autosomal recessive (OMIM:604360); Amyotrophic lateral sclerosis 5, juvenile (OMIM:602099); Charcot-Marie-Tooth disease, axonal, type 2X (OMIM:616668); Autosomal recessive spastic paraplegia type 11 (ORPHA:2822); Juvenile amyotrophic lateral sclerosis (ORPHA:300605); ORPHA:466775
SPG21	Mast syndrome (OMIM:248900); Autosomal recessive spastic paraplegia type 21 (ORPHA:101001)
SPG7	Spastic paraplegia 7, autosomal recessive (OMIM:607259); Spastic paraplegia type 7 (ORPHA:99013); Primary lateral sclerosis (ORPHA:35689)
SPI1	Agammaglobulinemia 10, autosomal dominant (OMIM:619707); Autosomal non-syndromic agammaglobulinemia (ORPHA:33110)
SPINK1	Pancreatitis, hereditary (OMIM:167800); Tropical calcific pancreatitis (OMIM:608189); ORPHA:700124;

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	Tropical pancreatitis (ORPHA:103918)
SPINK5	Netherton syndrome (OMIM:256500); Netherton syndrome (ORPHA:634)
SPINT2	Diarrhea 3, secretory sodium, congenital, with or without other congenital anomalies (OMIM:270420); ORPHA:563708
SPNS1	—
SPP1	Systemic lupus erythematosus (ORPHA:536); Pediatric systemic lupus erythematosus (ORPHA:93552)
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency (OMIM:612716); Dopa-responsive dystonia due to sepiapterin reductase deficiency (ORPHA:70594)
SPRED1	Legius syndrome (OMIM:611431); Legius syndrome (ORPHA:137605)
SPRED2	Noonan syndrome 14 (OMIM:619745); Noonan syndrome (ORPHA:648)
SPRN	—
SPRY2	Iga nephropathy, susceptibility to, 3 (OMIM:616818)
SPRY4	Hypogonadotropic hypogonadism 17 with or without anosmia (OMIM:615266); Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); ORPHA:363494; Kallmann syndrome (ORPHA:478); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432)
SPSB1	—
SPSB3	—
SPTA1	Spherocytosis, autosomal recessive (OMIM:270970); Elliptocytosis 2 (OMIM:130600); Pyropoikilocytosis, hereditary (OMIM:266140); Hereditary elliptocytosis (ORPHA:288); Hereditary spherocytosis (ORPHA:822)
SPTAN1	Epileptic encephalopathy, early infantile, 5 (OMIM:613477); Neuronopathy, distal hereditary motor, autosomal dominant 11 (OMIM:620528); Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia (OMIM:620538); Developmental delay with or without epilepsy (OMIM:620540); Infantile epileptic spasms syndrome (ORPHA:697160)
SPTB	Elliptocytosis-3 (OMIM:617948); Spherocytosis, type 2 (OMIM:616649); Hereditary elliptocytosis (ORPHA:288); Hereditary spherocytosis (ORPHA:822)
SPTBN1	Developmental delay, impaired speech, and behavioral abnormalities (OMIM:619475); ORPHA:528084
SPTBN2	Spinocerebellar ataxia, autosomal recessive 14 (OMIM:615386); Spinocerebellar ataxia 5 (OMIM:600224); Spectrin-associated autosomal recessive cerebellar ataxia (ORPHA:352403); Spinocerebellar ataxia type 5 (ORPHA:98766)
SPTBN5	—
SPTLC1	Amyotrophic lateral sclerosis 27, juvenile (OMIM:620285); Neuropathy, hereditary sensory and autonomic, type IA (OMIM:162400); Hereditary sensory and autonomic neuropathy type 1 (ORPHA:36386); Juvenile amyotrophic lateral sclerosis (ORPHA:300605)
SPTLC2	Neuropathy, hereditary sensory and autonomic, type IC (OMIM:613640); Hereditary sensory and autonomic neuropathy type 1 (ORPHA:36386)
SPX	—
SQSTM1	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3 (OMIM:616437); Paget disease of bone 3 (OMIM:167250); Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset (OMIM:617145); Myopathy, distal, with rimmed vacuoles (OMIM:617158); Behavioral variant of frontotemporal dementia (ORPHA:275864); Frontotemporal dementia with motor neuron disease (ORPHA:275872); Amyotrophic lateral sclerosis (ORPHA:803); Distal myopathy, Welander type (ORPHA:603)
SRC	Thrombocytopenia 6 (OMIM:616937); Colorectal cancer (OMIM:114500); ORPHA:480851
SRCAP	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities (OMIM:619595); Floating-Harbor syndrome (OMIM:136140); Floating-Harbor syndrome (ORPHA:2044); ORPHA:528084
SRD5A2	Pseudovaginal perineoscrotal hypospadias (OMIM:264600); ORPHA:1331; 46,XY difference of sex development due to 5-alpha-reductase 2 deficiency (ORPHA:753)
SRD5A3	Kahrizi syndrome (OMIM:612713); Congenital disorder of glycosylation, type IQ (OMIM:612379); SRD5A3-CDG (ORPHA:324737)
SREBF1	Mucoepithelial dysplasia, hereditary (OMIM:158310); Ichthyosis, follicular, with atrichia and photophobia syndrome 2 (OMIM:619016); Hirschsprung disease (ORPHA:388)
SREBF2	—
SRFBP1	—
SRGAP3	ORPHA:251615
SRI	—
SRP72	Bone marrow failure syndrome 1 (OMIM:614675); ORPHA:314399
SRPX	—
SRPX2	Rolandic epilepsy, impaired intellectual development, and speech dyspraxia (OMIM:300643); Bilateral perisylvian polymicrogyria (ORPHA:98889); Rolandic epilepsy-speech dyspraxia syndrome (ORPHA:163721); Self-limited epilepsy with centrotemporal spikes (ORPHA:1945)
SRR	—
SRY	46XY sex reversal 1 (OMIM:400044); 46XX sex reversal 1 (OMIM:400045); 46,XX testicular difference of sex development (ORPHA:393); 46,XY partial gonadal dysgenesis (ORPHA:251510); 46,XY complete gonadal dysgenesis (ORPHA:242); 46,XX ovotesticular difference of sex development (ORPHA:2138); 45,X/46,XY mixed gonadal dysgenesis (ORPHA:1772)
SSBP1	Optic atrophy with negative electroretinograms (OMIM:165510); ORPHA:90635
SSH1	—
SSPN	—
SSTR5	—
SSUH2	OMIM:621559; ORPHA:99789

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
SSX7	—
ST14	Ichthyosis, congenital, autosomal recessive 11 (OMIM:602400); Ichthyosis-hypotrichosis syndrome (ORPHA:91132)
ST20	—
ST3GAL1	—
ST3GAL2	—
ST3GAL3	Developmental and epileptic encephalopathy 15 (OMIM:615006); Intellectual developmental disorder, autosomal recessive 12 (OMIM:611090); ORPHA:697734
ST3GAL4	—
ST3GAL5	Salt and pepper developmental regression syndrome (OMIM:609056); ORPHA:370933
ST3GAL6	—
ST6GAL1	—
ST6GAL2	—
ST6GALNAC1	—
ST6GALNAC2	—
ST6GALNAC3	—
ST6GALNAC4	—
ST6GALNAC5	—
ST6GALNAC6	—
ST8SIA1	—
ST8SIA2	—
ST8SIA3	—
ST8SIA4	—
ST8SIA5	—
ST8SIA6	—
STAG1	Intellectual developmental disorder, autosomal dominant 47 (OMIM:617635); STAG1-related intellectual disability-facial dysmorphism-gastroesophageal reflux syndrome (ORPHA:502434)
STAG2	Holoprosencephaly 13, X-linked (OMIM:301043); Neurodevelopmental disorder, X-linked, with craniofacial abnormalities (OMIM:301022); Semilobar holoprosencephaly (ORPHA:220386); Alobar holoprosencephaly (ORPHA:93925); Xq25 microduplication syndrome (ORPHA:521258)
STAMBPL1	—
STAR	Lipoid congenital adrenal hyperplasia (OMIM:201710); ORPHA:325529; ORPHA:325524; Familial glucocorticoid deficiency (ORPHA:361)
STARD9	—
STAT1	Immunodeficiency 31C (OMIM:614162); Mycobacterial and viral infections, susceptibility to, autosomal recessive (OMIM:613796); Immunodeficiency 31A (OMIM:614892); ORPHA:391311; STAT1-related autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome (ORPHA:391487); ORPHA:319595
STAT3	Autoimmune disease, multisystem, infantile-onset, 1 (OMIM:615952); Hyper-IgE recurrent infection syndrome (OMIM:147060); Isolated permanent neonatal diabetes mellitus (ORPHA:99885); ORPHA:86872; ORPHA:512017; ORPHA:667662; Autosomal dominant hyper-IgE syndrome due to STAT3 deficiency (ORPHA:2314); Acute promyelocytic leukemia (ORPHA:520); ORPHA:438159
STAT4	Disabling pansclerotic morphea of childhood (OMIM:620443); OMIM:612253; Behçet disease (ORPHA:117); Oligoarticular juvenile idiopathic arthritis (ORPHA:85410); Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis (ORPHA:85408); Systemic lupus erythematosus (ORPHA:536); Pediatric systemic lupus erythematosus (ORPHA:93552)
STAT5B	Growth hormone insensitivity with immune dysregulation 2, autosomal dominant (OMIM:618985); Growth hormone insensitivity with immune dysregulation 1, autosomal recessive (OMIM:245590); ORPHA:220465; Acute promyelocytic leukemia (ORPHA:520)
STAT6	Hyper-IgE syndrome 6, autosomal dominant, with recurrent infections (OMIM:620532); Solitary fibrous tumor (ORPHA:2126)
STEAP3	Anemia, hypochromic microcytic, with iron overload 2 (OMIM:615234); Severe congenital hypochromic anemia with ringed sideroblasts (ORPHA:300298)
STIL	Microcephaly 7, primary, autosomal recessive (OMIM:612703); Septopreoptic holoprosencephaly (ORPHA:280195); ORPHA:99861; Semilobar holoprosencephaly (ORPHA:220386); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925); Autosomal recessive primary microcephaly (ORPHA:2512)
STIM1	Immunodeficiency 10 (OMIM:612783); Myopathy, tubular aggregate, 1 (OMIM:160565); Stormorken syndrome (OMIM:185070); Stormorken-Sjaastad-Langslet syndrome (ORPHA:3204); Tubular aggregate myopathy (ORPHA:2593); ORPHA:317430
STING1	STING-associated vasculopathy, infantile-onset (OMIM:615934); ORPHA:425120; ORPHA:481662
STK11	Testicular tumor, somatic (OMIM:273300); Pancreatic cancer (OMIM:260350); Peutz-Jeghers syndrome (OMIM:175200); Melanoma, cutaneous malignant (OMIM:155600); Peutz-Jeghers syndrome (ORPHA:2869)
STK11IP	—
STK19	—
STK35	—
STK36	Ciliary dyskinesia, primary, 46 (OMIM:619436); Primary ciliary dyskinesia (ORPHA:244)
STK39	—
STK4	Immunodeficiency-110 with lymphoproliferation (OMIM:614868); ORPHA:314689

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
STON1	—
STOX1	Preeclampsia/eclampsia 4 (OMIM:609404); Preeclampsia (ORPHA:275555)
STRA6	Microphthalmia, syndromic 9 (OMIM:601186); ORPHA:98938; Matthew-Wood syndrome (ORPHA:2470)
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy (OMIM:611087); Polyhydramnios-megalencephaly-symptomatic epilepsy syndrome (ORPHA:500533)
STRC	Spermatogenic failure 7 (OMIM:612997); Deafness, autosomal recessive 16 (OMIM:603720); Deafness, sensorineural, and male infertility (OMIM:611102); ORPHA:90636; Deafness-infertility syndrome (ORPHA:94064)
STRIP1	—
STS	Ichthyosis, X-linked (OMIM:308100); Syndromic recessive X-linked ichthyosis (ORPHA:281090); Recessive X-linked ichthyosis (ORPHA:461)
STT3A	Congenital disorder of glycosylation, type Iw (OMIM:615596); Congenital disorder of glycosylation, type Iw, autosomal dominant (OMIM:619714); STT3A-CDG (ORPHA:370921)
