

Gene_symbol	Disease_name	Disease_ID	Association_type
A2M	—		UNKNOWN
A4GALT	#111400 BLOOD GROUP, P1PK SYSTEMP(1) PHENOTYPE, INCLUDED;;P(2) PHENOTYPE, INCLUDED;;P1(k) PHENOTYPE, INCLUDED;;P2(k) PHENOTYPE, INCLUDED;;p PHENOTYPE, INCLUDED;;NOR POLYAGGLUTINATION SYNDROME, INCLUDED	OMIM:111400	MENDELIAN
A4GNT	—		UNKNOWN
AAAS	Achalasia-Addisonianism-Alacrima syndrome	OMIM:231550	MENDELIAN
AAAS	Triple A syndrome	ORPHA:869	UNKNOWN
AADAC	—		UNKNOWN
AADACL2	—		UNKNOWN
AAGAB	Keratoderma, palmoplantar, punctate type 1A	OMIM:148600	MENDELIAN
AAGAB	Punctate palmoplantar keratoderma type 1	ORPHA:79501	UNKNOWN
AANAT	—		UNKNOWN
AARS	—		UNKNOWN
AARS2	Combined oxidative phosphorylation deficiency 8	OMIM:614096	MENDELIAN
AARS2	Leukoencephalopathy, progressive, with ovarian failure	OMIM:615889	MENDELIAN
AASS	Hyperlysinemia, type I	OMIM:238700	MENDELIAN
AASS	Saccharopinuria	ORPHA:3124	UNKNOWN
AASS	Hyperlysinemia	ORPHA:2203	UNKNOWN
ABAT	GABA-transaminase deficiency	OMIM:613163	MENDELIAN
ABAT	Gamma-aminobutyric acid transaminase deficiency	ORPHA:2066	UNKNOWN
ABCA1	Tangier disease	OMIM:205400	MENDELIAN
ABCA1	HDL deficiency, familial, 1	OMIM:604091	MENDELIAN
ABCA1	Tangier disease	ORPHA:31150	UNKNOWN
ABCA1	Apolipoprotein A-I deficiency	ORPHA:425	UNKNOWN
ABCA10	—		UNKNOWN
ABCA12	Ichthyosis, congenital, autosomal recessive 4B	OMIM:242500	MENDELIAN
ABCA12	Ichthyosis, congenital, autosomal recessive 4A	OMIM:601277	MENDELIAN
ABCA12	Lamellar ichthyosis	ORPHA:313	UNKNOWN
ABCA12	Congenital ichthyosiform erythroderma	ORPHA:79394	UNKNOWN
ABCA12	Harlequin ichthyosis	ORPHA:457	UNKNOWN
ABCA13	—		UNKNOWN
ABCA2	Intellectual developmental disorder with poor growth and with or without seizures or ataxia	OMIM:618808	MENDELIAN
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3	OMIM:610921	MENDELIAN
ABCA3	Pediatric acute respiratory distress syndrome	ORPHA:685082	UNKNOWN
ABCA3	Idiopathic pulmonary fibrosis	ORPHA:2032	UNKNOWN
ABCA4	Stargardt disease 1	OMIM:248200	MENDELIAN
ABCA4	Macular degeneration, age-related, 2	OMIM:153800	POLYGENIC
ABCA4	Cone-rod dystrophy 3	OMIM:604116	MENDELIAN
ABCA4	Retinitis pigmentosa 19	OMIM:601718	MENDELIAN
ABCA4	Cone rod dystrophy	ORPHA:1872	UNKNOWN
ABCA4	Retinitis pigmentosa	ORPHA:791	UNKNOWN
ABCA4	Stargardt disease	ORPHA:827	UNKNOWN
ABCA5	Hypertrichosis terminalis, generalized, with or without gingival hyperplasia	OMIM:135400	MENDELIAN
ABCA5	Gingival fibromatosis-hypertrichosis syndrome	ORPHA:2026	UNKNOWN
ABCA7	Alzheimer disease 9, susceptibility to	OMIM:608907	POLYGENIC
ABCB1	Encephalopathy, acute transient	OMIM:620950	MENDELIAN
ABCB1	Inflammatory bowel disease 13	OMIM:612244	POLYGENIC
ABCB11	Cholestasis, progressive familial intrahepatic, 3	OMIM:602347	MENDELIAN
ABCB11	Cholestasis, benign recurrent intrahepatic, 2	OMIM:605479	MENDELIAN
ABCB11	Cholestasis, progressive familial intrahepatic 2	OMIM:601847	MENDELIAN
ABCB11	Intrahepatic cholestasis of pregnancy	ORPHA:69665	UNKNOWN
ABCB4	Gallbladder disease 1	OMIM:600803	MENDELIAN
ABCB4	Cholestasis, progressive familial intrahepatic, 3	OMIM:602347	MENDELIAN
ABCB4	Cholestasis, intrahepatic, of pregnancy 3	OMIM:614972	MENDELIAN
ABCB4	Low phospholipid-associated cholelithiasis	ORPHA:69663	UNKNOWN
ABCB4	Intrahepatic cholestasis of pregnancy	ORPHA:69665	UNKNOWN
ABCB5	—		UNKNOWN
ABCB6	Pseudohyperkalemia, familial, 2, due to red cell leak	OMIM:609153	MENDELIAN
ABCB6	Dyschromatosis universalis hereditaria 3	OMIM:615402	MENDELIAN
ABCB6	Microphthalmia, isolated, with coloboma 7	OMIM:614497	MENDELIAN
ABCB6	Dyschromatosis universalis hereditaria	ORPHA:241	UNKNOWN
ABCB6	Familial pseudohyperkalemia	ORPHA:90044	UNKNOWN
ABCB7	Anemia, sideroblastic, and spinocerebellar ataxia	OMIM:301310	MENDELIAN
ABCB7	X-linked sideroblastic anemia and spinocerebellar ataxia	ORPHA:2802	UNKNOWN
ABCC1	Deafness, autosomal dominant 77	OMIM:618915	MENDELIAN

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ABCC12	—		UNKNOWN
ABCC2	DUBIN-JOHNSON syndrome	OMIM:237500	MENDELIAN
ABCC2	Dubin-Johnson syndrome	ORPHA:234	UNKNOWN
ABCC3	—		UNKNOWN
ABCC4	—		UNKNOWN
ABCC6	Pseudoxanthoma elasticum	OMIM:264800	MENDELIAN
ABCC6	Pseudoxanthoma elasticum, forme fruste	OMIM:177850	MENDELIAN
ABCC6	Arterial calcification, generalized, of infancy, 1	OMIM:208000	MENDELIAN
ABCC6	Arterial calcification, generalized, of infancy, 2	OMIM:614473	MENDELIAN
ABCC6	Pseudoxanthoma elasticum	ORPHA:758	UNKNOWN
ABCC6	Generalized arterial calcification of infancy	ORPHA:51608	UNKNOWN
ABCC8	Diabetes mellitus, transient neonatal 2	OMIM:610374	MENDELIAN
ABCC8	Hyperinsulinemic hypoglycemia, familial, 1	OMIM:256450	MENDELIAN
ABCC8	Diabetes mellitus, noninsulin-dependent	OMIM:125853	MENDELIAN
ABCC8	Hypoglycemia of infancy, leucine-sensitive	OMIM:240800	MENDELIAN
ABCC8	Diabetes mellitus, permanent neonatal 3, with or without neurologic features	OMIM:618857	MENDELIAN
ABCC8	Maturity-onset diabetes of the young, type 12	OMIM:621196	MENDELIAN
ABCC8	Isolated permanent neonatal diabetes mellitus	ORPHA:99885	UNKNOWN
ABCC8	Transient neonatal diabetes mellitus	ORPHA:99886	UNKNOWN
ABCC8	Autosomal dominant hyperinsulinism due to SUR1 deficiency	ORPHA:276575	UNKNOWN
ABCC8	MODY	ORPHA:552	UNKNOWN
ABCC8	DEND syndrome	ORPHA:79134	UNKNOWN
ABCC9	Hypertrichotic osteochondrodysplasia	OMIM:239850	MENDELIAN
ABCC9	Cardiomyopathy, dilated, 10	OMIM:608569	MENDELIAN
ABCC9	Atrial fibrillation, familial, 12	OMIM:614050	MENDELIAN
ABCC9	Intellectual disability and myopathy syndrome	OMIM:619719	MENDELIAN
ABCC9	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
ABCC9	CantÃ³ syndrome	ORPHA:1517	UNKNOWN
ABCC9	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
ABCC9	Brugada syndrome	ORPHA:130	UNKNOWN
ABCD1	Adrenoleukodystrophy	OMIM:300100	MENDELIAN
ABCD1	Hirschsprung disease	ORPHA:388	UNKNOWN
ABCD1	CADD5	ORPHA:369942	UNKNOWN
ABCD1	Adrenomyeloneuropathy	ORPHA:139399	UNKNOWN
ABCD1	X-linked cerebral adrenoleukodystrophy	ORPHA:139396	UNKNOWN
ABCD3	Bile acid synthesis defect, congenital, 5	OMIM:616278	MENDELIAN
ABCD3	Oculopharyngodistal myopathy 5	OMIM:621446	MENDELIAN
ABCD3	Oculopharyngodistal myopathy	ORPHA:98897	UNKNOWN
ABCD4	Methylmalonic aciduria and homocystinuria, Cblj type	OMIM:614857	MENDELIAN
ABCE1	—		UNKNOWN
ABCG1	—		UNKNOWN
ABCG2	Uric acid concentration, serum, QTL1	OMIM:138900	MENDELIAN
ABCG5	Sitosterolemia 2	OMIM:618666	MENDELIAN
ABCG5	Sitosterolemia	ORPHA:2882	UNKNOWN
ABCG5	Homozygous familial hypercholesterolemia	ORPHA:391665	UNKNOWN
ABCG8	Gallbladder disease 4	OMIM:611465	POLYGENIC
ABCG8	Sitosterolemia 1	OMIM:210250	MENDELIAN
ABCG8	Sitosterolemia	ORPHA:2882	UNKNOWN
ABCG8	Homozygous familial hypercholesterolemia	ORPHA:391665	UNKNOWN
ABHD1	—		UNKNOWN
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract	OMIM:612674	MENDELIAN
ABHD5	Chanarin-Dorfman syndrome	OMIM:275630	MENDELIAN
ABHD5	Neutral lipid storage disease with ichthyosis	ORPHA:98907	UNKNOWN
ABI3BP	—		UNKNOWN
ABL1	Congenital heart defects and skeletal malformations syndrome	OMIM:617602	MENDELIAN
ABL1	Leukemia, chronic myeloid	OMIM:608232	MENDELIAN
ABL1	Chronic myeloid leukemia	ORPHA:521	UNKNOWN
ABL2	—		UNKNOWN
ACACA	Acetyl-CoA carboxylase deficiency	OMIM:613933	MENDELIAN
ACACB	—		UNKNOWN
ACAD10	—		UNKNOWN
ACAD11	—		UNKNOWN
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	OMIM:611283	MENDELIAN
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	ORPHA:79159	UNKNOWN
ACAD9	Mitochondrial complex I deficiency, nuclear type 20	OMIM:611126	MENDELIAN

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ACAD9	Acyl-CoA dehydrogenase 9 deficiency	ORPHA:99901	UNKNOWN
ACADL	—		UNKNOWN
ACADM	Acyl-CoA dehydrogenase, medium-chain, deficiency of	OMIM:201450	MENDELIAN
ACADM	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42	UNKNOWN
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of	OMIM:201470	MENDELIAN
ACADS	Short chain acyl-CoA dehydrogenase deficiency	ORPHA:26792	UNKNOWN
ACADSB	2-Methylbutyryl-CoA dehydrogenase deficiency	OMIM:610006	MENDELIAN
ACADSB	2-methylbutyryl-CoA dehydrogenase deficiency	ORPHA:79157	UNKNOWN
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency	OMIM:201475	MENDELIAN
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency	ORPHA:26793	UNKNOWN
ACAN	Spondyloepimetaphyseal dysplasia, Aggrecan type	OMIM:612813	MENDELIAN