STT3B	Congenital disorder of glycosylation, type Ix (OMIM:615597); STT3B-CDG (ORPHA:370924)
STX11	Hemophagocytic lymphohistiocytosis, familial, 4 (OMIM:603552); Familial hemophagocytic lymphohistiocytosis (ORPHA:540)
STX16	Pseudohypoparathyroidism, type IB (OMIM:603233); Pseudohypoparathyroidism type 1B (ORPHA:94089)
STXBP1	Developmental and epileptic encephalopathy 4 (OMIM:612164); STXBP1-related encephalopathy (ORPHA:599373); 9q33.3q34.11 microdeletion syndrome (ORPHA:495818)
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5 (OMIM:613101); Familial hemophagocytic lymphohistiocytosis (ORPHA:540)
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria) (OMIM:612073); Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria (ORPHA:1933)
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria) (OMIM:245400); Fatal infantile lactic acidosis with methylmalonic aciduria (ORPHA:17)
SUCO	—
SUFU	Joubert syndrome 32 (OMIM:617757); Meningioma, familial, susceptibility to (OMIM:607174); Basal cell nevus syndrome 2 (OMIM:620343); MEDULLOBLASTOMA (OMIM:155255); Gorlin syndrome (ORPHA:377); Microform holoprosencephaly (ORPHA:280200); ORPHA:251858; ORPHA:251863; Meningioma (ORPHA:2495); Isolated Joubert syndrome (ORPHA:475); ORPHA:263662
SUGCT	Glutaric aciduria III (OMIM:231690); Glutaric acidemia type 3 (ORPHA:35706)
SUGT1P3	—
SULF1	—
SULT1A1	—
SULT1C2	—
SULT2A1	—
SULT2B1	Ichthyosis, congenital, autosomal recessive 14 (OMIM:617571); Lamellar ichthyosis (ORPHA:313); Congenital ichthyosiform erythroderma (ORPHA:79394)
SULT4A1	—
SUMF1	Multiple sulfatase deficiency (OMIM:272200); Multiple sulfatase deficiency (ORPHA:585)
SUMO1	Orofacial cleft 10 (OMIM:613705); Oligodontia (ORPHA:99798)
SUMO2P10	—
SUMO4	OMIM:600320
SUOX	SULFOCYSTEINURIA (OMIM:272300); ORPHA:99731
SUPT16H	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum (OMIM:619480); 14q11.2 microduplication syndrome (ORPHA:261229)
SUPT3H	—
SURF1	Charcot-Marie-Tooth disease, type 4K (OMIM:616684); Mitochondrial complex IV deficiency, nuclear type 1 (OMIM:220110); ORPHA:391351
SUV39H2	—
SUZ12	Imagawa-Matsumoto syndrome (OMIM:618786); ORPHA:213711; ORPHA:659463; Weaver syndrome (ORPHA:3447)
SV2B	—
SYCE2	—
SYCP3	Spermatogenic failure 4 (OMIM:270960); Male infertility with azoospermia or oligozoospermia due to single gene mutation (ORPHA:399805)
SYK	Immunodeficiency 82 with systemic inflammation (OMIM:619381); ORPHA:695807
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders (OMIM:300491); Intellectual developmental disorder, X-linked 50 (OMIM:300115); X-linked epilepsy-learning disabilities-behavior disorders syndrome (ORPHA:85294)
SYN2	Schizophrenia (OMIM:181500)
SYN3	—
SYNE1	Emery-Dreifuss muscular dystrophy 4, autosomal dominant (OMIM:612998); Spinocerebellar ataxia, autosomal recessive 8 (OMIM:610743); Arthrogryposis multiplex congenita 3, myogenic type (OMIM:618484); Autosomal recessive ataxia, Beauce type (ORPHA:88644); Autosomal dominant Emery-Dreifuss muscular dystrophy (ORPHA:98853); Autosomal recessive myogenic arthrogryposis multiplex congenita (ORPHA:319332)
SYNE2	Emery-Dreifuss muscular dystrophy 5, autosomal dominant (OMIM:612999); Autosomal dominant Emery-Dreifuss muscular dystrophy (ORPHA:98853)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
SYNGR1	—
SYNM	—
SYNPO	—
SYNPR	—
SYP	Intellectual developmental disorder, X-linked 96 (OMIM:300802); ORPHA:777
SYT14	Spinocerebellar ataxia, autosomal recessive 11 (OMIM:614229); Autosomal recessive cerebellar ataxia-psychomotor delay syndrome (ORPHA:284271)
SYT17	—
SYT2	Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive (OMIM:619461); Myasthenic syndrome, congenital, 7, presynaptic (OMIM:616040); Presynaptic congenital myasthenic syndromes (ORPHA:98914)
SYTL3	—
SYTL5	—
TAAR1	—
TAAR9	—
TAB2	Congenital heart defects, multiple types, 2 (OMIM:614980); ORPHA:664404; ORPHA:664401
TAC3	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Hypogonadotropic hypogonadism 10 with or without anosmia (OMIM:614839); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432)
TACO1	Mitochondrial complex IV deficiency, nuclear type 8 (OMIM:619052); Mitochondrial complex IV deficiency, nuclear type 1 (OMIM:220110)
TACR3	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Hypogonadotropic hypogonadism 11 with or without anosmia (OMIM:614840); Kallmann syndrome (ORPHA:478); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432)
TACSTD2	Corneal dystrophy, gelatinous drop-like (OMIM:204870); Gelatinous drop-like corneal dystrophy (ORPHA:98957)
TAF1	Intellectual developmental disorder, X-linked syndromic 33 (OMIM:300966); Dystonia 3, torsion, X-linked (OMIM:314250); X-linked intellectual disability-global development delay-facial dysmorphism-sacral caudal remnant syndrome (ORPHA:480907); X-linked dystonia-parkinsonism (ORPHA:53351)
TAF10	—
TAF15	ORPHA:209916; Amyotrophic lateral sclerosis (ORPHA:803)
TAF1C	—
TAF1L	—
TAF2	Neurodevelopmental disorder with feeding difficulties, thin corpus callosum, and foot deformity (OMIM:615599); Microcephaly-thin corpus callosum-intellectual disability syndrome (ORPHA:397951)
TAF6	Alazami-Yuan syndrome (OMIM:617126); ORPHA:694946
TAFA2	—
TAFAZZIN	Barth syndrome (OMIM:302060); Familial isolated dilated cardiomyopathy (ORPHA:154); Barth syndrome (ORPHA:111)
TAL1	Leukemia, acute lymphocytic, susceptibility to, 1 (OMIM:613065); ORPHA:99861
TAL2	Leukemia, acute lymphocytic, susceptibility to, 1 (OMIM:613065)
TALDO1	Transaldolase deficiency (OMIM:606003); Transaldolase deficiency (ORPHA:101028)
TAP2	MHC class I deficiency 2 (OMIM:620813); ORPHA:34592
TARDBP	Amyotrophic lateral sclerosis 10 with or without frontotemporal dementia (OMIM:612069); ORPHA:700154; Frontotemporal dementia with motor neuron disease (ORPHA:275872); Amyotrophic lateral sclerosis (ORPHA:803)
TARID	—
TAS1R1	—
TAS1R3	—
TAS2R14	—
TAS2R16	OMIM:617956; Alcohol dependence (OMIM:103780)
TAS2R3	—
TAS2R38	THIOUREA TASTINGPHENYLTHIOCARBAMIDE TASTING, INCLUDED (OMIM:171200)
TAS2R46	—
TASOR	—
TAT	Tyrosine transaminase deficiency (OMIM:276600); Tyrosinemia type 2 (ORPHA:28378)
TATDN1	—
TAX1BP3	—
TBC1D23	Pontocerebellar hypoplasia, type 11 (OMIM:617695)
TBC1D24	Epilepsy, rolandic, with paroxysmal exercise-induced dystonia and writer's cramp (OMIM:608105); Doors syndrome (OMIM:220500); Developmental and epileptic encephalopathy 16 (OMIM:615338); Myoclonic epilepsy, familial infantile (OMIM:605021); Deafness, autosomal recessive 86 (OMIM:614617); Deafness, autosomal dominant 65 (OMIM:616044); ORPHA:90635; ORPHA:90636; DOORS syndrome (ORPHA:79500); Progressive myoclonic epilepsy with dystonia (ORPHA:352596); ORPHA:352587; Familial infantile myoclonic epilepsy (ORPHA:352582); Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome (ORPHA:163727); Epilepsy of infancy with migrating focal seizures (ORPHA:293181)
TBC1D4	OMIM:616087
TBC1D9	—
TBCAP3	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum (OMIM:617193); Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome (ORPHA:496641)
TBCE	Kenny-caffey syndrome, type 1 (OMIM:244460); Hypoparathyroidism-retardation-dysmorphism syndrome (OMIM:241410); Encephalopathy, progressive, with amyotrophy and optic atrophy (OMIM:617207); Sanjad-Sakati syndrome (ORPHA:2323); Autosomal recessive Kenny-Caffey syndrome (ORPHA:93324); Early-onset progressive encephalopathy-spastic ataxia-distal spinal muscular atrophy syndrome (ORPHA:496756)
TBCEL	—
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3 (OMIM:616900); TBCK-related encephalopathy-severe hypotonia-craniofacial dysmorphism syndrome (ORPHA:488632)
TBK1	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4 (OMIM:616439); Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8 (OMIM:617900); Autoinflammation with arthritis and vasculitis (OMIM:620880); Frontotemporal dementia with motor neuron disease (ORPHA:275872); Amyotrophic lateral sclerosis (ORPHA:803); Herpes simplex virus encephalitis (ORPHA:1930)
TBL1X	Hypothyroidism, congenital, nongoitrous, 8 (OMIM:301033)
TBL1XR1	Pierpont syndrome (OMIM:602342); Intellectual developmental disorder, autosomal dominant 41 (OMIM:616944); Pierpont syndrome (ORPHA:487825); Acute promyelocytic leukemia (ORPHA:520)
TBL1Y	DEAFNESS, Y-LINKED 2 (OMIM:400047)
TBP	Spinocerebellar ataxia 17 (OMIM:607136); Parkinson disease, late-onset (OMIM:168600); Spinocerebellar ataxia type 17 (ORPHA:98759)
TBX1	Conotruncal heart malformations (OMIM:217095); Digeorge syndrome (OMIM:188400); Tetralogy of Fallot (OMIM:187500); Velocardiofacial syndrome (OMIM:192430); ORPHA:685017; Tetralogy of Fallot (ORPHA:3303); ORPHA:665058; ORPHA:665044; 22q11.2 deletion syndrome (ORPHA:567); 22q11.2 duplication syndrome (ORPHA:1727)
TBX10	—
TBX15	Cousin syndrome (OMIM:260660); Pelviscapular dysplasia (ORPHA:93333)
TBX19	Acth deficiency, isolated (OMIM:201400); Congenital isolated ACTH deficiency (ORPHA:199296)
TBX2	Vertebral anomalies and variable endocrine and T-cell dysfunction (OMIM:618223)
TBX20	Atrial septal defect 4 (OMIM:611363); ORPHA:54260; Atrial septal defect, ostium secundum type (ORPHA:99103)
TBX21	Immunodeficiency 88 (OMIM:619630); Asthma, nasal polyps, and aspirin intolerance (OMIM:208550)
TBX22	Cleft palate, X-linked (OMIM:303400); Charge-Like syndrome, X-linked (OMIM:302905); Abruzzo-Erickson syndrome (ORPHA:921); ORPHA:324601
TBX3	Ulnar-Mammary syndrome (OMIM:181450); Ulnar-mammary syndrome (ORPHA:3138)
TBX4	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension (OMIM:147891); Amelia, autosomal recessive (OMIM:601360); Familial clubfoot due to 17q23.1q23.2 microduplication (ORPHA:238578); Coxopodopatellar syndrome (ORPHA:1509); ORPHA:275777; Tetraamelia-multiple malformations syndrome (ORPHA:3301); 17q23.1q23.2 microdeletion syndrome (ORPHA:261279)
TBX5	Holt-Oram syndrome (OMIM:142900); Holt-Oram syndrome (ORPHA:392); Romano-Ward syndrome (ORPHA:101016)
TBX6	Spondylocostal dysostosis 5 (OMIM:122600); Autosomal recessive spondylocostal dysostosis (ORPHA:2311); Autosomal dominant spondylocostal dysostosis (ORPHA:1797)
TBXA2R	Bleeding disorder, platelet-type, 13, susceptibility to (OMIM:614009); ORPHA:220443
TBXAS1	Ghosal hematodiaphyseal syndrome (OMIM:231095); Ghosal hematodiaphyseal dysplasia (ORPHA:1802)
TBXT	Sacral agenesis with vertebral anomalies (OMIM:615709); Neural tube defects, susceptibility to (OMIM:182940); ORPHA:178; ORPHA:397927
TCAP	Cardiomyopathy, familial hypertrophic, 25 (OMIM:607487); Muscular dystrophy, limb-girdle, type 2G (OMIM:601954); ORPHA:34514; Familial isolated dilated cardiomyopathy (ORPHA:154)
TCF19	—
TCF21	—
TCF3	Agammaglobulinemia 8, autosomal dominant (OMIM:616941); Agammaglobulinemia 8B, autosomal recessive (OMIM:619824); ORPHA:641375; ORPHA:585956; Autosomal non-syndromic agammaglobulinemia (ORPHA:33110)
TCF4	Corneal dystrophy, fuchs endothelial, 3 (OMIM:613267); Pitt-Hopkins syndrome (OMIM:610954); ORPHA:178469; Pitt-Hopkins syndrome (ORPHA:2896); Primary sclerosing cholangitis (ORPHA:171); Fuchs endothelial corneal dystrophy (ORPHA:98974)
TCF7	—
TCF7L1	—
TCF7L2	Diabetes mellitus, noninsulin-dependent (OMIM:125853); ORPHA:528084
TCIRG1	Osteopetrosis, autosomal recessive 1 (OMIM:259700); Intermediate osteopetrosis (ORPHA:210110); Autosomal recessive malignant osteopetrosis (ORPHA:667); Autosomal dominant severe congenital neutropenia (ORPHA:486); Dysosteosclerosis (ORPHA:1782)
TCN1	—
TCN2	Transcobalamin II deficiency (OMIM:275350); Transcobalamin deficiency (ORPHA:859)
TCOF1	Treacher Collins syndrome 1 (OMIM:154500); Treacher-Collins syndrome (ORPHA:861)
TCP1	Intellectual developmental disorder with polymicrogyria and seizures (OMIM:621021)
TCTE1	—
TCTN1	Joubert syndrome 13 (OMIM:614173); Meckel syndrome (ORPHA:564); Isolated Joubert syndrome (ORPHA:475)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
TCTN2	Joubert syndrome 24 (OMIM:616654); Meckel syndrome, type 8 (OMIM:613885); Meckel syndrome (ORPHA:564); Isolated Joubert syndrome (ORPHA:475)
TCTN3	Joubert syndrome 18 (OMIM:614815); Orofaciodigital syndrome IV (OMIM:258860); Orofaciodigital syndrome type 4 (ORPHA:2753); Orofaciodigital syndrome type 6 (ORPHA:2754); Meckel syndrome (ORPHA:564); Isolated Joubert syndrome (ORPHA:475)
TDO2	Hypertryptophanemia (OMIM:600627); Hypertryptophanemia (ORPHA:2224)
TDP1	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1 (OMIM:607250); Spinocerebellar ataxia with axonal neuropathy type 1 (ORPHA:94124)
TDRD7	Cataract, autosomal recessive congenital 4 (OMIM:613887)
TEAD1	Sveinsson chorioretinal atrophy (OMIM:108985); ORPHA:86813
TEC	—
TECPR2	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay (OMIM:615031); Hereditary sensory and autonomic neuropathy due to TECPR2 mutation (ORPHA:320385)
TECR	Intellectual developmental disorder, autosomal recessive 14 (OMIM:614020); ORPHA:88616
TECTA	Deafness, autosomal recessive 21 (OMIM:603629); Deafness, autosomal dominant nonsyndromic sensorineural 12 (OMIM:601543); ORPHA:90635; ORPHA:90636
TEK	Glaucoma 3, primary infantile, B (OMIM:600975); Venous malformations, multiple cutaneous and mucosal (OMIM:600195); Glaucoma 3, primary congenital, E (OMIM:617272); ORPHA:714806; Blue rubber bleb nevus (ORPHA:1059); Congenital glaucoma (ORPHA:98976); ORPHA:2451
TEKT2	—
TENM2	—
TENM4	Tremor, hereditary essential, 5 (OMIM:616736)
TERT	Pulmonary fibrosis, idiopathic (OMIM:178500); Melanoma, cutaneous malignant, susceptibility to, 9 (OMIM:615134); Aplastic anemia (OMIM:609135); Dyskeratosis congenita, autosomal dominant 2 (OMIM:613989); Dyskeratosis congenita, autosomal dominant, 1 (OMIM:127550); Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1 (OMIM:614742); Leukemia, acute myeloid (OMIM:601626); Idiopathic aplastic anemia (ORPHA:88); Adrenocortical carcinoma (ORPHA:1501); Idiopathic pulmonary fibrosis (ORPHA:2032); Hoyeraal-Hreidarsson syndrome (ORPHA:3322); ORPHA:146; Familial melanoma (ORPHA:618); Meningioma (ORPHA:2495); ORPHA:457246; Dyskeratosis congenita (ORPHA:1775)
TESHL	—
TET1	—
TET2	Myelodysplastic syndrome (OMIM:614286); Immunodeficiency 75 (OMIM:619126); Essential thrombocythemia (ORPHA:3318); Aggressive systemic mastocytosis (ORPHA:98850); Systemic mastocytosis with associated hematologic neoplasm (ORPHA:98849); ORPHA:86845; Myelodysplastic neoplasm with low blasts (ORPHA:98826); Primary myelofibrosis (ORPHA:824); Acquired idiopathic sideroblastic anemia (ORPHA:75564); ORPHA:100020; ORPHA:100019; ORPHA:664729
TEX14	Spermatogenic failure 23 (OMIM:617707); Male infertility with azoospermia or oligozoospermia due to single gene mutation (ORPHA:399805)
TEX22	—
TF	ATRANSFERRINEMIA (OMIM:209300); Congenital atransferrinemia (ORPHA:1195)
TFAM	Mitochondrial DNA depletion syndrome 15 (hepatocerebral type) (OMIM:617156)
TFAP2A	Branchiooculofacial syndrome (OMIM:113620); Branchio-oculo-facial syndrome (ORPHA:1297)
TFAP2B	Char syndrome (OMIM:169100); Patent ductus arteriosus 2 (OMIM:617035); Char syndrome (ORPHA:46627); ORPHA:466729
TFB1M	ORPHA:90641
TFE3	Intellectual developmental disorder, X-linked, syndromic, with pigmentary mosaicism and coarse facies (OMIM:301066); Renal cell carcinoma, xp11-associated (OMIM:300854); ORPHA:319308; ORPHA:163699; ORPHA:157791
TFF1	—
TFG	Neuropathy, hereditary motor and sensory, Okinawa type (OMIM:604484); Spastic paraplegia 57, autosomal recessive (OMIM:615658); ORPHA:209916; ORPHA:146; Hereditary motor and sensory neuropathy, Okinawa type (ORPHA:90117); Autosomal recessive spastic paraplegia type 57 (ORPHA:431329); ORPHA:435819
TFPI	—
TFR2	Hemochromatosis, type 3 (OMIM:604250); ORPHA:225123; ORPHA:648581
TFRC	Immunodeficiency 46 (OMIM:616740); ORPHA:476113
TG	Thyroid dyshormonogenesis 3 (OMIM:274700); Autoimmune thyroid disease, susceptibility to, 3 (OMIM:608175); Familial thyroid dyshormonogenesis (ORPHA:95716)
TGFB1	Cystic fibrosis (OMIM:219700); Inflammatory bowel disease, immunodeficiency, and encephalopathy (OMIM:618213); Camurati-Engelmann disease (OMIM:131300); Camurati-Engelmann disease (ORPHA:1328); Cystic fibrosis (ORPHA:586); ORPHA:565788
TGFB2	Loeys-Dietz syndrome 4 (OMIM:614816); Camurati-engelmann disease, type 2 (OMIM:606631); Loeys-Dietz syndrome (ORPHA:60030); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387)
TGFB3	Loeys-Dietz syndrome 5 (OMIM:615582); Arrhythmogenic right ventricular dysplasia 1 (OMIM:107970); Loeys-Dietz syndrome (ORPHA:60030); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); ORPHA:293888; ORPHA:293899; ORPHA:293910
TGFBI	Corneal dystrophy, lattice type I (OMIM:122200); Corneal dystrophy, Reis-Bucklers type (OMIM:608470); Corneal dystrophy, lattice type IIIA (OMIM:608471); Corneal dystrophy of bowman layer, type II (OMIM:602082); Corneal dystrophy, Avellino type (OMIM:607541); Corneal dystrophy, epithelial basement membrane (OMIM:121820); Groenouw type I corneal dystrophy (OMIM:121900); ORPHA:98961; Granular

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	corneal dystrophy type I (ORPHA:98962); Granular corneal dystrophy type II (ORPHA:98963); Lattice corneal dystrophy type I (ORPHA:98964); Thiel-Behnke corneal dystrophy (ORPHA:98960); ORPHA:98956
TGFBR1	Loeys-Dietz syndrome 1 (OMIM:609192); Multiple self-healing squamous epithelioma, susceptibility to (OMIM:132800); ORPHA:65748; Loeys-Dietz syndrome (ORPHA:60030); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); ORPHA:284973
TGFBR2	Loeys-Dietz syndrome 2 (OMIM:610168); Colorectal cancer, hereditary nonpolyposis, type 6 (OMIM:614331); Esophageal cancer, somatic (OMIM:133239); Squamous cell carcinoma of the esophagus (ORPHA:99977); Loeys-Dietz syndrome (ORPHA:60030); Lynch syndrome (ORPHA:144); Familial thoracic aortic aneurysm and aortic dissection (ORPHA:91387); ORPHA:284973
TGFBR3	Familial cerebral saccular aneurysm (ORPHA:231160)
TGIF1	Holoprosencephaly 4 (OMIM:142946); Septopreoptic holoprosencephaly (ORPHA:280195); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925)
TGM1	Ichthyosis, congenital, autosomal recessive 1 (OMIM:242300); Lamellar ichthyosis (ORPHA:313); Congenital ichthyosiform erythroderma (ORPHA:79394); Self-improving collodion baby (ORPHA:281122); Acral self-healing collodion baby (ORPHA:281127); Bathing suit ichthyosis (ORPHA:100976)
TGM2	—
TGM5	Peeling skin syndrome 2 (OMIM:609796); Acral peeling skin syndrome (ORPHA:263534)
TGM6	Spinocerebellar ataxia 35 (OMIM:613908); Spinocerebellar ataxia type 35 (ORPHA:276193); ORPHA:319465
TH	Segawa syndrome, autosomal recessive (OMIM:605407); Autosomal recessive dopa-responsive dystonia (ORPHA:101150)
TH2LCRR	—
THAP1	Dystonia 6, torsion (OMIM:602629); Primary dystonia, DYT6 type (ORPHA:98806)
THAP6	—
THBD	Hemolytic uremic syndrome, atypical, susceptibility to, 6 (OMIM:612926); Thrombophilia due to thrombomodulin defect (OMIM:614486); ORPHA:544472; ORPHA:436169
THBS1	—
THBS2	OMIM:603932; Ehlers-Danlos syndrome, classic-like, 3 (OMIM:620865)
THOC2	Intellectual developmental disorder, X-linked syndromic, Kumar type (OMIM:300957); Arthrogryposis multiplex congenita 7, X-linked (OMIM:301127); X-linked intellectual disability-short stature-overweight syndrome (ORPHA:457240)
THPO	Thrombocytopenia 9 (OMIM:620478); Amegakaryocytic thrombocytopenia, congenital, 2 (OMIM:620481); Thrombocythemia 1 (OMIM:187950); Familial thrombocytosis (ORPHA:71493); Congenital amegakaryocytic thrombocytopenia (ORPHA:3319); ORPHA:329319; ORPHA:397692
THRA	Hypothyroidism, congenital, nongoitrous, 6 (OMIM:614450); ORPHA:566231
THRB	Thyroid hormone resistance, generalized, autosomal recessive (OMIM:274300); Thyroid hormone resistance, generalized, autosomal dominant (OMIM:188570); Thyroid hormone resistance, selective pituitary (OMIM:145650); Resistance to thyroid hormone due to a mutation in thyroid hormone receptor beta (ORPHA:566243)