ACAN	Spondyloepiphyseal dysplasia, Kimberley type	OMIM:608361	MENDELIAN
ACAN	Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans	OMIM:165800	MENDELIAN
ACAN	Spondyloepimetaphyseal dysplasia, aggrecan type	ORPHA:171866	UNKNOWN
ACAN	Spondyloepiphyseal dysplasia, Kimberley type	ORPHA:93283	UNKNOWN
ACAN	Short stature-advanced bone age-early-onset osteoarthritis syndrome	ORPHA:435804	UNKNOWN
ACAT1	Alpha-methylacetoacetic aciduria	OMIM:203750	MENDELIAN
ACAT1	Beta-ketothiolase deficiency	ORPHA:134	UNKNOWN
ACAT2	Acetyl-CoA acetyltransferase-2 deficiency	OMIM:614055	MENDELIAN
ACBD5	Retinal dystrophy with leukodystrophy	OMIM:618863	MENDELIAN
ACBD6	Neurodevelopmental disorder with progressive movement abnormalities	OMIM:620785	MENDELIAN
ACCS	—		UNKNOWN
ACE	Renal tubular dysgenesis	OMIM:267430	MENDELIAN
ACKR3	Oculomotor-abducens synkinesis	OMIM:619215	MENDELIAN
ACLY	—		UNKNOWN
ACMSD	—		UNKNOWN
ACN9	—		UNKNOWN
ACO2	Optic atrophy 9	OMIM:616289	MENDELIAN
ACO2	Infantile cerebellar-retinal degeneration	OMIM:614559	MENDELIAN
ACOX1	Mitchell syndrome	OMIM:618960	MENDELIAN
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	OMIM:264470	MENDELIAN
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	ORPHA:2971	UNKNOWN
ACP1	—		UNKNOWN
ACP5	Spondyloenchondrodysplasia with immune dysregulation	OMIM:607944	MENDELIAN
ACP5	Spondyloenchondrodysplasia	ORPHA:1855	UNKNOWN
ACP6	—		UNKNOWN
ACR	Spermatogenic failure 87	OMIM:620500	MENDELIAN
ACSF3	Combined malonic and methylmalonic aciduria	OMIM:614265	MENDELIAN
ACSF3	Combined malonic and methylmalonic acidemia	ORPHA:289504	UNKNOWN
ACSL4	Intellectual developmental disorder, X-linked 63	OMIM:300387	MENDELIAN
ACSL4	Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome	ORPHA:86818	UNKNOWN
ACSL5	Diarrhea 13	OMIM:620357	MENDELIAN
ACSL6	—		UNKNOWN
ACSM2B	—		UNKNOWN
ACSM3	—		UNKNOWN
ACTA1	Nemaline myopathy 3	OMIM:161800	MENDELIAN
ACTA1	Myopathy, scapulohumeroperoneal	OMIM:616852	MENDELIAN
ACTA1	Congenital myopathy 2B, severe infantile, autosomal recessive	OMIM:620265	MENDELIAN
ACTA1	Congenital myopathy 2C, severe infantile, autosomal dominant	OMIM:620278	MENDELIAN
ACTA1	Zebra body myopathy	ORPHA:97240	UNKNOWN
ACTA1	Rigid spine syndrome	ORPHA:97244	UNKNOWN
ACTA1	Congenital fiber-type disproportion myopathy	ORPHA:2020	UNKNOWN
ACTA1	Typical nemaline myopathy	ORPHA:171436	UNKNOWN
ACTA1	Intermediate nemaline myopathy	ORPHA:171433	UNKNOWN
ACTA1	Severe congenital nemaline myopathy	ORPHA:171430	UNKNOWN
ACTA1	Childhood-onset nemaline myopathy	ORPHA:171439	UNKNOWN
ACTA2	Aortic aneurysm, familial thoracic 6	OMIM:611788	MENDELIAN
ACTA2	Moyamoya disease 5	OMIM:614042	MENDELIAN
ACTA2	Multisystemic smooth muscle dysfunction syndrome	OMIM:613834	MENDELIAN
ACTA2	Moyamoya disease	ORPHA:2573	UNKNOWN
ACTA2	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
ACTB	Baraitser-Winter syndrome 1	OMIM:243310	MENDELIAN
ACTB	Becker nevus syndrome	OMIM:604919	MENDELIAN
ACTB	Thrombocytopenia 8, with dysmorphic features and developmental delay	OMIM:620475	MENDELIAN

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ACTB	Congenital smooth muscle hamartoma with or without hemihypertrophy	OMIM:620470	MENDELIAN
ACTB	Dystonia, juvenile-onset	OMIM:607371	MENDELIAN
ACTB	Becker nevus syndrome	ORPHA:64755	UNKNOWN
ACTB	Developmental malformations-deafness-dystonia syndrome	ORPHA:79107	UNKNOWN
ACTB	Baraitser-Winter cerebrofrontofacial syndrome	ORPHA:2995	UNKNOWN
ACTC1	Atrial septal defect 5	OMIM:612794	MENDELIAN
ACTC1	Cardiomyopathy, dilated, 1R	OMIM:613424	MENDELIAN
ACTC1	Cardiomyopathy, familial hypertrophic, 11	OMIM:612098	MENDELIAN
ACTC1	Atrial septal defect, ostium secundum type	ORPHA:99103	UNKNOWN
ACTC1	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
ACTG1	Deafness, autosomal dominant 20	OMIM:604717	MENDELIAN
ACTG1	Baraitser-Winter syndrome 2	OMIM:614583	MENDELIAN
ACTG1	Baraitser-Winter cerebrofrontofacial syndrome	ORPHA:2995	UNKNOWN
ACTG2	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5	OMIM:619431	MENDELIAN
ACTG2	Visceral myopathy 1	OMIM:155310	MENDELIAN
ACTG2	Familial visceral myopathy	ORPHA:2604	UNKNOWN
ACTG2	Megacystis-microcolon-intestinal hypoperistalsis syndrome	ORPHA:2241	UNKNOWN
ACTL6B	Epileptic encephalopathy, early infantile, 76	OMIM:618468	MENDELIAN
ACTL6B	Intellectual developmental disorder with severe speech and ambulation defects	OMIM:618470	MENDELIAN
ACTL6B	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
ACTN1	Bleeding disorder, platelet-type, 15	OMIM:615193	MENDELIAN
ACTN1	Autosomal dominant macrothrombocytopenia	ORPHA:140957	UNKNOWN
ACTN2	Cardiomyopathy, dilated, 1AA, with or without LVNC	OMIM:612158	MENDELIAN
ACTN2	Myopathy, congenital with structured cores and Z-line abnormalities	OMIM:618654	MENDELIAN
ACTN2	Myopathy, distal, 6, adult onset	OMIM:618655	MENDELIAN
ACTN2	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
ACTN4	Focal segmental glomerulosclerosis 1	OMIM:603278	MENDELIAN
ACTN4	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
ACTR3C	—		UNKNOWN
ACTRT1	—		UNKNOWN
ACVR1	Fibrodysplasia ossificans progressiva	OMIM:135100	MENDELIAN
ACVR1	Fibrodysplasia ossificans progressiva	ORPHA:337	UNKNOWN
ACVR1B	Pancreatic cancer	OMIM:260350	MENDELIAN
ACVR1C	—		UNKNOWN
ACVR2A	—		UNKNOWN
ACVR2B	Heterotaxy, visceral, 4, autosomal	OMIM:613751	MENDELIAN
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2	OMIM:600376	MENDELIAN
ACVRL1	Hereditary hemorrhagic telangiectasia	ORPHA:774	UNKNOWN
ACY1	Aminoacylase 1 deficiency	OMIM:609924	MENDELIAN
ACY1	Aminoacylase 1 deficiency	ORPHA:137754	UNKNOWN
ADA	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-negative, due to adenosine deaminase deficiency	OMIM:102700	MENDELIAN
ADA	T-B-NK- severe combined immunodeficiency due to adenosine deaminase deficiency	ORPHA:277	UNKNOWN
ADA	Omenn syndrome	ORPHA:39041	UNKNOWN
ADAD2	—		UNKNOWN
ADAM10	Reticulate acropigmentation of Kitamura	OMIM:615537	MENDELIAN
ADAM12	—		UNKNOWN
ADAM17	Hypotichosis 16	OMIM:621490	MENDELIAN
ADAM17	Inflammatory skin and bowel disease, neonatal, 1	OMIM:614328	MENDELIAN
ADAM17	Neonatal erythroderma-autoinflammation-inflammatory bowel disease syndrome	ORPHA:294023	UNKNOWN
ADAM19	—		UNKNOWN
ADAM22	Epileptic encephalopathy, early infantile, 61	OMIM:617933	MENDELIAN
ADAM23	—		UNKNOWN
ADAM33	—		UNKNOWN
ADAM7	—		UNKNOWN
ADAM9	Cone-Rod dystrophy 9	OMIM:612775	MENDELIAN
ADAM9	Cone rod dystrophy	ORPHA:1872	UNKNOWN
ADAMTS1	—		UNKNOWN
ADAMTS10	Weill-Marchesani syndrome 1	OMIM:277600	MENDELIAN
ADAMTS10	Weill-Marchesani syndrome	ORPHA:3449	UNKNOWN
ADAMTS13	Thrombotic thrombocytopenic purpura, hereditary	OMIM:274150	MENDELIAN
ADAMTS16	—		UNKNOWN
ADAMTS17	Weill-Marchesani syndrome 4	OMIM:613195	MENDELIAN
ADAMTS17	Weill-Marchesani syndrome	ORPHA:3449	UNKNOWN
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus	OMIM:615458	MENDELIAN

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ADAMTS19	Cardiac valvular dysplasia 2	OMIM:620067	MENDELIAN
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	OMIM:225410	MENDELIAN
ADAMTS2	Dermatosparaxis Ehlers-Danlos syndrome	ORPHA:1901	UNKNOWN
ADAMTSL2	Geleophysic dysplasia 1	OMIM:231050	MENDELIAN
ADAMTSL2	Geleophysic dysplasia	ORPHA:2623	UNKNOWN
ADAMTSL2	Dermatosparaxis Ehlers-Danlos syndrome	ORPHA:1901	UNKNOWN
ADAMTSL3	—		UNKNOWN
ADAMTSL4	Ectopia lentis et pupillae	OMIM:225200	MENDELIAN
ADAMTSL4	Ectopia lentis	OMIM:225100	MENDELIAN
ADAMTSL4	Isolated ectopia lentis	ORPHA:1885	UNKNOWN
ADAR	Aicardi-Goutieres syndrome 6	OMIM:615010	MENDELIAN
ADAR	Dyschromatosis symmetrica hereditaria 1	OMIM:127400	MENDELIAN
ADAR	Dyschromatosis symmetrica hereditaria	ORPHA:41	UNKNOWN
ADAR	Aicardi-Goutières syndrome	ORPHA:51	UNKNOWN
ADAR	Familial infantile bilateral striatal necrosis	ORPHA:225154	UNKNOWN
ADARB1	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures	OMIM:618862	MENDELIAN
ADAT3	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies	OMIM:615286	MENDELIAN
ADAT3	Intellectual disability-strabismus syndrome	ORPHA:363528	UNKNOWN
ADC	—		UNKNOWN
ADCK3	—		UNKNOWN
ADCK4	—		UNKNOWN
ADCY1	Deafness, autosomal recessive 44	OMIM:610154	MENDELIAN
ADCY10	Hypercalciuria, absorptive, susceptibility to	OMIM:143870	POLYGENIC
ADCY10	Idiopathic hypercalciuria	ORPHA:2197	UNKNOWN