THSD7A	—
THSD8	—
TIALD	—
TICAM1	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6 (OMIM:614850); Herpes simplex virus encephalitis (ORPHA:1930)
TICRR	—
TIGD1	—
TILAM	—
TIMM44	—
TIMM8A	Mohr-Tranebjaerg syndrome (OMIM:304700); Mohr-Tranebjaerg syndrome (ORPHA:52368)
TIMP3	Sorsby fundus dystrophy (OMIM:136900); Sorsby fundus dystrophy (ORPHA:59181)
TINAG	—
TINF2	Dyskeratosis congenita, autosomal dominant 3 (OMIM:613990); Dyskeratosis congenita, autosomal dominant, 1 (OMIM:127550); Revesz syndrome (OMIM:268130); Hoyeraal-Hreidarsson syndrome (ORPHA:3322); Revesz syndrome (ORPHA:3088); Dyskeratosis congenita (ORPHA:1775)
TIRAP	OMIM:614382; OMIM:607948; OMIM:611162
TJP2	Cholestasis, progressive familial intrahepatic 4 (OMIM:615878); Hypercholanemia, familial (OMIM:607748); ORPHA:90635; ORPHA:238475; ORPHA:480483
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type) (OMIM:609560); Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3 (OMIM:617069); Autosomal recessive progressive external ophthalmoplegia (ORPHA:254886); Mitochondrial DNA depletion syndrome, myopathic form (ORPHA:254875)
TLDC2	—
TLL1	Atrial septal defect 6 (OMIM:613087); Atrial septal defect, ostium primum type (ORPHA:99106); Atrial septal defect, ostium secundum type (ORPHA:99103)
TLR1	OMIM:613223; OMIM:621601
TLR2	Colorectal cancer (OMIM:114500); Leprosy, susceptibility to (OMIM:246300); OMIM:607948
TLR3	OMIM:609423; Immunodeficiency 83, susceptibility to viral infections (OMIM:613002); Herpes simplex virus encephalitis (ORPHA:1930)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
TLR4	Behçet disease (ORPHA:117)
TLR5	OMIM:608556; OMIM:615557; OMIM:601744
TLR6	—
TLR9	—
TLX1	ORPHA:99861
TLX1NB	—
TLX2	—
TLX3	ORPHA:99861
TM4SF19	—
TMBIM7P	—
TMC1	Deafness, neurosensory, autosomal recessive 7 (OMIM:600974); Deafness, autosomal dominant 36 (OMIM:606705); ORPHA:90635; ORPHA:90636
TMC6	Epidermodysplasia verruciformis, susceptibility to, 1 (OMIM:226400); Inherited epidermodysplasia verruciformis (ORPHA:302)
TMC8	Epidermodysplasia verruciformis 2 (OMIM:618231); Inherited epidermodysplasia verruciformis (ORPHA:302)
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1 (OMIM:213980); Cerebrofaciothoracic dysplasia (ORPHA:1394)
TMEM114	—
TMEM126A	Optic atrophy 7 with or without auditory neuropathy (OMIM:612989); ORPHA:227976
TMEM127	PHEOCHROMOCYTOMA (OMIM:171300); ORPHA:404511; Hereditary pheochromocytoma-paraganglioma (ORPHA:29072)
TMEM135	—
TMEM138	Joubert syndrome 16 (OMIM:614465); Joubert syndrome with oculorenal defect (ORPHA:2318)
TMEM14B	—
TMEM151B	—
TMEM165	Congenital disorder of glycosylation, type IIK (OMIM:614727); ORPHA:314667
TMEM184C	—
TMEM186	—
TMEM187	—
TMEM216	Joubert syndrome 2 (OMIM:608091); Meckel syndrome, type 2 (OMIM:603194); Retinitis pigmentosa 98 (OMIM:620996); Orofaciodigital syndrome type 6 (ORPHA:2754); Retinitis pigmentosa (ORPHA:791); Joubert syndrome with oculorenal defect (ORPHA:2318); Meckel syndrome (ORPHA:564)
TMEM231	Meckel syndrome 11 (OMIM:615397); Joubert syndrome 20 (OMIM:614970); Orofaciodigital syndrome type 6 (ORPHA:2754); Joubert syndrome with oculorenal defect (ORPHA:2318); Meckel syndrome (ORPHA:564)
TMEM237	Joubert syndrome 14 (OMIM:614424); Joubert syndrome with renal defect (ORPHA:220497); Joubert syndrome with oculorenal defect (ORPHA:2318); Meckel syndrome (ORPHA:564); Isolated Joubert syndrome (ORPHA:475)
TMEM266	—
TMEM43	Arrhythmogenic right ventricular dysplasia, familial, 5 (OMIM:604400); Emery-Dreifuss muscular dystrophy 7, autosomal dominant (OMIM:614302); Auditory neuropathy, autosomal dominant 3 (OMIM:619832); Autosomal dominant Emery-Dreifuss muscular dystrophy (ORPHA:98853); ORPHA:293888; ORPHA:293899; ORPHA:293910
TMEM50B	—
TMEM52B	—
TMEM61	—
TMEM67	Rhyns syndrome (OMIM:602152); Joubert syndrome 6 (OMIM:610688); Coach syndrome 1 (OMIM:216360); Bardet-Biedl syndrome 14 (OMIM:615991); Meckel syndrome 3 (OMIM:607361); Nephronophthisis 11 (OMIM:613550); Joubert syndrome with hepatic defect (ORPHA:1454); Senior-Boichis syndrome (ORPHA:84081); RHYNS syndrome (ORPHA:140976); Meckel syndrome (ORPHA:564); Isolated Joubert syndrome (ORPHA:475)
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2 (OMIM:614052); TMEM70-related mitochondrial encephalo-cardio-myopathy (ORPHA:1194)
TMEM91	—
TMIE	Deafness, autosomal recessive 6 (OMIM:600971); ORPHA:90636
TMPO	Familial isolated dilated cardiomyopathy (ORPHA:154)
TMPPE	—
TMPRSS15	Enterokinase deficiency (OMIM:226200); ORPHA:168601
TMPRSS3	Deafness, autosomal recessive 8/10 (OMIM:601072); ORPHA:90636
TMPRSS5	—
TMPRSS6	Iron-Refractory iron deficiency anemia (OMIM:206200); IRIDA syndrome (ORPHA:209981)
TMTC3	Lissencephaly 8 (OMIM:617255); Periventricular nodular heterotopia (ORPHA:98892); Cobblestone lissencephaly without muscular or ocular involvement (ORPHA:352682)
TNF	Asthma, susceptibility to (OMIM:600807); Migraine with or without aura, susceptibility to, 1 (OMIM:157300); Immunodeficiency 127 (OMIM:620977); OMIM:611162
TNFRSF10B	Squamous cell carcinoma, head and neck (OMIM:275355); ORPHA:494547; ORPHA:494550; ORPHA:502363; ORPHA:502366; ORPHA:500478; ORPHA:500464
TNFRSF11A	Familial expansile osteolysis (OMIM:174810); Paget disease of bone 2, early-onset (OMIM:602080); Osteopetrosis, autosomal recessive 7 (OMIM:612301); Juvenile Paget disease (ORPHA:2801); ORPHA:178389; ORPHA:391490; ORPHA:85195; Dysosteosclerosis (ORPHA:1782)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
TNFRSF11B	Paget disease of bone 5, juvenile-onset (OMIM:239000); Familial calcium pyrophosphate deposition (ORPHA:1416); Juvenile Paget disease (ORPHA:2801)
TNFRSF13B	Immunoglobulin A deficiency 2 (OMIM:609529); Immunodeficiency, common variable, 2 (OMIM:240500); ORPHA:696907
TNFRSF13C	Immunodeficiency, common variable, 4 (OMIM:613494); Immunodeficiency, common variable, 2 (OMIM:240500); ORPHA:696925
TNFRSF1A	Periodic fever, familial, autosomal dominant (OMIM:142680); OMIM:614810; Intermittent hydrarthrosis (ORPHA:329967); Tumor necrosis factor receptor 1 associated periodic syndrome (ORPHA:32960)
TNFRSF1B	Sézary syndrome (ORPHA:3162); Classic mycosis fungoides (ORPHA:2584)
TNFRSF9	Immunodeficiency 109 with lymphoproliferation (OMIM:620282); ORPHA:664726
TNFSF11	Osteopetrosis, autosomal recessive 2 (OMIM:259710); Autosomal recessive malignant osteopetrosis (ORPHA:667)
TNFSF14	—
TNFSF4	OMIM:608446; Systemic lupus erythematosus (ORPHA:536); Narcolepsy type 1 (ORPHA:2073)
TNKS	—
TNNC1	Cardiomyopathy, dilated, 1Z (OMIM:611879); Cardiomyopathy, familial hypertrophic, 13 (OMIM:613243); Familial isolated dilated cardiomyopathy (ORPHA:154)
TNNI2	Arthrogryposis, distal, type 2B (OMIM:601680); Sheldon-Hall syndrome (ORPHA:1147); Distal arthrogryposis type 1 (ORPHA:1146)
TNNI3	Cardiomyopathy, familial restrictive, 1 (OMIM:115210); Cardiomyopathy, dilated, 2A (OMIM:611880); Cardiomyopathy, dilated, 1FF (OMIM:613286); Cardiomyopathy, familial hypertrophic, 7 (OMIM:613690); Familial isolated dilated cardiomyopathy (ORPHA:154); Familial isolated restrictive cardiomyopathy (ORPHA:75249)
TNNI3K	Cardiac conduction disease with or without dilated cardiomyopathy (OMIM:616117); ORPHA:436242
TNNT1	Nemaline myopathy 5C, autosomal dominant (OMIM:620389); Nemaline myopathy 5B, autosomal recessive, childhood-onset (OMIM:620386); Nemaline myopathy 5, Amish type (OMIM:605355); Amish nemaline myopathy (ORPHA:98902)
TNNT2	Cardiomyopathy, familial hypertrophic, 2 (OMIM:115195); Cardiomyopathy, dilated, 1D (OMIM:601494); Cardiomyopathy, familial restrictive, 3 (OMIM:612422); ORPHA:54260; Familial isolated dilated cardiomyopathy (ORPHA:154); Familial isolated restrictive cardiomyopathy (ORPHA:75249)
TNNT3	Arthrogryposis, distal, type 2B2 (OMIM:618435); Sheldon-Hall syndrome (ORPHA:1147); Distal arthrogryposis type 1 (ORPHA:1146)
TNP1	—
TNR	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus (OMIM:619653)
TNRC6B	Global developmental delay with speech and behavioral abnormalities (OMIM:619243); ORPHA:528084
TNS2	—
TNS3	—
TNXA	—
TNXB	Vesicoureteral reflux 8 (OMIM:615963); Ehlers-Danlos syndrome, classic-like (OMIM:606408); ORPHA:289365; Classical-like Ehlers-Danlos syndrome type 1 (ORPHA:230839)
TOMM20L	—
TOMM22	—
TOMM40L	—
TOMT	—
TOP1	—
TOP2A	—
TOPORS	Retinitis pigmentosa 31 (OMIM:609923); Orofaciodigital syndrome type 6 (ORPHA:2754); Retinitis pigmentosa (ORPHA:791)
TOR1A	Dystonia 1, torsion, autosomal dominant (OMIM:128100); Arthrogryposis multiplex congenita 5 (OMIM:618947); Early-onset generalized limb-onset dystonia (ORPHA:256); Myoclonus-dystonia syndrome (ORPHA:36899)
TOX	—
TP53	Glioma susceptibility 1 (OMIM:137800); Nasopharyngeal carcinoma (OMIM:607107); Pancreatic cancer (OMIM:260350); Papilloma of choroid plexus (OMIM:260500); Basal cell carcinoma 7 (OMIM:614740); Osteosarcoma (OMIM:259500); Breast cancer (OMIM:114480); Bone marrow failure syndrome 5 (OMIM:618165); Li-Fraumeni syndrome (OMIM:151623); Colorectal cancer (OMIM:114500); Hepatocellular carcinoma (OMIM:114550); Adrenocortical carcinoma, pediatric (OMIM:202300); ORPHA:251579; ORPHA:251576; ORPHA:210159; Adrenocortical carcinoma (ORPHA:1501); Papilloma of choroid plexus (ORPHA:2807); Essential thrombocythemia (ORPHA:3318); Hereditary breast and/or ovarian cancer syndrome (ORPHA:145); ORPHA:585909; Cushing disease (ORPHA:96253); ORPHA:52688; Familial pancreatic carcinoma (ORPHA:1333); ORPHA:667662; ORPHA:70573; Osteosarcoma (ORPHA:668); ORPHA:251899; ORPHA:293199; ORPHA:67038; Li-Fraumeni syndrome (ORPHA:524); ORPHA:99757; ORPHA:99756
TP53BP1	—
TP53RK	Galloway-Mowat syndrome 4 (OMIM:617730); Galloway-Mowat syndrome (ORPHA:2065)
TP63	Rapp-Hodgkin syndrome (OMIM:129400); Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3 (OMIM:604292); Limb-Mammary syndrome (OMIM:603543); Adult syndrome (OMIM:103285); Orofacial cleft 8 (OMIM:618149); Premature ovarian failure 21 (OMIM:620311); Hay-Wells syndrome (OMIM:106260); Split-Hand/foot malformation 4 (OMIM:605289); Isolated cleft lip (ORPHA:199302); Cleft lip/palate (ORPHA:199306); Classic bladder exstrophy (ORPHA:93930); ADULT syndrome (ORPHA:978);

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	Ankyloblepharon filiforme adnatum-cleft palate syndrome (ORPHA:1072); EEC syndrome (ORPHA:1896); Isolated split hand-split foot malformation (ORPHA:2440); Cleft lip and alveolus (ORPHA:141291); Limb-mammary syndrome (ORPHA:69085)
TP73	Ciliary dyskinesia, primary, 47, and lissencephaly (OMIM:619466); ORPHA:70573
TPCN2	OMIM:612267
TPH1	—
TPH2	Major depressive disorder (OMIM:608516); Attention deficit-hyperactivity disorder, susceptibility to, 7 (OMIM:613003)
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type) (OMIM:614458); ORPHA:293955
TPM1	Cardiomyopathy, familial hypertrophic, 3 (OMIM:115196); Cardiomyopathy, dilated, 1Y (OMIM:611878); ORPHA:54260; Familial isolated dilated cardiomyopathy (ORPHA:154)
TPM2	Congenital myopathy 23 (OMIM:609285); Arthrogryposis, distal, type 1A (OMIM:108120); Congenital fiber-type disproportion myopathy (ORPHA:2020); Typical nemaline myopathy (ORPHA:171436); Childhood-onset nemaline myopathy (ORPHA:171439); Sheldon-Hall syndrome (ORPHA:1147); Distal arthrogryposis type 1 (ORPHA:1146); Cap myopathy (ORPHA:171881)
TPM3	Nemaline myopathy 1 (OMIM:609284); Myopathy, congenital, with fiber-type disproportion (OMIM:255310); ORPHA:178342; Congenital fiber-type disproportion myopathy (ORPHA:2020); Intermediate nemaline myopathy (ORPHA:171433); Childhood-onset nemaline myopathy (ORPHA:171439); ORPHA:476406; Cap myopathy (ORPHA:171881)
TPMT	THIOPURINE S-METHYLTRANSFERASE DEFICIENCY (OMIM:610460)
TPO	Thyroid hormonogenesis, genetic defect in, 2A (OMIM:274500); Familial thyroid dyshormonogenesis (ORPHA:95716)
TPP1	Spinocerebellar ataxia, autosomal recessive 7 (OMIM:609270); Ceroid lipofuscinosis, neuronal, 2 (OMIM:204500); ORPHA:699769; ORPHA:699761; ORPHA:699751; Childhood-onset autosomal recessive slowly progressive spinocerebellar ataxia (ORPHA:284324)
TPR	Intellectual developmental disorder, autosomal recessive 79 (OMIM:620393); ORPHA:146; ORPHA:88616
TPRN	Deafness, autosomal recessive 79 (OMIM:613307); ORPHA:90636
TPTE	—
TRADD	—
TRAF3	Immunodeficiency 132B (OMIM:621096); Immunodeficiency 132A (OMIM:614849); Herpes simplex virus encephalitis (ORPHA:1930)
TRAF3IP2	Candidiasis, familial, 8 (OMIM:615527); OMIM:614070; Chronic mucocutaneous candidiasis (ORPHA:1334)
TRAF6	Autosomal dominant hypohidrotic ectodermal dysplasia (ORPHA:1810)
TRAK1	Epileptic encephalopathy, early infantile, 68 (OMIM:618201); Non-specific early-onset epileptic encephalopathy (ORPHA:442835)
TRAP1	—
TRAPPC2L	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis (OMIM:618331)
TRAPPC4	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy (OMIM:618741); ORPHA:528084
TRAPPC9	Intellectual developmental disorder, autosomal recessive 13 (OMIM:613192); Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome (ORPHA:352530); ORPHA:88616
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 1, with or without atrial dysfunction and/or dilated cardiomyopathy (OMIM:604772); Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness (OMIM:615441); Romano-Ward syndrome (ORPHA:101016); Catecholaminergic polymorphic ventricular tachycardia (ORPHA:3286)
TREH	Trehalase deficiency (OMIM:612119); Trehalase deficiency (ORPHA:103909)
TREM2	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (OMIM:221770); Alzheimer disease 17, susceptibility to (OMIM:615080); Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2 (OMIM:618193); Nasu-Hakola disease (ORPHA:2770); Behavioral variant of frontotemporal dementia (ORPHA:275864); Early-onset autosomal dominant Alzheimer disease (ORPHA:1020); Amyotrophic lateral sclerosis (ORPHA:803); Progressive non-fluent aphasia (ORPHA:100070); Semantic dementia (ORPHA:100069)
TRERF1	—
TREX1	Chilblain lupus 1 (OMIM:610448); Aicardi-Goutieres syndrome 1 (OMIM:225750); Systemic lupus erythematosus (OMIM:152700); Vasculopathy, retinal, with cerebral leukodystrophy (OMIM:192315); Aicardi-Goutières syndrome (ORPHA:51); Systemic lupus erythematosus (ORPHA:536); ORPHA:481662; Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations (ORPHA:247691)
TRHR	Hypothyroidism, congenital, nongoitrous, 7 (OMIM:618573); Resistance to thyrotropin-releasing hormone syndrome (ORPHA:99832)
TRIB1AL	—
TRIB2	—
TRIB3	—
TRIM13	—
TRIM21	—
TRIM24	ORPHA:146
TRIM25	—
TRIM32	Muscular dystrophy, limb-girdle, type 2H (OMIM:254110); Bardet-Biedl syndrome 11 (OMIM:615988); Bardet-Biedl syndrome (ORPHA:110); TRIM32-related limb-girdle muscular dystrophy R8 (ORPHA:1878)
TRIM33	Developmental dysplasia of the hip 4 (OMIM:621311); ORPHA:146; Dislocation of the hip-dysmorphism

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	syndrome (ORPHA:2412)
TRIM37	Mulibrey nanism (OMIM:253250); Mulibrey nanism (ORPHA:2576)
TRIM5	—
TRIM59	—
TRIO	Intellectual developmental disorder, autosomal dominant 63, with macrocephaly (OMIM:618825); Intellectual developmental disorder, autosomal dominant 44, with microcephaly (OMIM:617061); Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome (ORPHA:476126)
TRIOBP	Deafness, autosomal recessive 28 (OMIM:609823); ORPHA:90636
TRIP11	Osteochondrodysplasia (OMIM:184260); Achondrogenesis, type IA (OMIM:200600); Odontochondrodysplasia (ORPHA:166272); Achondrogenesis type 1A (ORPHA:93299)
TRIP12	Clark-Baraitser syndrome (OMIM:617752); ORPHA:528084
TRIP13	Mosaic variegated aneuploidy syndrome 3 (OMIM:617598); Oocyte maturation defect 9 (OMIM:619011); Mosaic variegated aneuploidy syndrome (ORPHA:1052); Nephroblastoma (ORPHA:654)
TRMU	Deafness, aminoglycoside-induced (OMIM:580000); Liver failure, infantile, transient (OMIM:613070); ORPHA:90641; ORPHA:217371; Mitochondrial myopathy with reversible cytochrome C oxidase deficiency (ORPHA:254864)
TRNT1	Retinitis pigmentosa and erythrocytic microcytosis (OMIM:616959); Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay (OMIM:616084); ORPHA:369861
TROAP	—
TRPA1	Episodic pain syndrome, familial, 1 (OMIM:615040); ORPHA:581271; ORPHA:391389
TRPC3	Spinocerebellar ataxia 41 (OMIM:616410); Spinocerebellar ataxia type 41 (ORPHA:458798)
TRPC4	—
TRPC6	Focal segmental glomerulosclerosis 2 (OMIM:603965); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
TRPM1	Night blindness, congenital stationary (complete), 1C, autosomal recessive (OMIM:613216); ORPHA:714079
TRPM2	—
TRPM3	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures (OMIM:620224); Cataract 50 with or without glaucoma (OMIM:620253); ORPHA:178469
TRPM4	Progressive familial heart block, type IB (OMIM:604559); Erythrokeratodermia veriabilis et progressiva 6 (OMIM:618531); Progressive symmetric erythrokeratodermia (ORPHA:316); Brugada syndrome (ORPHA:130); Hereditary progressive cardiac conduction defect (ORPHA:871)
TRPM6	Hypomagnesemia 1, intestinal (OMIM:602014); ORPHA:30924
TRPM7	Amyotrophic lateral sclerosis-parkinsonism/dementia complex 1 (OMIM:105500); ORPHA:90020; Autosomal dominant macrothrombocytopenia (ORPHA:140957)
TRPS1	Trichorhinophalangeal syndrome, type I (OMIM:190350); Trichorhinophalangeal syndrome, type III (OMIM:190351); Trichorhinophalangeal syndrome type 1 (ORPHA:77258); Trichorhinophalangeal syndrome type 2 (ORPHA:502)
TRPV1	—
TRPV3	Palmoplantar keratoderma, nonepidermolytic, focal 2 (OMIM:616400); Olmsted syndrome 1 (OMIM:614594); Mutilating palmoplantar keratoderma with periorificial keratotic plaques (ORPHA:659); ORPHA:448264
TRPV4	Spondylometaphyseal dysplasia, Kozlowski type (OMIM:184252); Digital arthropathy-brachydactyly, familial (OMIM:606835); Neuronopathy, distal hereditary motor, type VIII (OMIM:600175); Scapuloperoneal spinal muscular atrophy (OMIM:181405); Parastremmatic dwarfism (OMIM:168400); Hereditary motor and sensory neuropathy, type IIC (OMIM:606071); Brachyolmia type 3 (OMIM:113500); Metatropic dysplasia (OMIM:156530); Avascular necrosis of femoral head, primary, 2 (OMIM:617383); Spondyloepiphyseal dysplasia, Maroteaux type (OMIM:184095); OMIM:613508; ORPHA:263482; ORPHA:99937; Autosomal dominant congenital benign spinal muscular atrophy (ORPHA:1216); Familial avascular necrosis of femoral head (ORPHA:86820); Metatropic dysplasia (ORPHA:2635); ORPHA:431255; Spondylometaphyseal dysplasia, Kozlowski type (ORPHA:93314); Autosomal dominant brachyolmia (ORPHA:93304); Familial digital arthropathy-brachydactyly (ORPHA:85169)
TRPV5	—
TRRAP	Deafness, autosomal dominant 75 (OMIM:618778); Developmental delay with or without dysmorphic facies and autism (OMIM:618454); ORPHA:90635
TSC1	Focal cortical dysplasia of taylor (OMIM:607341); LYMPHANGIOLEIOMYOMATOSIS (OMIM:606690); Tuberous sclerosis-1 (OMIM:191100); ORPHA:210159; ORPHA:269008; Tuberous sclerosis complex (ORPHA:805); Lymphangioleiomyomatosis (ORPHA:538)
TSC2	Tuberous sclerosis-2 (OMIM:613254); Focal cortical dysplasia of taylor (OMIM:607341); LYMPHANGIOLEIOMYOMATOSIS (OMIM:606690); ORPHA:210159; ORPHA:269001; ORPHA:269008; Tuberous sclerosis complex (ORPHA:805); Lymphangioleiomyomatosis (ORPHA:538); ORPHA:88924
TSEN2	Pontocerebellar hypoplasia, type 2B (OMIM:612389); Pontocerebellar hypoplasia type 2 (ORPHA:2524)
TSEN34	Pontocerebellar hypoplasia, type 2C (OMIM:612390); Pontocerebellar hypoplasia type 2 (ORPHA:2524)
TSEN54	Pontocerebellar hypoplasia, type 5 (OMIM:610204); Pontocerebellar hypoplasia, type 4 (OMIM:225753); Pontocerebellar hypoplasia, type 2A (OMIM:277470); Pontocerebellar hypoplasia type 2 (ORPHA:2524); Pontocerebellar hypoplasia type 4 (ORPHA:166063)
TSFM	Combined oxidative phosphorylation deficiency 3 (OMIM:610505); ORPHA:168566