ADCY3	Obesity, susceptibility to, BMIQ19	OMIM:617885	POLYGENIC
ADCY5	Dyskinesia with orofacial involvement, autosomal recessive	OMIM:619647	MENDELIAN
ADCY5	Neurodevelopmental disorder with hyperkinetic movements and dyskinesia	OMIM:619651	MENDELIAN
ADCY5	Dyskinesia, familial, with facial myokymia	OMIM:606703	MENDELIAN
ADCY5	Benign hereditary chorea	ORPHA:1429	UNKNOWN
ADCY5	Familial dyskinesia and facial myokymia	ORPHA:324588	UNKNOWN
ADCY6	Lethal congenital contracture syndrome 8	OMIM:616287	MENDELIAN
ADCY6	Hypomyelination neuropathy-arthrogryposis syndrome	ORPHA:2680	UNKNOWN
ADCY9	—		UNKNOWN
ADCYAP1	—		UNKNOWN
ADD1	Hypertension, essential	OMIM:145500	POLYGENIC
ADD2	—		UNKNOWN
ADGRA2	—		UNKNOWN
ADGRG1	Polymicrogyria, bilateral frontoparietal	OMIM:606854	MENDELIAN
ADGRG1	Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian)	OMIM:615752	MENDELIAN
ADGRG1	Bilateral frontoparietal polymicrogyria	ORPHA:101070	UNKNOWN
ADGRG1	Bilateral perisylvian polymicrogyria	ORPHA:98889	UNKNOWN
ADGRG2	Vas deferens, congenital bilateral aplasia of, X-linked	OMIM:300985	MENDELIAN
ADGRG2	Congenital bilateral absence of vas deferens	ORPHA:48	UNKNOWN
ADGRL1	Developmental delay, behavioral abnormalities, and neuropsychiatric disorders	OMIM:620065	MENDELIAN
ADH1A	—		UNKNOWN
ADH1B	Alcohol dependence	OMIM:103780	POLYGENIC
ADH1C	Alcohol dependence	OMIM:103780	POLYGENIC
ADH1C	Parkinson disease, late-onset	OMIM:168600	POLYGENIC
ADH4	—		UNKNOWN
ADH5	AMED syndrome, digenic	OMIM:619151	MENDELIAN
ADH7	—		UNKNOWN
ADIPOQ	Adiponectin deficiency	OMIM:612556	MENDELIAN
ADIPOR1	—		UNKNOWN
ADK	Hypermethioninemia due to adenosine kinase deficiency	OMIM:614300	MENDELIAN
ADM	—		UNKNOWN
ADNP	Helsmoortel-van der Aa syndrome	OMIM:615873	MENDELIAN
ADNP	Helsmoortel-Van der Aa syndrome	ORPHA:404448	UNKNOWN
ADORA1	—		UNKNOWN
ADORA2A	Acute encephalopathy with biphasic seizures and late reduced diffusion	ORPHA:363549	UNKNOWN
ADORA3	—		UNKNOWN
ADRA1A	—		UNKNOWN
ADRA2A	Lipodystrophy, familial partial, type 8	OMIM:620679	MENDELIAN
ADRA2B	Familial adult myoclonic epilepsy	ORPHA:86814	UNKNOWN
ADRA2C	—		UNKNOWN
ADRB1	Short sleep, familial natural, 2	OMIM:618591	MENDELIAN

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ADRB2	—		UNKNOWN
ADRB3	Obesity	OMIM:601665	POLYGENIC
ADRBK2	—		UNKNOWN
ADSL	Adenylosuccinase deficiency	OMIM:103050	MENDELIAN
ADSL	Adenylosuccinate lyase deficiency	ORPHA:46	UNKNOWN
ADTRP	—		UNKNOWN
AEBP1	Ehlers-Danlos syndrome, classic-like, 2	OMIM:618000	MENDELIAN
AEBP1	Classical-like Ehlers-Danlos syndrome type 2	ORPHA:536532	UNKNOWN
AFF2	Intellectual developmental disorder, X-linked 109	OMIM:309548	MENDELIAN
AFF2	FRAXE intellectual disability	ORPHA:100973	UNKNOWN
AFF3	KINSSHIP syndrome	OMIM:619297	MENDELIAN
AFF4	CHOPS syndrome	OMIM:616368	MENDELIAN
AFF4	Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome	ORPHA:444077	UNKNOWN
AFG3L2	Spinocerebellar ataxia 28	OMIM:610246	MENDELIAN
AFG3L2	Optic atrophy 12	OMIM:618977	MENDELIAN
AFG3L2	Spastic ataxia 5, autosomal recessive	OMIM:614487	MENDELIAN
AFG3L2	Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome	ORPHA:313772	UNKNOWN
AFG3L2	Spinocerebellar ataxia type 28	ORPHA:101109	UNKNOWN
AFP	Alpha-Fetoprotein deficiency	OMIM:615969	MENDELIAN
AFP	Alpha-Fetoprotein, hereditary persistence of	OMIM:615970	MENDELIAN
AGA	Aspartylglucosaminuria	OMIM:208400	MENDELIAN
AGA	Aspartylglucosaminuria	ORPHA:93	UNKNOWN
AGBL1	Corneal dystrophy, fuchs endothelial, 8	OMIM:615523	MENDELIAN
AGBL1	Fuchs endothelial corneal dystrophy	ORPHA:98974	UNKNOWN
AGBL4	—		UNKNOWN
AGER	—		UNKNOWN
AGGF1	Capillary-lymphatic-venous malformation with segmental distribution	ORPHA:90308	UNKNOWN
AGK	Sengers syndrome	OMIM:212350	MENDELIAN
AGK	Cataract, autosomal recessive congenital 5	OMIM:614691	MENDELIAN
AGK	Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome	ORPHA:1369	UNKNOWN
AGL	Glycogen storage disease III	OMIM:232400	MENDELIAN
AGL	Glycogen storage disease due to glycogen debranching enzyme deficiency	ORPHA:366	UNKNOWN
AGMO	—		UNKNOWN
AGO1	—		UNKNOWN
AGO2	—		UNKNOWN
AGPAT2	Lipodystrophy, congenital generalized, type 1	OMIM:608594	MENDELIAN
AGPS	Rhizomelic chondrodysplasia punctata, type 3	OMIM:600121	MENDELIAN
AGRN	Myasthenic syndrome, congenital, 8	OMIM:615120	MENDELIAN
AGRP	Obesity	OMIM:601665	POLYGENIC
AGT	Renal tubular dysgenesis	OMIM:267430	MENDELIAN
AGTPBP1	Neurodegeneration, childhood-onset, with cerebellar atrophy	OMIM:618276	MENDELIAN
AGTPBP1	Pontocerebellar hypoplasia type 1	ORPHA:2254	UNKNOWN
AGTR1	Renal tubular dysgenesis	OMIM:267430	MENDELIAN
AGTR1	Hypertension, essential	OMIM:145500	POLYGENIC
AGXT	Hyperoxaluria, primary, type I	OMIM:259900	MENDELIAN
AGXT	Primary hyperoxaluria type 1	ORPHA:93598	UNKNOWN
AGXT2	Beta-aminoisobutyric acid, urinary excretion of	OMIM:210100	MENDELIAN
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	OMIM:613752	MENDELIAN
AHCY	S-adenosylhomocysteine hydrolase deficiency	ORPHA:88618	UNKNOWN
AHDC1	Xia-Gibbs syndrome	OMIM:615829	MENDELIAN
AHDC1	AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome	ORPHA:412069	UNKNOWN
AHI1	Joubert syndrome 3	OMIM:608629	MENDELIAN
AHI1	Joubert syndrome with ocular anomaly	ORPHA:220493	UNKNOWN
AHI1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
AHI1	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
AHNAK2	—		UNKNOWN
AHR	Retinitis pigmentosa 85	OMIM:618345	MENDELIAN
AHR	Foveal hypoplasia 3	OMIM:620958	MENDELIAN
AHR	Retinitis pigmentosa	ORPHA:791	UNKNOWN
AHRR	—		UNKNOWN
AHSG	Alopecia-intellectual disability syndrome 1	OMIM:203650	MENDELIAN
AHSG	Alopecia-intellectual disability syndrome	ORPHA:2850	UNKNOWN
AHSP	—		UNKNOWN
AICDA	Immunodeficiency with hyper-igm, type 2	OMIM:605258	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
AIF1	—		UNKNOWN
AIFM1	Cowchock syndrome	OMIM:310490	MENDELIAN
AIFM1	Spondyloepimetaphyseal dysplasia, X-linked, with mental deterioration	OMIM:300232	MENDELIAN
AIFM1	Combined oxidative phosphorylation deficiency 6	OMIM:300816	MENDELIAN
AIFM1	Deafness, X-linked 5	OMIM:300614	MENDELIAN
AIFM1	X-linked Charcot-Marie-Tooth disease type 4	ORPHA:101078	UNKNOWN
AIFM1	Severe X-linked mitochondrial encephalomyopathy	ORPHA:238329	UNKNOWN
AIFM1	Leukoencephalopathy-spondyloepimetaphyseal dysplasia syndrome	ORPHA:83629	UNKNOWN
AIMP1	Leukodystrophy, hypomyelinating, 3	OMIM:260600	MENDELIAN
AIP	Pituitary adenoma, growth hormone-secreting, 1	OMIM:102200	MENDELIAN
AIP	Pituitary adenoma 4, ACTH-secreting, somatic	OMIM:219090	MENDELIAN
AIP	Acromegaly	ORPHA:963	UNKNOWN
AIP	Prolactinoma	ORPHA:2965	UNKNOWN
AIP	Pituitary gigantism	ORPHA:99725	UNKNOWN
AIPL1	Leber congenital amaurosis 4	OMIM:604393	MENDELIAN
AIPL1	Retinitis pigmentosa	OMIM:268000	MENDELIAN
AIPL1	Leber congenital amaurosis	ORPHA:65	UNKNOWN
AIPL1	Cone rod dystrophy	ORPHA:1872	UNKNOWN
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	OMIM:240300	MENDELIAN
AIRE	Autoimmune polyendocrinopathy type 1	ORPHA:3453	UNKNOWN
AK1	Adenylate kinase deficiency, hemolytic anemia due to	OMIM:612631	MENDELIAN
AK2	Reticular dysgenesis	OMIM:267500	MENDELIAN
AK2	Reticular dysgenesis	ORPHA:33355	UNKNOWN
AK7	Spermatogenic failure 27	OMIM:617965	MENDELIAN
AK8	—		UNKNOWN
AKAP1	—		UNKNOWN
AKAP10	—		UNKNOWN
AKAP13	—		UNKNOWN
AKAP2	—		UNKNOWN
AKAP9	Long QT syndrome 11	OMIM:611820	MENDELIAN
AKAP9	Brugada syndrome	ORPHA:130	UNKNOWN
AKR1B1	—		UNKNOWN
AKR1C2	46,xy sex reversal 8	OMIM:614279	MENDELIAN
AKR1C3	—		UNKNOWN
AKR1C4	46,xy sex reversal 8	OMIM:614279	POLYGENIC
AKR1D1	Bile acid synthesis defect, congenital, 2	OMIM:235555	MENDELIAN
AKR1D1	Congenital bile acid synthesis defect type 2	ORPHA:79303	UNKNOWN
AKR7A2	—		UNKNOWN
AKR7A3	—		UNKNOWN
AKT1	Cowden syndrome 6	OMIM:615109	MENDELIAN
AKT1	Breast cancer	OMIM:114480	MENDELIAN
AKT1	Proteus syndrome, somatic	OMIM:176920	MENDELIAN
AKT1	Colorectal cancer	OMIM:114500	MENDELIAN
AKT1	Ovarian cancer	OMIM:167000	MENDELIAN
AKT1	Proteus syndrome	ORPHA:744	UNKNOWN
AKT1	Meningioma	ORPHA:2495	UNKNOWN
AKT2	Hypoinsulinemic hypoglycemia with hemihypertrophy	OMIM:240900	MENDELIAN
AKT2	Diabetes mellitus, noninsulin-dependent	OMIM:125853	MENDELIAN
AKT2	Hypoinsulinemic hypoglycemia and body hemihypertrophy	ORPHA:293964	UNKNOWN
AKT2	AKT2-related familial partial lipodystrophy	ORPHA:79085	UNKNOWN
AKT3	Megalencephaly-Polymicrogyria-Polydactyly-Hydrocephalus syndrome 2	OMIM:615937	MENDELIAN
AKT3	Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome	ORPHA:83473	UNKNOWN
AKT3	Hemimegalencephaly	ORPHA:99802	UNKNOWN
ALAD	Porphyria, acute hepatic	OMIM:612740	MENDELIAN
ALAD	Porphyria due to ALA dehydratase deficiency	ORPHA:100924	UNKNOWN
ALAS2	Anemia, sideroblastic, X-linked	OMIM:300751	MENDELIAN
ALAS2	Protoporphyria, erythropoietic, X-linked	OMIM:300752	MENDELIAN
ALAS2	X-linked sideroblastic anemia	ORPHA:75563	UNKNOWN
ALB	Familial dysalbuminemic hyperthyroxinemia	OMIM:615999	MENDELIAN
ALB	Analbuminemia	OMIM:616000	MENDELIAN
ALB	Congenital analbuminemia	ORPHA:86816	UNKNOWN
ALCAM	—		UNKNOWN
ALDH16A1	—		UNKNOWN
ALDH18A1	Spastic paraplegia 9B, autosomal recessive	OMIM:616586	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
ALDH18A1	Cutis laxa, autosomal dominant 3	OMIM:616603	MENDELIAN
ALDH18A1	Spastic paraplegia 9A, autosomal dominant	OMIM:601162	MENDELIAN
ALDH18A1	Cutis laxa, autosomal recessive, type IIIA	OMIM:219150	MENDELIAN
ALDH18A1	Autosomal dominant cutis laxa	ORPHA:90348	UNKNOWN
ALDH18A1	ALDH18A1-related De Barsy syndrome	ORPHA:35664	UNKNOWN
ALDH18A1	Autosomal dominant spastic paraplegia type 9B	ORPHA:447757	UNKNOWN
ALDH18A1	Autosomal dominant spastic paraplegia type 9A	ORPHA:447753	UNKNOWN
ALDH18A1	Autosomal recessive spastic paraplegia type 9B	ORPHA:447760	UNKNOWN
ALDH1A1	—		UNKNOWN
ALDH1A2	Diaphragmatic hernia 4, with cardiovascular defects	OMIM:620025	MENDELIAN
ALDH1A3	Microphthalmia, isolated 8	OMIM:615113	MENDELIAN
ALDH1A3	Isolated complete colobomatous microphthalmia	ORPHA:35612	UNKNOWN
ALDH1B1	—		UNKNOWN
ALDH2	Alcohol sensitivity, acute	OMIM:610251	MENDELIAN
ALDH3A2	Sjogren-Larsson syndrome	OMIM:270200	MENDELIAN
ALDH3A2	Sjögren-Larsson syndrome	ORPHA:816	UNKNOWN
ALDH4A1	Hyperprolinemia, type II	OMIM:239510	MENDELIAN
ALDH4A1	Hyperprolinemia type 2	ORPHA:79101	UNKNOWN
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency	OMIM:271980	MENDELIAN
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency	ORPHA:22	UNKNOWN
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency	OMIM:614105	MENDELIAN
ALDH7A1	Epilepsy, pyridoxine-dependent	OMIM:266100	MENDELIAN
ALDH7A1	Pyridoxine-dependent-developmental and epileptic encephalopathy	ORPHA:3006	UNKNOWN
ALDOA	Glycogen storage disease XII	OMIM:611881	MENDELIAN
ALDOA	Glycogen storage disease due to aldolase A deficiency	ORPHA:57	UNKNOWN
ALDOB	Fructose intolerance, hereditary	OMIM:229600	MENDELIAN
ALDOB	Hereditary fructose intolerance	ORPHA:469	UNKNOWN
ALG1	Congenital disorder of glycosylation, type Ik	OMIM:608540	MENDELIAN
ALG1	ALG1-CDG	ORPHA:79327	UNKNOWN
ALG10	—		UNKNOWN
ALG10B	Long QT syndrome 2	OMIM:613688	POLYGENIC
ALG11	Congenital disorder of glycosylation, type Ip	OMIM:613661	MENDELIAN
ALG11	ALG11-CDG	ORPHA:280071	UNKNOWN
ALG12	Congenital disorder of glycosylation, type Ig	OMIM:607143	MENDELIAN
ALG12	ALG12-CDG	ORPHA:79324	UNKNOWN
ALG13	Epileptic encephalopathy, early infantile, 36	OMIM:300884	MENDELIAN
ALG13	ALG13-CDG	ORPHA:324422	UNKNOWN
ALG14	Myasthenic syndrome, congenital, 15	OMIM:616227	MENDELIAN
ALG14	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies	OMIM:619031	MENDELIAN
ALG14	Myopathy, epilepsy, and progressive cerebral atrophy	OMIM:619036	MENDELIAN
ALG14	Congenital myasthenic syndrome with glycosylation defect	ORPHA:353327	UNKNOWN
ALG2	Myasthenic syndrome, congenital, 14	OMIM:616228	MENDELIAN
ALG2	Congenital disorder of glycosylation, type Ii	OMIM:607906	MENDELIAN
ALG2	Congenital myasthenic syndrome with glycosylation defect	ORPHA:353327	UNKNOWN
ALG2	ALG2-CDG	ORPHA:79326	UNKNOWN
ALG3	Congenital disorder of glycosylation, type Id	OMIM:601110	MENDELIAN
ALG3	ALG3-CDG	ORPHA:79321	UNKNOWN
ALG5	Polycystic kidney disease 7	OMIM:620056	MENDELIAN
ALG5	Autosomal dominant polycystic kidney disease	ORPHA:730	UNKNOWN
ALG6	Congenital disorder of glycosylation, type Ic	OMIM:603147	MENDELIAN
ALG6	ALG6-CDG	ORPHA:79320	UNKNOWN
ALG8	Congenital disorder of glycosylation, type Ih	OMIM:608104	MENDELIAN
ALG8	Polycystic liver disease 3 with or without kidney cysts	OMIM:617874	MENDELIAN
ALG8	Isolated polycystic liver disease	ORPHA:2924	UNKNOWN
ALG8	ALG8-CDG	ORPHA:79325	UNKNOWN
ALG9	Congenital disorder of glycosylation, type II	OMIM:608776	MENDELIAN
ALG9	Gillessen-Kaesbach-Nishimura syndrome	OMIM:263210	MENDELIAN
ALG9	Autosomal dominant polycystic kidney disease	ORPHA:730	UNKNOWN
ALG9	ALG9-CDG	ORPHA:79328	UNKNOWN
ALK	Ganglioneuroma	ORPHA:251992	UNKNOWN
ALK	Large/giant congenital melanocytic nevus	ORPHA:626	UNKNOWN
ALK	Neuroblastoma	ORPHA:635	UNKNOWN
ALKBH2	—		UNKNOWN
ALKBH3	—		UNKNOWN
ALMS1	Alstrom syndrome	OMIM:203800	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
ALMS1	Alström syndrome	ORPHA:64	UNKNOWN
ALMS1	Leber congenital amaurosis	ORPHA:65	UNKNOWN
ALMS1	Severe early-childhood-onset retinal dystrophy	ORPHA:364055	UNKNOWN
ALOX12	—		UNKNOWN
ALOX12B	Ichthyosis, congenital, autosomal recessive 2	OMIM:242100	MENDELIAN
ALOX12B	Lamellar ichthyosis	ORPHA:313	UNKNOWN
ALOX12B	Congenital ichthyosiform erythroderma	ORPHA:79394	UNKNOWN
ALOX12B	Self-improving collodion baby	ORPHA:281122	UNKNOWN
ALOX15	—		UNKNOWN
ALOX5	Asthma, susceptibility to	OMIM:600807	POLYGENIC
ALOX5AP	Ischemic stroke, susceptibility to	OMIM:601367	POLYGENIC
ALOXE3	Ichthyosis, congenital, autosomal recessive 2	OMIM:242100	MENDELIAN
ALOXE3	Ichthyosis, congenital, autosomal recessive 3	OMIM:606545	MENDELIAN
ALOXE3	Lamellar ichthyosis	ORPHA:313	UNKNOWN
ALOXE3	Congenital ichthyosiform erythroderma	ORPHA:79394	UNKNOWN
ALOXE3	Self-improving collodion baby	ORPHA:281122	UNKNOWN
ALPL	Hypophosphatasia, adult	OMIM:146300	MENDELIAN
ALPL	Hypophosphatasia, infantile	OMIM:241500	MENDELIAN
ALPL	Hypophosphatasia, childhood	OMIM:241510	MENDELIAN
ALPP	—		UNKNOWN
ALS2	Amyotrophic lateral sclerosis 2, juvenile	OMIM:205100	MENDELIAN
ALS2	Spastic paralysis, infantile-onset ascending	OMIM:607225	MENDELIAN
ALS2	Primary lateral sclerosis, juvenile	OMIM:606353	MENDELIAN
ALS2	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
ALS2	Infantile-onset ascending hereditary spastic paralysis	ORPHA:293168	UNKNOWN
ALS2	Juvenile primary lateral sclerosis	ORPHA:247604	UNKNOWN
ALS2CL	—		UNKNOWN
ALX1	Frontonasal dysplasia 3	OMIM:613456	MENDELIAN
ALX1	Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome	ORPHA:306542	UNKNOWN
ALX3	Frontonasal dysplasia 1	OMIM:136760	MENDELIAN
ALX3	Frontorhiny	ORPHA:391474	UNKNOWN
ALX4	Craniosynostosis 5, susceptibility to	OMIM:615529	POLYGENIC
ALX4	Parietal foramina 2	OMIM:609597	MENDELIAN
ALX4	Frontonasal dysplasia 2	OMIM:613451	MENDELIAN
ALX4	Frontonasal dysplasia-alopecia-genital anomalies syndrome	ORPHA:228390	UNKNOWN
ALX4	Enlarged parietal foramina	ORPHA:60015	UNKNOWN
ALX4	Non-syndromic sagittal craniosynostosis	ORPHA:35093	UNKNOWN
ALX4	Potocki-Shaffer syndrome	ORPHA:52022	UNKNOWN
AMACR	Alpha-methylacyl-CoA racemase deficiency	OMIM:614307	MENDELIAN
AMACR	Bile acid synthesis defect, congenital, 4	OMIM:214950	MENDELIAN
AMACR	Congenital bile acid synthesis defect type 4	ORPHA:79095	UNKNOWN
AMBN	Amelogenesis imperfecta, type IF	OMIM:616270	MENDELIAN
AMD1	—		UNKNOWN
AMELX	Amelogenesis imperfecta, type IE	OMIM:301200	MENDELIAN
AMELY	—		UNKNOWN
AMER1	Osteopathia striata with cranial sclerosis	OMIM:300373	MENDELIAN
AMER1	Osteopathia striata-cranial sclerosis syndrome	ORPHA:2780	UNKNOWN
AMH	Persistent mullerian duct syndrome, types I and II	OMIM:261550	MENDELIAN
AMH	Persistent Müllerian duct syndrome	ORPHA:2856	UNKNOWN
AMHR2	Persistent mullerian duct syndrome, types I and II	OMIM:261550	MENDELIAN
AMHR2	Persistent Müllerian duct syndrome	ORPHA:2856	UNKNOWN
AMN	Imerslund-Grasbeck syndrome 2	OMIM:618882	MENDELIAN
AMN	Imerslund-Grasbeck syndrome	ORPHA:35858	UNKNOWN
AMPD1	Myopathy due to myoadenylate deaminase deficiency	OMIM:615511	MENDELIAN
AMPD1	Adenosine monophosphate deaminase deficiency	ORPHA:45	UNKNOWN
AMPD2	Spastic paraplegia 63, autosomal recessive	OMIM:615686	MENDELIAN
AMPD2	Pontocerebellar hypoplasia, type 9	OMIM:615809	MENDELIAN
AMPD2	Autosomal recessive spastic paraplegia type 63	ORPHA:401805	UNKNOWN
AMPD3	AMP deaminase deficiency, erythrocytic	OMIM:612874	MENDELIAN
AMPD3	Adenosine monophosphate deaminase deficiency	ORPHA:45	UNKNOWN