TSG101	—
TSHB	Hypothyroidism, congenital, nongoitrous, 4 (OMIM:275100); Isolated thyroid-stimulating hormone deficiency (ORPHA:90674)
TSHR	Hyperthyroidism, nonautoimmune (OMIM:609152); Hyperthyroidism, familial gestational (OMIM:603373);

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	Hypothyroidism, congenital, nongoitrous, 1 (OMIM:275200); Hypothyroidism due to TSH receptor mutations (ORPHA:90673); Familial gestational hyperthyroidism (ORPHA:99819); Thyroid hypoplasia (ORPHA:95720); Athyreosis (ORPHA:95713); Familial hyperthyroidism due to mutations in TSH receptor (ORPHA:424)
TSHZ1	Aural atresia, congenital (OMIM:607842); ORPHA:141074
TSLP	—
TSNAX	—
TSPAN12	Exudative vitreoretinopathy 5 (OMIM:613310); Familial exudative vitreoretinopathy (ORPHA:891)
TSPAN17	—
TSPAN31	—
TSPAN7	Intellectual developmental disorder, X-linked 58 (OMIM:300210); ORPHA:777
TSPAN8	—
TSPEAR	Tooth agenesis, selective, 10 (OMIM:620173); Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis (OMIM:618180); Deafness, autosomal recessive 98 (OMIM:614861); ORPHA:685067
TSPO	—
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome (OMIM:608800); Sudden infant death-dysgenesis of the testes syndrome (ORPHA:168593)
TSR1	—
TSR2	Diamond-Blackfan anemia 14 with mandibulofacial dysostosis (OMIM:300946); Diamond-Blackfan anemia (ORPHA:124)
TSSC4	—
TSSK4	—
TST	—
TTBK2	Spinocerebellar ataxia 11 (OMIM:604432); Spinocerebellar ataxia type 11 (ORPHA:98767)
TTC19	Mitochondrial complex III deficiency, nuclear type 2 (OMIM:615157); ORPHA:1460
TTC21B	Short-Rib thoracic dysplasia 4 with or without polydactyly (OMIM:613819); Nephronophthisis 12 (OMIM:613820); Jeune syndrome (ORPHA:474); ORPHA:93591
TTC33	—
TTC7A	Gastrointestinal defects and immunodeficiency syndrome (OMIM:243150); Combined immunodeficiency-multiple intestinal atresia (ORPHA:436252)
TTC8	Retinitis pigmentosa 51 (OMIM:613464); Bardet-Biedl syndrome 8 (OMIM:615985); Bardet-Biedl syndrome (ORPHA:110); Retinitis pigmentosa (ORPHA:791)
TTI2	Intellectual developmental disorder, autosomal recessive 39 (OMIM:615541); Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome (ORPHA:391307)
TTLL1	—
TTLL5	Cone-Rod dystrophy 19 (OMIM:615860); Cone rod dystrophy (ORPHA:1872)
TTN	Congenital myopathy 5 with cardiomyopathy (OMIM:611705); Myopathy, myofibrillar, 9, with early respiratory failure (OMIM:603689); Muscular dystrophy, limb-girdle, autosomal recessive 10 (OMIM:608807); Cardiomyopathy, dilated, 1G (OMIM:604145); Cardiomyopathy, familial hypertrophic, 9 (OMIM:613765); Tibial muscular dystrophy, tardive (OMIM:600334); ORPHA:289377; Hereditary myopathy with early respiratory failure (ORPHA:178464); Hereditary atrial fibrillation (ORPHA:334); ORPHA:707983; Familial isolated dilated cardiomyopathy (ORPHA:154); Autosomal recessive centronuclear myopathy (ORPHA:169186); ORPHA:293888; ORPHA:293899; Classic multiminicore myopathy (ORPHA:324604); ORPHA:293910; ORPHA:140922; ORPHA:466921; Tibial muscular dystrophy (ORPHA:609)
TTPA	Ataxia with isolated vitamin E deficiency (OMIM:277460); Ataxia with vitamin E deficiency (ORPHA:96)
TTR	Carpal tunnel syndrome (OMIM:115430); Dystransthyretinemic euthyroidal hyperthyroxinemia (OMIM:145680); Amyloidosis, hereditary, transthyretin-related (OMIM:105210); ATTRV30M amyloidosis (ORPHA:85447); ATTRV122I amyloidosis (ORPHA:85451); ORPHA:597939
TUB	Retinal dystrophy and obesity (OMIM:616188); Retinitis pigmentosa (ORPHA:791)
TUBA1A	Lissencephaly 3 (OMIM:611603); Lissencephaly due to TUBA1A mutation (ORPHA:171680); Fetal akinesia deformation sequence (ORPHA:994); Congenital fibrosis of extraocular muscles (ORPHA:45358); Tubulinopathy-associated dysgyria (ORPHA:467166)
TUBA1B	—
TUBA1C	Oocyte/zygote/embryo maturation arrest 24 (OMIM:621232)
TUBA8	Macrothrombocytopenia, isolated, 2, autosomal dominant (OMIM:619840); Polymicrogyria with optic nerve hypoplasia (ORPHA:250972); Autosomal dominant macrothrombocytopenia (ORPHA:140957)
TUBB1	Macrothrombocytopenia, autosomal dominant, tubb1-related (OMIM:613112); Autosomal dominant macrothrombocytopenia (ORPHA:140957)
TUBB2B	Cortical dysplasia, complex, with other brain malformations 7 (OMIM:610031); Congenital fibrosis of extraocular muscles (ORPHA:45358); Polymicrogyria due to TUBB2B mutation (ORPHA:300573); Tubulinopathy-associated dysgyria (ORPHA:467166); Dysequilibrium syndrome (ORPHA:1766)
TUBB3	Cortical dysplasia, complex, with other brain malformations 1 (OMIM:614039); Fibrosis of extraocular muscles, congenital, 3A, with or without extraocularinvolvement (OMIM:600638); Congenital fibrosis of extraocular muscles (ORPHA:45358); Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation (ORPHA:300570); Tubulinopathy-associated dysgyria (ORPHA:467166)
TUBB6	Facial palsy, congenital, with ptosis and velopharyngeal dysfunction (OMIM:617732)
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, 3 (OMIM:616335); Autosomal recessive chorioretinopathy-microcephaly syndrome (ORPHA:2518)
TUBGCP5	—
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1 (OMIM:251270); Autosomal recessive

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	chorioretinopathy-microcephaly syndrome (ORPHA:2518)
TUFM	Combined oxidative phosphorylation deficiency 4 (OMIM:610678); ORPHA:254925
TULP1	Retinitis pigmentosa 14 (OMIM:600132); Leber congenital amaurosis 15 (OMIM:613843); Leber congenital amaurosis (ORPHA:65); Retinitis pigmentosa (ORPHA:791)
TUSC3	Intellectual developmental disorder, autosomal recessive 7 (OMIM:611093); ORPHA:88616
TWIST1	Robinow-Sorauf syndrome (OMIM:180750); Sweeney-Cox syndrome (OMIM:617746); Saethre-Chotzen syndrome (OMIM:101400); Craniosynostosis 1 (OMIM:123100); Non-syndromic sagittal craniosynostosis (ORPHA:35093); Non-syndromic bicoronal craniosynostosis (ORPHA:35099); Saethre-Chotzen syndrome (ORPHA:794)
TWIST2	Ablepharon-Macrostomia syndrome (OMIM:200110); Focal facial dermal dysplasia 3, Setleis type (OMIM:227260); Barber-Say syndrome (OMIM:209885); Barber-Say syndrome (ORPHA:1231); Ablepharon macrostomia syndrome (ORPHA:920); Focal facial dermal dysplasia type III (ORPHA:1807)
TXNIP	—
TXNRD2	Glucocorticoid deficiency 5 (OMIM:617825); Familial glucocorticoid deficiency (ORPHA:361); Familial isolated dilated cardiomyopathy (ORPHA:154)
TYK2	Immunodeficiency 35 (OMIM:611521); ORPHA:98842; ORPHA:300865; ORPHA:331226
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type) (OMIM:603041); Mitochondrial neurogastrointestinal encephalomyopathy (ORPHA:298)
TYMS	Dyskeratosis congenita, digenic (OMIM:620040); Dyskeratosis congenita (ORPHA:1775)
TYMSOS	—
TYR	Albinism, oculocutaneous, type IB (OMIM:606952); Albinism, oculocutaneous, type IA (OMIM:203100); SKIN/HAIR/EYE PIGMENTATION, VARIATION IN, 3; SHEP3 (OMIM:601800); Oculocutaneous albinism type 1A (ORPHA:79431); Oculocutaneous albinism type 1B (ORPHA:79434); Waardenburg syndrome type 2 (ORPHA:895); ORPHA:352737; ORPHA:352734
TYRO3	—
TYROBP	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (OMIM:221770); Nasu-Hakola disease (ORPHA:2770)
TYRP1	Albinism, oculocutaneous, type III (OMIM:203290); OMIM:612271; Oculocutaneous albinism type 3 (ORPHA:79433)
TYW1	—
UACA	—
UBA1	VEXAS syndrome, somatic (OMIM:301054); Spinal muscular atrophy, X-linked 2 (OMIM:301830); ORPHA:596753; Infantile-onset X-linked spinal muscular atrophy (ORPHA:1145)
UBA2	ACCES syndrome (OMIM:619959); Aplasia cutis congenita (ORPHA:1114)
UBA6	—
UBAC2	Behçet disease (ORPHA:117)
UBE2A	Intellectual developmental disorder, X-linked syndromic, Nascimento type (OMIM:300860); X-linked intellectual disability, Nascimento type (ORPHA:163956)
UBE2B	—
UBE2NL	—
UBE3A	Angelman syndrome (OMIM:105830); 15q11q13 microduplication syndrome (ORPHA:238446); Angelman syndrome due to maternal 15q11q13 deletion (ORPHA:98794); Angelman syndrome due to paternal uniparental disomy of chromosome 15 (ORPHA:98795); Angelman syndrome due to a point mutation (ORPHA:411511); Angelman syndrome due to imprinting defect in 15q11-q13 (ORPHA:411515)
UBE3C	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities (OMIM:620270)
UBE4A	Neurodevelopmental disorder with hypotonia and gross motor and seech delay (OMIM:619639); ORPHA:88616
UBIAD1	Corneal dystrophy, crystalline, of schnyder (OMIM:121800); ORPHA:98967
UBQLN2	Amyotrophic lateral sclerosis 15 with or without frontotemporal dementia (OMIM:300857); Amyotrophic lateral sclerosis (ORPHA:803)
UBR1	Johanson-Blizzard syndrome (OMIM:243800); Johanson-Blizzard syndrome (ORPHA:2315)
UBR3	—
UBR7	Li-Campeau syndrome (OMIM:619189); ORPHA:528084
UBTF	Neurodegeneration, childhood-onset, with brain atrophy (OMIM:617672); Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder (ORPHA:500180)
UCHL1	Spastic paraplegia 79A, autosomal dominant (OMIM:620221); Spastic paraplegia 79, autosomal recessive (OMIM:615491); Parkinson disease 5, susceptibility to (OMIM:613643); Young-onset Parkinson disease (ORPHA:2828); ORPHA:352654
UCN	—
UCP1	—
UCP2	OMIM:607447; Hyperinsulinism due to UCP2 deficiency (ORPHA:276556)
UCP3	OBESITY (OMIM:601665)
UFD1	22q11.2 deletion syndrome (ORPHA:567)
UGCG	—
UGGT1	Congenital disorder of glycosylation, type IICC (OMIM:621381)
UGGT2	—
UGT1A8	—
UGT2A1	—

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
UGT2A2	—
UGT2B10	—
UGT2B17	OMIM:612560
UGT2B4	—
UGT2B7	—
UGT8	—
UIMC1	—
ULK4	—
UMOD	Tubulointerstitial kidney disease, autosomal dominant, 1 (OMIM:162000); ORPHA:88950
UMPS	Orotic aciduria (OMIM:258900); Hereditary orotic aciduria (ORPHA:30)
UNC119	Immunodeficiency 13 (OMIM:615518); Cone-rod dystrophy 24 (OMIM:620342); ORPHA:228000; Cone rod dystrophy (ORPHA:1872)
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3 (OMIM:608898); Familial hemophagocytic lymphohistiocytosis (ORPHA:540)
UNC5C	—
UNC5CL	—
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2 (OMIM:616801); ORPHA:700333