AMT	Glycine encephalopathy 2	OMIM:620398	MENDELIAN
ANAPC1	Rothmund-thomson syndrome, type 1	OMIM:618625	MENDELIAN
ANAPC1	Rothmund-Thomson syndrome type 1	ORPHA:221008	UNKNOWN
ANG	Amyotrophic lateral sclerosis 9	OMIM:611895	MENDELIAN
ANG	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
ANGPT1	Angioedema, hereditary, 5	OMIM:619361	MENDELIAN
ANGPT2	Lymphatic malformation 10	OMIM:619369	MENDELIAN
ANGPTL3	Hypobetalipoproteinemia, familial, 2	OMIM:605019	MENDELIAN
ANGPTL5	—		UNKNOWN
ANGPTL6	Familial cerebral saccular aneurysm	ORPHA:231160	UNKNOWN
ANK1	Spherocytosis, type 1	OMIM:182900	MENDELIAN
ANK1	Hereditary spherocytosis	ORPHA:822	UNKNOWN
ANK1	8p11.2 deletion syndrome	ORPHA:251066	UNKNOWN
ANK2	Cardiac arrhythmia, ankyrin-B-related	OMIM:600919	MENDELIAN
ANK3	Intellectual developmental disorder, autosomal recessive 37	OMIM:615493	MENDELIAN
ANK3	ANK3-related intellectual disability-sleep disturbance syndrome	ORPHA:356996	UNKNOWN
ANKH	Chondrocalcinosis 2	OMIM:118600	MENDELIAN
ANKH	Cranio metaphyseal dysplasia	OMIM:123000	MENDELIAN
ANKH	Familial calcium pyrophosphate deposition	ORPHA:1416	UNKNOWN
ANKH	Cranio metaphyseal dysplasia	ORPHA:1522	UNKNOWN
ANKK1	—		UNKNOWN
ANKLE2	Microcephaly 16, primary, autosomal recessive	OMIM:616681	MENDELIAN
ANKLE2	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
ANKRD1	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
ANKRD11	KBG syndrome	OMIM:148050	MENDELIAN
ANKRD11	KBG syndrome	ORPHA:2332	UNKNOWN
ANKRD11	16q24.3 microdeletion syndrome	ORPHA:261250	UNKNOWN
ANKRD17	Chopra-Amiel-Gordon syndrome	OMIM:619504	MENDELIAN
ANKRD26	Thrombocytopenia 2	OMIM:188000	MENDELIAN
ANKRD28	—		UNKNOWN
ANKRD44	—		UNKNOWN
ANKRD52	—		UNKNOWN
ANKS1A	—		UNKNOWN
ANKS1B	—		UNKNOWN
ANKS6	Nephronophthisis 16	OMIM:615382	MENDELIAN
ANLN	Focal segmental glomerulosclerosis 8	OMIM:616032	MENDELIAN
ANLN	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
ANO10	Spinocerebellar ataxia, autosomal recessive 10	OMIM:613728	MENDELIAN
ANO10	Adult-onset autosomal recessive cerebellar ataxia	ORPHA:284289	UNKNOWN
ANO3	Dystonia 24	OMIM:615034	MENDELIAN
ANO3	Cranio-cervical dystonia with laryngeal and upper-limb involvement	ORPHA:420485	UNKNOWN
ANO5	Miyoshi muscular dystrophy 3	OMIM:613319	MENDELIAN
ANO5	Muscular dystrophy, limb-girdle, type 2L	OMIM:611307	MENDELIAN
ANO5	Gnathodiaphyseal dysplasia	OMIM:166260	MENDELIAN
ANO5	Gnathodiaphyseal dysplasia	ORPHA:53697	UNKNOWN
ANO5	Distal anoctaminopathy	ORPHA:399096	UNKNOWN
ANO5	Isolated asymptomatic elevation of creatine phosphokinase	ORPHA:206599	UNKNOWN
ANO5	Anoctamin-5-related limb-girdle muscular dystrophy R12	ORPHA:206549	UNKNOWN
ANO6	Scott syndrome	OMIM:262890	MENDELIAN
ANO6	Scott syndrome	ORPHA:806	UNKNOWN
ANO7	—		UNKNOWN
ANP32A	—		UNKNOWN
ANTXR1	Hemangioma, capillary infantile	OMIM:602089	POLYGENIC
ANTXR1	GAPO syndrome	OMIM:230740	MENDELIAN
ANTXR1	GAPO syndrome	ORPHA:2067	UNKNOWN
ANTXR2	Hyaline fibromatosis syndrome	OMIM:228600	MENDELIAN
ANTXR2	Juvenile hyaline fibromatosis	ORPHA:2028	UNKNOWN
ANTXR2	Infantile systemic hyalinosis	ORPHA:2176	UNKNOWN
ANXA1	—		UNKNOWN
ANXA11	Amyotrophic lateral sclerosis 23	OMIM:617839	MENDELIAN
ANXA11	Inclusion body myopathy and brain white matter abnormalities	OMIM:619733	MENDELIAN
ANXA11	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
ANXA5	Pregnancy loss, recurrent, susceptibility to, 3	OMIM:614391	POLYGENIC
AOAH	—		UNKNOWN
AOC1	—		UNKNOWN
AP1S1	MEDNIK syndrome	OMIM:609313	MENDELIAN
AP1S1	MEDNIK syndrome	ORPHA:171851	UNKNOWN
AP1S2	Pettigrew syndrome	OMIM:304340	MENDELIAN
AP1S2	Fried syndrome	ORPHA:85335	UNKNOWN
AP1S2	X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome	ORPHA:85329	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
AP1S2	X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome	ORPHA:1568	UNKNOWN
AP1S3	Psoriasis 15, pustular, susceptibility to	OMIM:616106	POLYGENIC
AP1S3	Generalized pustular psoriasis	ORPHA:247353	UNKNOWN
AP2S1	Hypocalciuric hypercalcemia, familial, type III	OMIM:600740	MENDELIAN
AP3B1	Hermansky-Pudlak syndrome 2	OMIM:608233	MENDELIAN
AP3B2	Developmental and epileptic encephalopathy 48	OMIM:617276	MENDELIAN
AP3B2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
AP3D1	Hermansky-Pudlak syndrome 10	OMIM:617050	MENDELIAN
AP3D1	X-linked recessive ocular albinism	ORPHA:54	UNKNOWN
AP3D1	Ocular albinism with late-onset sensorineural deafness	ORPHA:1000	UNKNOWN
AP4B1	Spastic paraplegia 47, autosomal recessive	OMIM:614066	MENDELIAN
AP4B1	Severe intellectual disability and progressive spastic paraplegia	ORPHA:280763	UNKNOWN
AP4E1	Stuttering, familial persistent, 1	OMIM:184450	MENDELIAN
AP4E1	Spastic paraplegia 51, autosomal recessive	OMIM:613744	MENDELIAN
AP4E1	Severe intellectual disability and progressive spastic paraplegia	ORPHA:280763	UNKNOWN
AP4M1	Spastic paraplegia 50, autosomal recessive	OMIM:612936	MENDELIAN
AP4M1	Severe intellectual disability and progressive spastic paraplegia	ORPHA:280763	UNKNOWN
AP4S1	Spastic paraplegia 52, autosomal recessive	OMIM:614067	MENDELIAN
AP4S1	Severe intellectual disability and progressive spastic paraplegia	ORPHA:280763	UNKNOWN
AP5Z1	Spastic paraplegia 48, autosomal recessive	OMIM:613647	MENDELIAN
AP5Z1	Autosomal recessive spastic paraplegia type 48	ORPHA:306511	UNKNOWN
APAF1	—		UNKNOWN
APBA2	—		UNKNOWN
APBB1	—		UNKNOWN
APBB1IP	—		UNKNOWN
APBB2	—		UNKNOWN
APBB3	—		UNKNOWN
APC	Desmoid disease, hereditary	OMIM:135290	MENDELIAN
APC	Gastric adenocarcinoma and proximal polyposis of the stomach	OMIM:619182	MENDELIAN
APC	Adenomatous polyposis coli	OMIM:175100	MENDELIAN
APC	Colorectal cancer	OMIM:114500	MENDELIAN
APC	Hepatocellular carcinoma	OMIM:114550	MENDELIAN
APC	Gastric cancer, somatic	OMIM:613659	MENDELIAN
APC	Attenuated familial adenomatous polyposis	ORPHA:220460	UNKNOWN
APC	Cenani-Lenz syndrome	ORPHA:3258	UNKNOWN
APC	5q22 microdeletion syndrome	ORPHA:261584	UNKNOWN
APC	Desmoid tumor	ORPHA:873	UNKNOWN
APC	Familial adenomatous polyposis	ORPHA:733	UNKNOWN
APC2	Cortical dysplasia, complex, with other brain malformations 10	OMIM:618677	MENDELIAN
APC2	Intellectual developmental disorder, autosomal recessive 74	OMIM:617169	MENDELIAN
APC2	Sotos syndrome	ORPHA:821	UNKNOWN
APCDD1	Hypotrichosis 1	OMIM:605389	MENDELIAN
APCDD1	Hypotrichosis simplex	ORPHA:55654	UNKNOWN
APCS	—		UNKNOWN
APEX1	—		UNKNOWN
APEX2	—		UNKNOWN
APH1A	—		UNKNOWN
APH1B	—		UNKNOWN
APITD1	—		UNKNOWN
APITD1-CORT	—		UNKNOWN
APLF	—		UNKNOWN
APLNR	—		UNKNOWN
APOA1	Amyloidosis, hereditary systemic 3	OMIM:620657	MENDELIAN
APOA1	Hypoalphalipoproteinemia, primary, 2, intermediate	OMIM:619836	MENDELIAN
APOA1	Hypoalphalipoproteinemia, primary, 2	OMIM:618463	MENDELIAN
APOA1	Apolipoprotein A-I deficiency	ORPHA:425	UNKNOWN
APOA2	Hypercholesterolemia, familial, 1	OMIM:143890	POLYGENIC
APOA2	Apolipoprotein A-II deficiency]	OMIM:621417	MENDELIAN
APOA4	Tubulointerstitial kidney disease, autosomal dominant 6	OMIM:621106	MENDELIAN
APOA5	Hypertriglyceridemia, familial	OMIM:145750	POLYGENIC
APOA5	Hyperlipoproteinemia, type V	OMIM:144650	MENDELIAN
APOB	Hypobetalipoproteinemia, familial, 1	OMIM:615558	MENDELIAN
APOB	Hypercholesterolemia, familial, 2	OMIM:144010	MENDELIAN
APOB	Homozygous familial hypercholesterolemia	ORPHA:391665	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
APOBEC1	—		UNKNOWN
APOBEC3B	—		UNKNOWN
APOBEC3G	—		UNKNOWN
APOBEC3H	—		UNKNOWN
APOC1	—		UNKNOWN
APOC2	Apolipoprotein C-II deficiency	OMIM:207750	MENDELIAN
APOC3	Hyperalphalipoproteinemia 2	OMIM:614028	MENDELIAN
APOC4	—		UNKNOWN
APOC4-APOC2	—		UNKNOWN
APOD	—		UNKNOWN
APOE	Lipoprotein glomerulopathy	OMIM:611771	MENDELIAN
APOE	Sea-Blue histiocyte disease	OMIM:269600	MENDELIAN
APOE	Macular degeneration, age-related, 1	OMIM:603075	POLYGENIC
APOE	Alzheimer disease-4	OMIM:606889	MENDELIAN
APOE	Alzheimer disease 2	OMIM:104310	MENDELIAN
APOE	Hyperlipoproteinemia, type III	OMIM:617347	MENDELIAN
APOE	Alzheimer disease 3	OMIM:607822	POLYGENIC
APOE	Dysbetalipoproteinemia	ORPHA:412	UNKNOWN
APOH	—		UNKNOWN
APOL1	Focal segmental glomerulosclerosis 4, susceptibility to	OMIM:612551	POLYGENIC
APOL1	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
APOL2	Schizophrenia	OMIM:181500	POLYGENIC
APOL3	—		UNKNOWN
APOL4	—		UNKNOWN
APOL6	—		UNKNOWN
APOM	—		UNKNOWN
APOPT1	—		UNKNOWN
APP	Cerebral amyloid angiopathy, APP-related	OMIM:605714	MENDELIAN
APP	Alzheimer disease	OMIM:104300	MENDELIAN
APP	Early-onset autosomal dominant Alzheimer disease	ORPHA:1020	UNKNOWN
APP	ABeta amyloidosis, Arctic type	ORPHA:324723	UNKNOWN
APP	ABeta amyloidosis, Italian type	ORPHA:324713	UNKNOWN
APP	ABetaL34V amyloidosis	ORPHA:324703	UNKNOWN
APP	ABeta amyloidosis, Iowa type	ORPHA:324708	UNKNOWN
APP	ABeta amyloidosis, Dutch type	ORPHA:100006	UNKNOWN
APRT	Adenine phosphoribosyltransferase deficiency	OMIM:614723	MENDELIAN
APRT	Adenine phosphoribosyltransferase deficiency	ORPHA:976	UNKNOWN
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	OMIM:208920	MENDELIAN
APTX	Coenzyme Q10 deficiency, primary, 1	OMIM:607426	MENDELIAN
APTX	Ataxia-oculomotor apraxia type 1	ORPHA:1168	UNKNOWN
AQP2	Diabetes insipidus, nephrogenic, 2	OMIM:125800	MENDELIAN
AQP2	Arginine vasopressin resistance	ORPHA:223	UNKNOWN
AQP4	Megalencephalic leukoencephalopathy with subcortical cysts 4, remitting	OMIM:620448	MENDELIAN
AQP4	Megalencephalic leukoencephalopathy with subcortical cysts	ORPHA:2478	UNKNOWN
AQP5	Palmoplantar keratoderma, Bothnian type	OMIM:600231	MENDELIAN
AQP5	Diffuse palmoplantar keratoderma, Bothnian type	ORPHA:2337	UNKNOWN
AR	Spinal and bulbar muscular atrophy, X-linked 1	OMIM:313200	MENDELIAN
AR	Androgen insensitivity syndrome	OMIM:300068	MENDELIAN
AR	Prostate cancer, susceptibility to	OMIM:301120	POLYGENIC
AR	Reifenstein syndrome	OMIM:312300	MENDELIAN
AR	Hypospadias 1, X-linked	OMIM:300633	MENDELIAN
AR	Partial androgen insensitivity syndrome	ORPHA:90797	UNKNOWN
AR	Non-syndromic posterior hypospadias	ORPHA:95706	UNKNOWN
AR	Complete androgen insensitivity syndrome	ORPHA:99429	UNKNOWN
AR	Kennedy disease	ORPHA:481	UNKNOWN
ARAF	—		UNKNOWN
AREG	—		UNKNOWN
AREL1	—		UNKNOWN
ARF4	—		UNKNOWN
ARFGEF2	Periventricular heterotopia with microcephaly, autosomal recessive	OMIM:608097	MENDELIAN
ARFGEF2	Periventricular nodular heterotopia	ORPHA:98892	UNKNOWN
ARFRP1	—		UNKNOWN
ARG1	Argininemia	OMIM:207800	MENDELIAN
ARG1	Argininemia	ORPHA:90	UNKNOWN
ARHGAP18	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
ARHGAP24	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
ARHGAP26	Juvenile myelomonocytic leukemia	OMIM:607785	MENDELIAN
ARHGAP29	Cleft lip/palate	ORPHA:199306	UNKNOWN
ARHGAP31	Adams-Oliver syndrome 1	OMIM:100300	MENDELIAN
ARHGAP31	Adams-Oliver syndrome	ORPHA:974	UNKNOWN
ARHGAP6	—		UNKNOWN
ARHGAP9	—		UNKNOWN
ARHGDIA	Nephrotic syndrome, type 8	OMIM:615244	MENDELIAN
ARHGDIA	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
ARHGEF10	Slowed nerve conduction velocity, autosomal dominant	OMIM:608236	MENDELIAN
ARHGEF11	—		UNKNOWN
ARHGEF12	—		UNKNOWN
ARHGEF4	—		UNKNOWN
ARHGEF7	—		UNKNOWN
ARHGEF9	Developmental and epileptic encephalopathy 8	OMIM:300607	MENDELIAN
ARHGEF9	Hyperekplexia-epilepsy syndrome	ORPHA:163985	UNKNOWN
ARID1A	Coffin-Siris syndrome 2	OMIM:614607	MENDELIAN
ARID1A	Coffin-Siris syndrome	ORPHA:1465	UNKNOWN
ARID1B	Coffin-Siris syndrome 1	OMIM:135900	MENDELIAN
ARID1B	Coffin-Siris syndrome	ORPHA:1465	UNKNOWN
ARID1B	6q25.2q25.3 microdeletion syndrome	ORPHA:251056	UNKNOWN
ARID2	Coffin-Siris syndrome 6	OMIM:617808	MENDELIAN
ARID2	Coffin-Siris syndrome	ORPHA:1465	UNKNOWN
ARID4A	—		UNKNOWN
ARID4B	—		UNKNOWN
ARID5A	—		UNKNOWN
ARID5B	—		UNKNOWN
ARIH1	—		UNKNOWN
ARL13B	Joubert syndrome 8	OMIM:612291	MENDELIAN
ARL13B	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
ARL14EP	—		UNKNOWN
ARL2BP	Retinitis pigmentosa with or without situs inversus	OMIM:615434	MENDELIAN
ARL2BP	Retinitis pigmentosa	ORPHA:791	UNKNOWN
ARL3	Joubert syndrome 35	OMIM:618161	MENDELIAN
ARL3	Retinitis pigmentosa 83	OMIM:618173	MENDELIAN
ARL3	Retinitis pigmentosa	ORPHA:791	UNKNOWN
ARL3	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
ARL6	Bardet-Biedl syndrome 3	OMIM:600151	MENDELIAN
ARL6	Retinitis pigmentosa	OMIM:268000	MENDELIAN
ARL6	Bardet-Biedl syndrome 1	OMIM:209900	POLYGENIC
ARL6	Retinitis pigmentosa 55	OMIM:613575	MENDELIAN
ARL6	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
ARL6	Retinitis pigmentosa	ORPHA:791	UNKNOWN
ARL6IP1	Spastic paraplegia 61, autosomal recessive	OMIM:615685	MENDELIAN
ARL6IP1	Autosomal recessive spastic paraplegia type 61	ORPHA:401780	UNKNOWN
ARL6IP5	—		UNKNOWN
ARL6IP6	Cutis marmorata telangiectatica congenita	ORPHA:1556	UNKNOWN
ARMC4	—		UNKNOWN
ARMC5	ACTH-independent macronodular adrenal hyperplasia 2	OMIM:615954	POLYGENIC
ARMC5	Cushing syndrome due to bilateral macronodular adrenocortical disease	ORPHA:189427	UNKNOWN
ARNT	—		UNKNOWN
ARNT2	Webb-Dattani syndrome	OMIM:615926	MENDELIAN
ARNT2	Septo-optic dysplasia spectrum	ORPHA:3157	UNKNOWN
ARNTL	—		UNKNOWN
ARPC3	—		UNKNOWN
ARSA	Metachromatic leukodystrophy	OMIM:250100	MENDELIAN
ARSA	Metachromatic leukodystrophy, adult form	ORPHA:309271	UNKNOWN
ARSA	Metachromatic leukodystrophy, juvenile form	ORPHA:309263	UNKNOWN
ARSA	Metachromatic leukodystrophy, late infantile form	ORPHA:309256	UNKNOWN
ARSB	Mucopolysaccharidosis, type VI	OMIM:253200	MENDELIAN
ARSE	—		UNKNOWN
ARSF	—		UNKNOWN
ARSH	—		UNKNOWN
ARSI	Autosomal recessive spastic paraplegia type 66	ORPHA:401815	UNKNOWN
ARV1	Epileptic encephalopathy, early infantile, 38	OMIM:617020	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
ARVCF	22q11.2 deletion syndrome	ORPHA:567	UNKNOWN
ARX	Partington syndrome	OMIM:309510	MENDELIAN
ARX	Developmental and epileptic encephalopathy 1	OMIM:308350	MENDELIAN
ARX	Corpus callosum, agenesis of, with abnormal genitalia	OMIM:300004	MENDELIAN
ARX	Intellectual developmental disorder, X-linked 29	OMIM:300419	MENDELIAN
ARX	Lissencephaly, X-linked, 2	OMIM:300215	MENDELIAN
ARX	X-linked spasticity-intellectual disability-epilepsy syndrome	ORPHA:3175	UNKNOWN
ARX	Corpus callosum agenesis-abnormal genitalia syndrome	ORPHA:2508	UNKNOWN
ARX	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
ARX	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
ARX	Partington syndrome	ORPHA:94083	UNKNOWN
ARX	X-linked lissencephaly with abnormal genitalia	ORPHA:452	UNKNOWN
AS3MT	—		UNKNOWN
ASAH1	Spinal muscular atrophy with progressive myoclonic epilepsy	OMIM:159950	MENDELIAN
ASAH1	Farber lipogranulomatosis	OMIM:228000	MENDELIAN
ASAH1	Farber disease	ORPHA:333	UNKNOWN
ASAH1	Spinal muscular atrophy-progressive myoclonic epilepsy syndrome	ORPHA:2590	UNKNOWN
ASAH2	—		UNKNOWN
ASB1	—		UNKNOWN
ASB10	Glaucoma 1, open angle, F	OMIM:603383	MENDELIAN
ASCC1	Barrett esophagus	OMIM:614266	MENDELIAN
ASCC1	Spinal muscular atrophy with congenital bone fractures 2	OMIM:616867	MENDELIAN
ASCC3	Intellectual developmental disorder, autosomal recessive 81	OMIM:620700	MENDELIAN
ASCL1	Haddad syndrome	ORPHA:99803	UNKNOWN
ASH1L	Intellectual developmental disorder, autosomal dominant 52	OMIM:617796	MENDELIAN
ASIC3	—		UNKNOWN
ASL	Argininosuccinic aciduria	OMIM:207900	MENDELIAN
ASL	Argininosuccinic aciduria	ORPHA:23	UNKNOWN
ASMT	—		UNKNOWN
ASMTL	—		UNKNOWN
ASNS	Asparagine synthetase deficiency	OMIM:615574	MENDELIAN
ASPA	Canavan disease	OMIM:271900	MENDELIAN
ASPA	Mild Canavan disease	ORPHA:314918	UNKNOWN
ASPA	Severe Canavan disease	ORPHA:314911	UNKNOWN
ASPH	Facial dysmorphism, lens dislocation, anterior segment abnormalities, and spontaneous filtering blebs	OMIM:601552	MENDELIAN
ASPM	Microcephaly 5, primary, autosomal recessive	OMIM:608716	MENDELIAN
ASPM	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
ASPN	Hand osteoarthritis	OMIM:607850	POLYGENIC
ASPRV1	Ichthyosis, lamellar, autosomal dominant	OMIM:146750	MENDELIAN
ASPRV1	Lamellar ichthyosis	ORPHA:313	UNKNOWN
ASPSCR1	Alveolar soft part sarcoma	OMIM:606243	MENDELIAN
ASS1	Citrullinemia, classic	OMIM:215700	MENDELIAN
ASTN2	—		UNKNOWN
ASXL1	Myelodysplastic syndrome	OMIM:614286	MENDELIAN
ASXL1	Bohring-Opitz syndrome	OMIM:605039	MENDELIAN
ASXL1	Bohring-Opitz syndrome	ORPHA:97297	UNKNOWN
ASXL1	Aggressive systemic mastocytosis	ORPHA:98850	UNKNOWN
ASXL1	Systemic mastocytosis with associated hematologic neoplasm	ORPHA:98849	UNKNOWN
ASXL2	Shashi-Pena syndrome	OMIM:617190	MENDELIAN
ASXL3	Bainbridge-Ropers syndrome	OMIM:615485	MENDELIAN
ASXL3	Bainbridge-Ropers syndrome	ORPHA:352577	UNKNOWN
ATAD3A	Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal	OMIM:618810	MENDELIAN
ATAD3A	Harel-Yoon syndrome	OMIM:617183	MENDELIAN