UNC93A	—
UNC93B1	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1 (OMIM:610551); Herpes simplex virus encephalitis (ORPHA:1930)
UNG	Immunodeficiency with hyper IgM, type 5 (OMIM:608106); ORPHA:101092
UNKL	—
UPB1	Beta-ureidopropionase deficiency (OMIM:613161); Beta-ureidopropionase deficiency (ORPHA:65287)
UPF3B	Intellectual developmental disorder, X-linked syndromic 14 (OMIM:300676); Lujan-Fryns syndrome (ORPHA:776); ORPHA:777
UPK3A	ORPHA:93100
UQCRB	Mitochondrial complex III deficiency, nuclear type 3 (OMIM:615158); ORPHA:1460
UQCRFS1	Mitochondrial complex III deficiency, nuclear type 10 (OMIM:618775); ORPHA:1460
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4 (OMIM:615159); ORPHA:1460
URB1	—
UROC1	Urocanase deficiency (OMIM:276880); Urocanic aciduria (ORPHA:210128)
UROD	Porphyria cutanea tarda (OMIM:176100); ORPHA:443062; Hepatoerythropoietic porphyria (ORPHA:95159)
UROS	Porphyria, congenital erythropoietic (OMIM:263700); Congenital erythropoietic porphyria (ORPHA:79277)
USB1	Poikiloderma with neutropenia (OMIM:604173); ORPHA:221046; Dyskeratosis congenita (ORPHA:1775)
USF1	OMIM:602491
USH1C	Usher syndrome, type I (OMIM:276900); Usher syndrome, type IC (OMIM:276904); Deafness, neurosensory, autosomal recessive 18 (OMIM:602092); ORPHA:90636; Usher syndrome type 1 (ORPHA:231169)
USH1G	Usher syndrome, type IG (OMIM:606943); Usher syndrome type 1 (ORPHA:231169)
USH2A	Usher syndrome, type IIA (OMIM:276901); Retinitis pigmentosa 39 (OMIM:613809); Retinitis pigmentosa (ORPHA:791); Usher syndrome type 2 (ORPHA:231178)
USP1	—
USP15	—
USP2	—
USP26	Spermatogenic failure, X-linked, 6 (OMIM:301101); ORPHA:276234
USP3	—
USP46	—
USP7	Chromosome 16p13.2 deletion syndrome (OMIM:616863); Hao-Fountain syndrome due to 16p13.2 microdeletion (ORPHA:500055); ORPHA:643538
USP9X	Intellectual developmental disorder, X-linked 99 (OMIM:300919); Intellectual developmental disorder, X-linked 99, syndromic, female-restricted (OMIM:300968); X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability (ORPHA:480880); ORPHA:777
USP9Y	Spermatogenic failure, Y-linked, 2 (OMIM:415000); Chromosome Y microdeletion syndrome (ORPHA:1646)
UST	—
UTF1	—
UTP14C	—
UTP23	—
UTP4	ORPHA:168583
UTRN	—
UTS2B	—
UVSSA	UV-sensitive syndrome 3 (OMIM:614640); ORPHA:178338
VAMP7	46,XY partial gonadal dysgenesis (ORPHA:251510)
VANGL1	Sacral defect with anterior meningocele (OMIM:600145); Neural tube defects, susceptibility to (OMIM:182940); Caudal regression syndrome (ORPHA:3027)
VANGL2	Neural tube defects, susceptibility to (OMIM:182940); Isolated exencephaly (ORPHA:563612); Isolated anencephaly (ORPHA:563609)
VAPB	Amyotrophic lateral sclerosis 8 (OMIM:608627); Spinal muscular atrophy, late-onset, Finkel type (OMIM:182980); Amyotrophic lateral sclerosis (ORPHA:803); Autosomal dominant adult-onset proximal spinal muscular atrophy (ORPHA:209335)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
VAX1	Microphthalmia, syndromic 11 (OMIM:614402)
VAX2	—
VCAM1	—
VCAN	Wagner vitreoretinopathy (OMIM:143200); Wagner disease (ORPHA:898)
VCF1	—
VCL	Cardiomyopathy, dilated, 1W (OMIM:611407); Cardiomyopathy, familial hypertrophic, 15 (OMIM:613255); Familial isolated dilated cardiomyopathy (ORPHA:154)
VCP	Charcot-Marie-Tooth disease, axonal, type 2Y (OMIM:616687); Inclusion body myopathy with early-onset paget disease with or without frontotemporal dementia 1 (OMIM:167320); Frontotemporal dementia and/or amyotrophic lateral sclerosis 6 (OMIM:613954); Inclusion body myopathy with Paget disease of bone and frontotemporal dementia (ORPHA:52430); Autosomal dominant Charcot-Marie-Tooth disease type 2Y (ORPHA:435387); Behavioral variant of frontotemporal dementia (ORPHA:275864); Frontotemporal dementia with motor neuron disease (ORPHA:275872); Adult-onset distal myopathy due to VCP mutation (ORPHA:329478); Spastic paraplegia-Paget disease of bone syndrome (ORPHA:329475); Amyotrophic lateral sclerosis (ORPHA:803); Progressive non-fluent aphasia (ORPHA:100070)
VCX3A	—
VDR	Rickets, vitamin D-resistant, type IIA (OMIM:277440); Hypocalcemic vitamin D-resistant rickets (ORPHA:93160)
VEGFA	OMIM:603933
VEGFC	Lymphatic malformation 4 (OMIM:615907); ORPHA:569821
vert	—
VGLL4	—
VHL	PHEOCHROMOCYTOMA (OMIM:171300); von Hippel-Lindau syndrome (OMIM:193300); Renal cell carcinoma, nonpapillary (OMIM:144700); Erythrocytosis, familial, 2 (OMIM:263400); ORPHA:238557; Von Hippel-Lindau disease (ORPHA:892); Sporadic pheochromocytoma/secreting paraganglioma (ORPHA:276621); Hereditary pheochromocytoma-paraganglioma (ORPHA:29072)
VIM	Cataract 30, multiple types (OMIM:116300); ORPHA:98984; ORPHA:675396
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2 (OMIM:613404); Arthrogryposis-renal dysfunction-cholestasis syndrome (ORPHA:2697)
VKORC1	Coumarin resistance (OMIM:122700); Vitamin K-dependent clotting factors, combined deficiency of, 2 (OMIM:607473); Hereditary combined deficiency of vitamin K-dependent clotting factors (ORPHA:98434)
VLDLR	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1 (OMIM:224050); Dysequilibrium syndrome (ORPHA:1766)
VMA21	Myopathy, X-linked, with excessive autophagy (OMIM:310440); ORPHA:25980
VNN1	—
VPS13A	Choreoacanthocytosis (OMIM:200150); Choreoacanthocytosis (ORPHA:2388)
VPS13B	Cohen syndrome (OMIM:216550); Cohen syndrome (ORPHA:193)
VPS33B	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive (OMIM:620009); Cholestasis, progressive familial intrahepatic, 12 (OMIM:620010); Arthrogryposis, renal dysfunction, and cholestasis 1 (OMIM:208085); Arthrogryposis-renal dysfunction-cholestasis syndrome (ORPHA:2697)
VPS35	Parkinson disease 17 (OMIM:614203); Hereditary late-onset Parkinson disease (ORPHA:411602)
VPS37A	Spastic paraplegia 53, autosomal recessive (OMIM:614898); Autosomal recessive spastic paraplegia type 53 (ORPHA:319199)
VPS54	—
VRK1	Neuronopathy, distal hereditary motor, autosomal recessive 10 (OMIM:620542); Pontocerebellar hypoplasia type 1A (OMIM:607596); ORPHA:423894; Pontocerebellar hypoplasia type 1 (ORPHA:2254)
VRK2	—
VSIG4	—
VSX1	Craniofacial anomalies and anterior segment dysgenesis syndrome (OMIM:614195); Keratoconus 1 (OMIM:148300); Posterior polymorphous corneal dystrophy (ORPHA:98973)
VSX2	Microphthalmia with coloboma 3 (OMIM:610092); Microphthalmia, isolated 2 (OMIM:610093); ORPHA:98938
VWF	von Willebrand disease, type 1 (OMIM:193400); von Willebrand disease, type 3 (OMIM:277480); Von willebrand disease, type 2 (OMIM:613554); ORPHA:166078; ORPHA:166084; ORPHA:166087; ORPHA:166090; ORPHA:166096; ORPHA:166093
WAC	Desanto-Shinawi syndrome (OMIM:616708); Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion (ORPHA:284169); Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to WAC point mutation (ORPHA:466950)
WAS	Wiskott-Aldrich syndrome (OMIM:301000); Neutropenia, severe congenital, X-linked (OMIM:300299); Thrombocytopenia 1 (OMIM:313900); X-linked severe congenital neutropenia (ORPHA:86788); ORPHA:852; Wiskott-Aldrich syndrome (ORPHA:906)
WASF3	—
WASHC4	Intellectual developmental disorder, autosomal recessive 43 (OMIM:615817); ORPHA:88616
WASHC5	Spastic paraplegia 8, autosomal dominant (OMIM:603563); Ritscher-Schinzel syndrome 1 (OMIM:220210); Autosomal dominant spastic paraplegia type 8 (ORPHA:100989); 3C syndrome (ORPHA:7)
WDFY3	Microcephaly 18, primary, autosomal dominant (OMIM:617520); ORPHA:528084
WDPCP	Congenital heart defects, hamartomas of tongue, and polysyndactyly (OMIM:217085); Bardet-Biedl syndrome 15 (OMIM:615992); Bardet-Biedl syndrome (ORPHA:110); Heart defect-tongue hamartoma-polysyndactyly syndrome (ORPHA:1338)
WDR11	Hypogonadotropic hypogonadism 7 without anosmia (OMIM:146110); Intellectual developmental disorder,

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
	autosomal recessive 78 (OMIM:620237); Hypogonadotropic hypogonadism 14 with or without anosmia (OMIM:614858); Pituitary stalk interruption syndrome (ORPHA:95496); Kallmann syndrome (ORPHA:478); Normosmic congenital hypogonadotropic hypogonadism (ORPHA:432)
WDR13	—
WDR19	Senior-Loken syndrome 8 (OMIM:616307); Cranioectodermal dysplasia 4 (OMIM:614378); Short-Rib thoracic dysplasia 5 with or without polydactyly (OMIM:614376); Nephronophthisis 13 (OMIM:614377); Spermatogenic failure 72 (OMIM:619867); Senior-Loken syndrome (ORPHA:3156); Cranioectodermal dysplasia (ORPHA:1515); Jeune syndrome (ORPHA:474); ORPHA:93592
WDR3	—
WDR35	Short-Rib thoracic dysplasia 7 with or without polydactyly (OMIM:614091); Cranioectodermal dysplasia 2 (OMIM:613610); Cranioectodermal dysplasia (ORPHA:1515); Short rib-polydactyly syndrome, Verma-Naumoff type (ORPHA:93271); ORPHA:498497
WDR36	Glaucoma 1, open angle, G (OMIM:609887)
WDR4	Microcephaly, growth deficiency, seizures, and brain malformations (OMIM:618346); Galloway-Mowat syndrome 6 (OMIM:618347); Galloway-Mowat syndrome (ORPHA:2065)
WDR41	—
WDR45	Neurodegeneration with brain iron accumulation 5 (OMIM:300894); Beta-propeller protein-associated neurodegeneration (ORPHA:329284); Infantile epileptic spasms syndrome (ORPHA:697160)
WDR45B	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures (OMIM:617977)
WDR5B	—
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations (OMIM:604317); Autosomal recessive primary microcephaly (ORPHA:2512)
WDR72	Amelogenesis imperfecta, hypomaturation type, iia3 (OMIM:613211); ORPHA:100033; ORPHA:402041
WDR81	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2 (OMIM:610185); Hydrocephalus, congenital, 3, with brain anomalies (OMIM:617967); ORPHA:269505; ORPHA:269510; Dysequilibrium syndrome (ORPHA:1766)
WFS1	Deafness, autosomal dominant 6 (OMIM:600965); Cataract, nuclear total (OMIM:116400); Wolfram-Like syndrome, autosomal dominant (OMIM:614296); Wolfram syndrome 1 (OMIM:222300); ORPHA:90635; ORPHA:98991; Wolfram-like syndrome (ORPHA:411590); Wolfram syndrome (ORPHA:3463)