ATAD3A	Ocular anomalies-axonal neuropathy-developmental delay syndrome	ORPHA:496790	UNKNOWN
ATCAY	Cerebellar ataxia, Cayman type	OMIM:601238	MENDELIAN
ATCAY	Cerebellar ataxia, Cayman type	ORPHA:94122	UNKNOWN
ATF3	—		UNKNOWN
ATF5	—		UNKNOWN
ATF6	Achromatopsia 7	OMIM:616517	MENDELIAN
ATF6	Cone rod dystrophy	ORPHA:1872	UNKNOWN
ATF6	Achromatopsia	ORPHA:49382	UNKNOWN
ATG7	Spinocerebellar ataxia, autosomal recessive 31	OMIM:619422	MENDELIAN
ATIC	AICA-ribosiduria due to ATIC deficiency	OMIM:608688	MENDELIAN
ATIC	AICA-ribosiduria	ORPHA:250977	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
ATL1	Spastic paraplegia 3, autosomal dominant	OMIM:182600	MENDELIAN
ATL1	Neuropathy, hereditary sensory, type ID	OMIM:613708	MENDELIAN
ATL1	Hereditary sensory and autonomic neuropathy type 1	ORPHA:36386	UNKNOWN
ATL1	Autosomal dominant spastic paraplegia type 3	ORPHA:100984	UNKNOWN
ATL3	Neuropathy, hereditary sensory, type IF	OMIM:615632	MENDELIAN
ATL3	Hereditary sensory and autonomic neuropathy type 1	ORPHA:36386	UNKNOWN
ATM	Ataxia-telangiectasia	OMIM:208900	MENDELIAN
ATM	Breast cancer	OMIM:114480	POLYGENIC
ATM	Mantle cell lymphoma	ORPHA:52416	UNKNOWN
ATM	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
ATM	Ataxia-telangiectasia	ORPHA:100	UNKNOWN
ATM	Familial colorectal cancer Type X	ORPHA:440437	UNKNOWN
ATN1	Dentatorubral-pallidoluysian atrophy	OMIM:125370	MENDELIAN
ATN1	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies	OMIM:618494	MENDELIAN
ATN1	Dentatorubral pallidoluysian atrophy	ORPHA:101	UNKNOWN
ATOH7	Persistent hyperplastic primary vitreous, autosomal recessive	OMIM:221900	MENDELIAN
ATOH7	Persistent hyperplastic primary vitreous	ORPHA:91495	UNKNOWN
ATP10A	Angelman syndrome due to imprinting defect in 15q11-q13	ORPHA:411515	UNKNOWN
ATP10D	—		UNKNOWN
ATP11B	—		UNKNOWN
ATP13A2	Kufor-Rakeb syndrome	OMIM:606693	MENDELIAN
ATP13A2	Spastic paraplegia 78, autosomal recessive	OMIM:617225	MENDELIAN
ATP13A2	Autosomal recessive spastic paraplegia type 78	ORPHA:513436	UNKNOWN
ATP13A2	Kufor-Rakeb syndrome	ORPHA:306674	UNKNOWN
ATP13A2	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
ATP13A2	CLN12 disease	ORPHA:314632	UNKNOWN
ATP13A4	—		UNKNOWN
ATP13A5	—		UNKNOWN
ATP1A1	Hypomagnesemia, seizures, and impaired intellectual development 2	OMIM:618314	MENDELIAN
ATP1A1	Charcot-Marie-Tooth disease, axonal, type 2DD	OMIM:618036	MENDELIAN
ATP1A1	Primary hypomagnesemia-refractory seizures-intellectual disability syndrome	ORPHA:564178	UNKNOWN
ATP1A2	Alternating hemiplegia of childhood 1	OMIM:104290	MENDELIAN
ATP1A2	Developmental and epileptic encephalopathy 98	OMIM:619605	MENDELIAN
ATP1A2	Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies	OMIM:619602	MENDELIAN
ATP1A2	Migraine, familial hemiplegic, 2	OMIM:602481	MENDELIAN
ATP1A2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
ATP1A2	Familial or sporadic hemiplegic migraine	ORPHA:569	UNKNOWN
ATP1A2	Alternating hemiplegia of childhood	ORPHA:2131	UNKNOWN
ATP1A3	Dystonia 12	OMIM:128235	MENDELIAN
ATP1A3	Developmental and epileptic encephalopathy 99	OMIM:619606	MENDELIAN
ATP1A3	Cerebellar ataxia, areflexia, pes cavus, optic atrophy, and sensorineural hearing loss	OMIM:601338	MENDELIAN
ATP1A3	Alternating hemiplegia of childhood 2	OMIM:614820	MENDELIAN
ATP1A3	Rapid-onset dystonia-parkinsonism	ORPHA:71517	UNKNOWN
ATP1A3	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
ATP1A3	Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome	ORPHA:1171	UNKNOWN
ATP1A3	Alternating hemiplegia of childhood	ORPHA:2131	UNKNOWN
ATP1B1	Hypertension, essential	OMIM:145500	MENDELIAN
ATP1B4	—		UNKNOWN
ATP2A1	Brody myopathy	OMIM:601003	MENDELIAN
ATP2A2	Acrokeratosis verruciformis	OMIM:101900	MENDELIAN
ATP2A2	Rhabdomyolysis, susceptibility to, 2	OMIM:621236	POLYGENIC
ATP2A2	Darier-White disease	OMIM:124200	MENDELIAN
ATP2A2	Darier disease	ORPHA:218	UNKNOWN
ATP2A2	Acrokeratosis verruciformis of Hopf	ORPHA:79151	UNKNOWN
ATP2A3	—		UNKNOWN
ATP2B1	Intellectual developmental disorder, autosomal dominant 66	OMIM:619910	MENDELIAN
ATP2B2	Deafness, autosomal dominant 82	OMIM:619804	MENDELIAN
ATP2B3	Spinocerebellar ataxia, X-linked 1	OMIM:302500	MENDELIAN
ATP2B3	X-linked non progressive cerebellar ataxia	ORPHA:314978	UNKNOWN
ATP2B4	—		UNKNOWN
ATP2C1	Benign chronic pemphigus	OMIM:169600	MENDELIAN
ATP2C1	Hailey-Hailey disease	ORPHA:2841	UNKNOWN
ATP5A1	—		UNKNOWN
ATP5B	—		UNKNOWN
ATP5C1	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
ATP5E	—		UNKNOWN
ATP5G1	—		UNKNOWN
ATP5G2	—		UNKNOWN
ATP5G3	—		UNKNOWN
ATP5I	—		UNKNOWN
ATP5J	—		UNKNOWN
ATP5O	—		UNKNOWN
ATP5SL	—		UNKNOWN
ATP6AP2	Parkinsonism with spasticity, X-linked	OMIM:300911	MENDELIAN
ATP6AP2	Congenital disorder of glycosylation, type IIr	OMIM:301045	MENDELIAN
ATP6AP2	Intellectual developmental disorder, X-linked syndromic, Hedera type	OMIM:300423	MENDELIAN
ATP6AP2	X-linked intellectual disability, Hedera type	ORPHA:93952	UNKNOWN
ATP6AP2	X-linked parkinsonism-spasticity syndrome	ORPHA:363654	UNKNOWN
ATP6V0A1	Neurodevelopmental disorder with epilepsy and brain atrophy	OMIM:619971	MENDELIAN
ATP6V0A1	Developmental and epileptic encephalopathy 104	OMIM:619970	MENDELIAN
ATP6V0A2	Wrinkly skin syndrome	OMIM:278250	MENDELIAN
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA	OMIM:219200	MENDELIAN
ATP6V0A2	Wrinkly skin syndrome	ORPHA:2834	UNKNOWN
ATP6V0A2	Autosomal recessive cutis laxa type 2, classic type	ORPHA:357074	UNKNOWN
ATP6V0A4	Distal renal tubular acidosis 3, with or without sensorineural hearing loss	OMIM:602722	MENDELIAN
ATP6V0C	Epilepsy, early-onset, 3, with or without developmental delay	OMIM:620465	MENDELIAN
ATP6V0E2	—		UNKNOWN
ATP6V1B1	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss	OMIM:267300	MENDELIAN
ATP6V1B2	Zimmermann-Laband syndrome 2	OMIM:616455	MENDELIAN
ATP6V1B2	Deafness, congenital, and onychodystrophy, autosomal dominant	OMIM:124480	MENDELIAN
ATP6V1B2	DOORS syndrome	ORPHA:79500	UNKNOWN
ATP6V1B2	Autosomal dominant deafness-onychodystrophy syndrome	ORPHA:79499	UNKNOWN
ATP6V1B2	Zimmermann-Laband syndrome	ORPHA:3473	UNKNOWN
ATP7A	Menkes disease	OMIM:309400	MENDELIAN
ATP7A	Spinal muscular atrophy, distal, X-linked 3	OMIM:300489	MENDELIAN
ATP7A	Occipital horn syndrome	OMIM:304150	MENDELIAN
ATP7A	Hirschsprung disease	ORPHA:388	UNKNOWN
ATP7A	Occipital horn syndrome	ORPHA:198	UNKNOWN
ATP7A	Menkes disease	ORPHA:565	UNKNOWN
ATP7B	Wilson disease	OMIM:277900	MENDELIAN
ATP7B	Wilson disease	ORPHA:905	UNKNOWN
ATP8A2	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4	OMIM:615268	MENDELIAN
ATP8A2	Dysequilibrium syndrome	ORPHA:1766	UNKNOWN
ATP8B1	Cholestasis, benign recurrent intrahepatic 1	OMIM:243300	MENDELIAN
ATP8B1	Cholestasis, intrahepatic, of pregnancy, 1	OMIM:147480	MENDELIAN
ATP8B1	Cholestasis, progressive familial intrahepatic, 3	OMIM:602347	MENDELIAN
ATP8B1	Cholestasis, progressive familial intrahepatic 1	OMIM:211600	MENDELIAN
ATP8B1	Intrahepatic cholestasis of pregnancy	ORPHA:69665	UNKNOWN
ATP9A	Neurodevelopmental disorder with poor growth and behavioral abnormalities	OMIM:620242	MENDELIAN
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	OMIM:604273	MENDELIAN
ATPAF2	Isolated ATP synthase deficiency	ORPHA:254913	UNKNOWN
ATR	Cutaneous telangiectasia and cancer syndrome, familial	OMIM:614564	MENDELIAN
ATR	Seckel syndrome 1	OMIM:210600	MENDELIAN
ATR	Seckel syndrome	ORPHA:808	UNKNOWN
ATRIP	Seckel syndrome	ORPHA:808	UNKNOWN
ATRN	—		UNKNOWN
ATRNL1	—		UNKNOWN
ATRX	Intellectual disability-hypotonic facies syndrome, X-linked	OMIM:309580	MENDELIAN
ATRX	Alpha-thalassemia/impaired intellectual development syndrome	OMIM:301040	MENDELIAN
ATRX	Alpha-Thalassemia myelodysplasia syndrome	OMIM:300448	MENDELIAN
ATRX	Cushing disease	ORPHA:96253	UNKNOWN
ATRX	X-linked alpha-thalassemia-intellectual disability syndrome	ORPHA:847	UNKNOWN
ATRX	Neuroendocrine tumor of stomach	ORPHA:100075	UNKNOWN
ATRX	Alpha-thalassemia-myelodysplastic syndrome	ORPHA:231401	UNKNOWN
ATXN1	Spinocerebellar ataxia 1	OMIM:164400	MENDELIAN
ATXN1	Spinocerebellar ataxia type 1	ORPHA:98755	UNKNOWN
ATXN10	Spinocerebellar ataxia 10	OMIM:603516	MENDELIAN
ATXN10	Spinocerebellar ataxia type 10	ORPHA:98761	UNKNOWN
ATXN2	Spinocerebellar ataxia 2	OMIM:183090	POLYGENIC