WHRN	Usher syndrome, type IID (OMIM:611383); Deafness, autosomal recessive 31 (OMIM:607084); ORPHA:90636; Usher syndrome type 2 (ORPHA:231178)
WIPF1	Wiskott-Aldrich syndrome 2 (OMIM:614493); ORPHA:714493
WNK1	Neuropathy, hereditary sensory and autonomic, type II (OMIM:201300); Pseudohypoaldosteronism, type IIC (OMIM:614492); Hereditary sensory and autonomic neuropathy type 2 (ORPHA:970); ORPHA:88940
WNK4	Pseudohypoaldosteronism, type IIB (OMIM:614491); ORPHA:88939
WNT10A	Odontoonychodermal dysplasia (OMIM:257980); Schopf-Schulz-Passarge syndrome (OMIM:224750); Tooth agenesis, selective, 4 (OMIM:150400); ORPHA:2721; Autosomal recessive hypohidrotic ectodermal dysplasia (ORPHA:248); Schöpf-Schulz-Passarge syndrome (ORPHA:50944); Oligodontia (ORPHA:99798)
WNT10B	Split-Hand/foot malformation 6 (OMIM:225300); Tooth agenesis, selective, 8 (OMIM:617073); Isolated split hand-split foot malformation (ORPHA:2440); Oligodontia (ORPHA:99798)
WNT3	Tetraamelia, autosomal recessive (OMIM:273395); Tetraamelia-multiple malformations syndrome (ORPHA:3301)
WNT4	Mullerian aplasia and hyperandrogenism (OMIM:158330); 46,XX sex reversal with dysgenesis of kidneys, adrenals, and lungs (OMIM:611812); Mayer-Rokitansky-Küster-Hauser syndrome type 2 (ORPHA:2578); SERKAL syndrome (ORPHA:139466); Müllerian aplasia and hyperandrogenism (ORPHA:247768)
WNT5A	Robinow syndrome, autosomal dominant 1 (OMIM:180700); Autosomal dominant Robinow syndrome (ORPHA:3107)
WNT7A	Ulna and fibula, absence of, with severe limb deficiency (OMIM:276820); Santos syndrome (OMIM:613005); Fibular aplasia or hypoplasia, femoral bowing and poly-, syn-, and oligodactyly (OMIM:228930); ORPHA:2854; Phocomelia, Schinzel type (ORPHA:2879)
WRAP53	Dyskeratosis congenita, autosomal recessive, 3 (OMIM:613988); Dyskeratosis congenita (ORPHA:1775)
WRN	Werner syndrome (OMIM:277700); Werner syndrome (ORPHA:902)
WT1	Frasier syndrome (OMIM:136680); Nephrotic syndrome, type 4 (OMIM:256370); Denys-Drash syndrome (OMIM:194080); Wilms tumor, aniridia, genitourinary anomalies and impaired intellectual development syndrome (OMIM:194072); Wilms tumor 1 (OMIM:194070); Mesothelioma, malignant (OMIM:156240); Meacham syndrome (OMIM:608978); Aniridia (OMIM:106210); Frasier syndrome (ORPHA:347); Desmoplastic small round cell tumor (ORPHA:83469); 46,XY partial gonadal dysgenesis (ORPHA:251510); Denys-Drash syndrome (ORPHA:220); 46,XY complete gonadal dysgenesis (ORPHA:242); Meacham syndrome (ORPHA:3097); WAGR syndrome (ORPHA:893); Nephroblastoma (ORPHA:654); Hereditary steroid-resistant nephrotic syndrome (ORPHA:656)
WTAPP1	—
WWC1	OMIM:615602
WWOX	Developmental and epileptic encephalopathy 28 (OMIM:616211); Spinocerebellar ataxia, autosomal recessive 12 (OMIM:614322); Esophageal cancer, somatic (OMIM:133239); 46,XY partial gonadal dysgenesis (ORPHA:251510); Squamous cell carcinoma of the esophagus (ORPHA:99977); ORPHA:708171; Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency (ORPHA:284282)
WWTR1	ORPHA:157791; ORPHA:673556

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
XBP1	OMIM:612371
XDH	Xanthinuria, type I (OMIM:278300); ORPHA:93601
XG	—
XGY2	—
XIAP	Lymphoproliferative syndrome, X-linked, 1 (OMIM:308240); Lymphoproliferative syndrome, X-linked, 2 (OMIM:300635); X-linked lymphoproliferative disease due to XIAP deficiency (ORPHA:538934)
XK	Mcleod syndrome (OMIM:300842); ORPHA:59306
XKR6	—
XPA	Xeroderma pigmentosum, complementation group A (OMIM:278700); Xeroderma pigmentosum (ORPHA:910)
XPC	Xeroderma pigmentosum, group C (OMIM:278720); Xeroderma pigmentosum (ORPHA:910)
XPNPEP2	Acquired angioedema (OMIM:300909); Renin-angiotensin-aldosterone system-blocker-induced angioedema (ORPHA:100057)
XPNPEP3	Nephronophthisis-Like nephropathy 1 (OMIM:613159); ORPHA:93589
XRCC1	Spinocerebellar ataxia, autosomal recessive 26 (OMIM:617633)
XRCC2	Premature ovarian failure 17 (OMIM:619146); Spermatogenic failure 50 (OMIM:619145); FANCONI ANEMIA, COMPLEMENTATION GROUP U (OMIM:617247); Fanconi anemia (ORPHA:84); Male infertility with azoospermia or oligozoospermia due to single gene mutation (ORPHA:399805); ORPHA:227535
XRCC3	OMIM:613972; Breast cancer (OMIM:114480)
XRCC4	Short stature, microcephaly, and endocrine dysfunction (OMIM:616541); LIG4 syndrome (ORPHA:99812); Microcephalic primordial dwarfism-insulin resistance syndrome (ORPHA:436182)
XRCC5	—
XRCC6	—
XYLT1	Pseudoxanthoma elasticum (OMIM:264800); Desbuquois dysplasia 2 (OMIM:615777); XYLT1-CDG (ORPHA:370930); Desbuquois syndrome (ORPHA:1425)
XYLT2	Spondyloocular syndrome (OMIM:605822); Pseudoxanthoma elasticum (OMIM:264800); Spondylo-ocular syndrome (ORPHA:85194)
YAE1	—
YARS1	Charcot-Marie-Tooth disease, dominant intermediate C (OMIM:608323); Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2 (OMIM:619418); ORPHA:100045
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2 (OMIM:613561); Mitochondrial myopathy and sideroblastic anemia (ORPHA:2598)
YBX2	—
YWHAE	17p13.3 microduplication syndrome (ORPHA:217385); ORPHA:213711; ORPHA:261257; Miller-Dieker syndrome (ORPHA:531); ORPHA:457246
YY1	Gabriele-De vries syndrome (OMIM:617557); Insulinoma (ORPHA:97279); Gabriele-de Vries syndrome (ORPHA:506358)
ZAN	—
ZAP70	Immunodeficiency 48 (OMIM:269840); Autoimmune disease, multisystem, infantile-onset, 2 (OMIM:617006); Combined immunodeficiency due to ZAP70 deficiency (ORPHA:911)
ZBTB16	ORPHA:99861; Acute promyelocytic leukemia (ORPHA:520)
ZBTB18	Intellectual developmental disorder, autosomal dominant 22 (OMIM:612337); Distal deletion 1q syndrome (ORPHA:36367)
ZBTB24	Immunodeficiency-Centromeric instability-facial anomalies syndrome2 (OMIM:614069); ICF syndrome (ORPHA:2268)
ZBTB25	—
ZBTB40	—
ZBTB41	—
ZBTB7A	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin (OMIM:619769); ORPHA:694956
ZC3H14	Intellectual developmental disorder, autosomal recessive 56 (OMIM:617125); ORPHA:88616
ZCCHC12	—
ZCCHC13	—
ZCCHC7	—
ZCCHC8	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 5 (OMIM:618674)
ZDHHC14	—
ZDHHC15	—
ZDHHC17	—
ZDHHC2	—
ZDHHC24	—
ZDHHC6	—
ZDHHC9	Intellectual developmental disorder, X-linked syndromic, Raymond type (OMIM:300799); Lujan-Fryns syndrome (ORPHA:776)
ZEB1	Corneal dystrophy, posterior polymorphous, 3 (OMIM:609141); Corneal dystrophy, fuchs endothelial, 6 (OMIM:613270); Posterior polymorphous corneal dystrophy (ORPHA:98973); Fuchs endothelial corneal dystrophy (ORPHA:98974)
ZEB2	Mowat-Wilson syndrome (OMIM:235730); Mowat-Wilson syndrome due to a ZEB2 point mutation (ORPHA:261552); Mowat-Wilson syndrome due to monosomy 2q22 (ORPHA:261537); Large/giant congenital melanocytic nevus (ORPHA:626)
ZFAT	Autoimmune thyroid disease, susceptibility to, 3 (OMIM:608175)

Gene	Malattie associate (HPO / OMIM / Orphanet / DECIPHER)
ZFHX3	Atrial fibrillation 8, susceptibility to (OMIM:613055); Epilepsy, idiopathic generalized 20 (OMIM:621500); Prostate cancer (OMIM:176807); Spinocerebellar ataxia 4 (OMIM:600223)
ZFHX4	ORPHA:91411
ZFP36	—
ZFP36L1	—
ZFP36L2	Oocyte maturation defect 13 (OMIM:620154); Female infertility due to oocyte meiotic arrest (ORPHA:488191); ORPHA:675396
ZFP69	—
ZFP90	—
ZFP91	—
ZFPM2	Diaphragmatic hernia 3 (OMIM:610187); Tetralogy of Fallot (OMIM:187500); 46,xy sex reversal 9 (OMIM:616067); 46,XY partial gonadal dysgenesis (ORPHA:251510); Tetralogy of Fallot (ORPHA:3303); Congenital diaphragmatic hernia (ORPHA:2140)
ZFYVE26	Spastic paraplegia 15, autosomal recessive (OMIM:270700); Kjellin syndrome (ORPHA:100996)
ZFYVE27	—
ZHX3	—
ZIC1	Craniosynostosis 6 (OMIM:616602); Structural brain anomalies with impaired intellectual development and craniosynostosis (OMIM:618736); Non-syndromic bicoronal craniosynostosis (ORPHA:35099); ORPHA:269212; ORPHA:269215
ZIC2	Holoprosencephaly 5 (OMIM:609637); Septopreoptic holoprosencephaly (ORPHA:280195); Semilobar holoprosencephaly (ORPHA:220386); Microform holoprosencephaly (ORPHA:280200); Midline interhemispheric variant of holoprosencephaly (ORPHA:93926); Lobar holoprosencephaly (ORPHA:93924); Alobar holoprosencephaly (ORPHA:93925)
ZIC3	VACTERL association, X-linked (OMIM:314390); Heterotaxy, visceral, 1, X-linked (OMIM:306955); ORPHA:216718; ORPHA:157769
ZIC4	ORPHA:269212; ORPHA:269215
ZKSCAN5	—
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy (OMIM:608612); Restrictive dermopathy 1 (OMIM:275210); Hutchinson-Gilford progeria syndrome (ORPHA:740); Mandibuloacral dysplasia with type B lipodystrophy (ORPHA:90154); Restrictive dermopathy (ORPHA:1662)
ZMYM3	Intellectual developmental disorder, X-linked 112 (OMIM:301111)
ZNF175	—
ZNF213	—
ZNF276	—
ZNF300	—
ZNF335	Microcephaly 10, primary, autosomal recessive (OMIM:615095); Microcephalic primordial dwarfism due to ZNF335 deficiency (ORPHA:329228)
ZNF341	Hyper-IgE recurrent infection syndrome 3, autosomal recessive (OMIM:618282); ORPHA:641368
ZNF365	OMIM:605990; Narcolepsy type 2 (ORPHA:83465); Narcolepsy type 1 (ORPHA:2073)
ZNF384	—
ZNF41	—
ZNF419	—
ZNF420	—
ZNF423	Nephronophthisis 14 (OMIM:614844); Joubert syndrome with oculorenal defect (ORPHA:2318); ORPHA:93591
ZNF433	—
ZNF469	Brittle cornea syndrome (OMIM:229200); Brittle cornea syndrome (ORPHA:90354)
ZNF480	—
ZNF513	Retinitis pigmentosa 58 (OMIM:613617); Retinitis pigmentosa (ORPHA:791)
ZNF518A	—
ZNF526	Dentici-Novelli neurodevelopmental syndrome (OMIM:619877); ORPHA:528084
ZNF575	—
ZNF585A	—
ZNF592	CAMOS syndrome (ORPHA:83472)
ZNF627	—
ZNF644	Myopia 21, autosomal dominant (OMIM:614167)
ZNF674	—
ZNF687	Paget disease of bone 6 (OMIM:616833)
ZNF691	—
ZNF711	Intellectual developmental disorder, X-linked 97 (OMIM:300803); ORPHA:777
ZNF750	Seborrhea-Like dermatitis with psoriasiform elements (OMIM:610227); ORPHA:168606
ZNF75D	—
ZNF80	—
ZNF81	ORPHA:777
ZPBP	Spermatogenic failure 66 (OMIM:619799); ORPHA:171709
ZPBP2	—
ZRANB2	—
ZRANB3	—
ZRSR2	Orofaciodigital syndrome XXI (OMIM:301132)