ATXN2	Parkinson disease, late-onset	OMIM:168600	POLYGENIC

Gene_symbol	Disease_name	Disease_ID	Association_type
ATXN2	Spinocerebellar ataxia type 2	ORPHA:98756	UNKNOWN
ATXN2	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
ATXN3	Machado-Joseph disease	OMIM:109150	MENDELIAN
ATXN3	Parkinson disease, late-onset	OMIM:168600	POLYGENIC
ATXN3	Machado-Joseph disease type 1	ORPHA:276238	UNKNOWN
ATXN3	Machado-Joseph disease type 3	ORPHA:276244	UNKNOWN
ATXN3	Machado-Joseph disease type 2	ORPHA:276241	UNKNOWN
ATXN3L	—		UNKNOWN
ATXN7	Spinocerebellar ataxia 7	OMIM:164500	MENDELIAN
ATXN7	Spinocerebellar ataxia type 7	ORPHA:94147	UNKNOWN
ATXN8	Spinocerebellar ataxia 8	OMIM:608768	MENDELIAN
ATXN8	Spinocerebellar ataxia type 8	ORPHA:98760	UNKNOWN
ATXN8OS	Spinocerebellar ataxia 8	OMIM:608768	MENDELIAN
ATXN8OS	Parkinson disease, late-onset	OMIM:168600	POLYGENIC
ATXN8OS	Spinocerebellar ataxia type 8	ORPHA:98760	UNKNOWN
AUH	3-methylglutaconic aciduria, type I	OMIM:250950	MENDELIAN
AUH	3-methylglutaconic aciduria type 1	ORPHA:67046	UNKNOWN
AUP1	—		UNKNOWN
AURKA	Colorectal cancer	OMIM:114500	POLYGENIC
AURKB	—		UNKNOWN
AURKC	Male infertility with large-headed, multiflagellar, polyploid spermatozoa	OMIM:243060	MENDELIAN
AUTS2	Intellectual developmental disorder, autosomal dominant 26	OMIM:615834	MENDELIAN
AUTS2	Autism spectrum disorder due to AUTS2 deficiency	ORPHA:352490	UNKNOWN
AVP	Diabetes insipidus, Neurohypophyseal type	OMIM:125700	MENDELIAN
AVP	Hereditary arginine vasopressin deficiency	ORPHA:30925	UNKNOWN
AVPR1A	—		UNKNOWN
AVPR1B	—		UNKNOWN
AVPR2	Diabetes insipidus, nephrogenic, X-linked	OMIM:304800	MENDELIAN
AVPR2	Nephrogenic syndrome of inappropriate antidiuresis	OMIM:300539	MENDELIAN
AVPR2	Arginine vasopressin resistance	ORPHA:223	UNKNOWN
AXIN1	Craniometadiaphyseal osteosclerosis with hip dysplasia	OMIM:620558	MENDELIAN
AXIN1	Hepatocellular carcinoma	OMIM:114550	MENDELIAN
AXIN1	Caudal duplication anomaly	OMIM:607864	MENDELIAN
AXIN2	Colorectal cancer	OMIM:114500	MENDELIAN
AXIN2	Oligodontia-colorectal cancer syndrome	OMIM:608615	MENDELIAN
AXIN2	AXIN2-related polyposis	ORPHA:401911	UNKNOWN
AXL	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
B2M	Amyloidosis, hereditary systemic 6	OMIM:620659	MENDELIAN
B2M	Immunodeficiency 43	OMIM:241600	MENDELIAN
B2M	Variant ABeta2M amyloidosis	ORPHA:314652	UNKNOWN
B3GALNT1	#111400 BLOOD GROUP, P1PK SYSTEMP(1) PHENOTYPE, INCLUDED;;P(2) PHENOTYPE, INCLUDED;;P1(k) PHENOTYPE, INCLUDED;;P2(k) PHENOTYPE, INCLUDED;;p PHENOTYPE, INCLUDED;;NOR POLYAGGLUTINATION SYNDROME, INCLUDED	OMIM:111400	MENDELIAN
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11)	OMIM:615181	MENDELIAN
B3GALNT2	Walker-Warburg syndrome	ORPHA:899	UNKNOWN
B3GALNT2	Muscle-eye-brain disease	ORPHA:588	UNKNOWN
B3GALT1	—		UNKNOWN
B3GALT2	—		UNKNOWN
B3GALT5	—		UNKNOWN
B3GALT6	Al-Gazali syndrome	OMIM:609465	MENDELIAN
B3GALT6	Ehlers-Danlos syndrome, spondylodysplastic type, 2	OMIM:615349	MENDELIAN
B3GALT6	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	OMIM:271640	MENDELIAN
B3GALT6	B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:536467	UNKNOWN
B3GALTTL	—		UNKNOWN
B3GAT1	—		UNKNOWN
B3GAT2	—		UNKNOWN
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects	OMIM:245600	MENDELIAN
B3GNT1	—		UNKNOWN
B3GNT2	—		UNKNOWN
B3GNT3	—		UNKNOWN
B3GNT4	—		UNKNOWN
B3GNT5	—		UNKNOWN
B3GNT6	—		UNKNOWN
B3GNT7	—		UNKNOWN
B3GNT8	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
B4GALNT1	Spastic paraplegia 26, autosomal recessive	OMIM:609195	MENDELIAN
B4GALNT1	Autosomal recessive spastic paraplegia type 26	ORPHA:101006	UNKNOWN
B4GALNT3	—		UNKNOWN
B4GALNT4	—		UNKNOWN
B4GALT1	Congenital disorder of glycosylation, type IID	OMIM:607091	MENDELIAN
B4GALT1	Combined low LDL and fibrinogen	OMIM:620364	MENDELIAN
B4GALT1	B4GALT1-CDG	ORPHA:79332	UNKNOWN
B4GALT2	—		UNKNOWN
B4GALT3	—		UNKNOWN
B4GALT4	—		UNKNOWN
B4GALT5	—		UNKNOWN
B4GALT6	—		UNKNOWN
B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic type, 1	OMIM:130070	MENDELIAN
B4GALT7	B4GALT7-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:75496	UNKNOWN
B9D1	Meckel syndrome, type 9	OMIM:614209	MENDELIAN
B9D1	Joubert syndrome 27	OMIM:617120	MENDELIAN
B9D1	Meckel syndrome	ORPHA:564	UNKNOWN
B9D1	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
B9D2	Meckel syndrome, type 10	OMIM:614175	MENDELIAN
B9D2	Meckel syndrome	ORPHA:564	UNKNOWN
B9D2	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
BAALC	—		UNKNOWN
BAAT	Bile acid conjugation defect 1	OMIM:619232	MENDELIAN
BABAM1	—		UNKNOWN
BACE1	—		UNKNOWN
BACH1	—		UNKNOWN
BAG3	Charcot-Marie-Tooth disease, axonal, type 2JJ	OMIM:621095	MENDELIAN
BAG3	Neuronopathy, distal hereditary motor, autosomal dominant 15	OMIM:621094	MENDELIAN
BAG3	Myopathy, myofibrillar, 6	OMIM:612954	MENDELIAN
BAG3	Cardiomyopathy, dilated, 1HH	OMIM:613881	MENDELIAN
BAG3	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
BAG6	—		UNKNOWN
BANF1	Nestor-Guillermo progeria syndrome	OMIM:614008	MENDELIAN
BANK1	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
BAP1	Tumor predisposition syndrome	OMIM:614327	MENDELIAN
BAP1	Melanoma, uveal, susceptibility to, 2	OMIM:606661	POLYGENIC
BAP1	Kury-Isidor syndrome	OMIM:619762	MENDELIAN
BAP1	Familial melanoma	ORPHA:618	UNKNOWN
BAP1	Meningioma	ORPHA:2495	UNKNOWN
BAP1	Pleural mesothelioma	ORPHA:50251	UNKNOWN
BARD1	Breast cancer	OMIM:114480	POLYGENIC
BARD1	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
BARX2	—		UNKNOWN
BAX	Leukemia, acute lymphocytic, susceptibility to, 1	OMIM:613065	MENDELIAN
BAX	Colorectal cancer	OMIM:114500	MENDELIAN
BAZ1B	Williams syndrome	ORPHA:904	UNKNOWN
BAZ2B	—		UNKNOWN
BBC3	—		UNKNOWN
BBIP1	Bardet-Biedl syndrome 18	OMIM:615995	MENDELIAN
BBIP1	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
BBS1	Bardet-Biedl syndrome 1	OMIM:209900	MENDELIAN
BBS1	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
BBS1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
BBS10	Bardet-Biedl syndrome 10	OMIM:615987	MENDELIAN
BBS10	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
BBS12	Bardet-Biedl syndrome 12	OMIM:615989	MENDELIAN
BBS12	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
BBS2	Retinitis pigmentosa 74	OMIM:616562	MENDELIAN
BBS2	Bardet-Biedl syndrome 2	OMIM:615981	MENDELIAN
BBS2	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
BBS2	Retinitis pigmentosa	ORPHA:791	UNKNOWN
BBS4	Bardet-Biedl syndrome 4	OMIM:615982	MENDELIAN
BBS4	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
BBS5	Bardet-Biedl syndrome 5	OMIM:615983	MENDELIAN
BBS5	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
BBS7	Bardet-Biedl syndrome 7	OMIM:615984	MENDELIAN
BBS7	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
BBS9	Bardet-Biedl syndrome 9	OMIM:615986	MENDELIAN
BBS9	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
BCAM	Blood group, Lutheran null	OMIM:247420	MENDELIAN
BCAP31	Deafness, dystonia, and cerebral hypomyelination	OMIM:300475	MENDELIAN
BCAP31	CADDS	ORPHA:369942	UNKNOWN
BCAP31	Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome	ORPHA:369939	UNKNOWN
BCAS2	—		UNKNOWN
BCAT1	—		UNKNOWN
BCAT2	Hypervalinemia or hyperleucine-isoleucinemia	OMIM:618850	MENDELIAN
BCHE	Butyrylcholinesterase deficiency	OMIM:617936	MENDELIAN
BCHE	Hereditary butyrylcholinesterase deficiency	ORPHA:132	UNKNOWN
BCKDHA	Maple syrup urine disease	OMIM:248600	MENDELIAN
BCKDHB	Maple syrup urine disease, type Ib	OMIM:620698	MENDELIAN
BCKDK	Branched-chain keto acid dehydrogenase kinase deficiency	OMIM:614923	MENDELIAN
BCL10	Testicular tumor, somatic	OMIM:273300	POLYGENIC
BCL10	Mesothelioma, malignant	OMIM:156240	POLYGENIC
BCL10	Lymphoma, non-hodgkin, familial	OMIM:605027	POLYGENIC
BCL10	Gastric lymphoma, primary	OMIM:137245	MENDELIAN
BCL10	Immunodeficiency 37	OMIM:616098	MENDELIAN
BCL10	MALT lymphoma	ORPHA:52417	UNKNOWN
BCL11A	Intellectual developmental disorder with persistence of fetal hemoglobin	OMIM:617101	MENDELIAN
BCL11A	Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome	ORPHA:251380	UNKNOWN
BCL11A	2p15p16.1 microdeletion syndrome	ORPHA:261349	UNKNOWN
BCL2	Follicular lymphoma	ORPHA:545	UNKNOWN
BCL2A1	—		UNKNOWN
BCL2L1	—		UNKNOWN
BCL2L11	—		UNKNOWN
BCL2L2	—		UNKNOWN
BCL2L2-PABPN1	—		UNKNOWN
BCL3	—		UNKNOWN
BCL6	Follicular lymphoma	ORPHA:545	UNKNOWN
BCL7A	—		UNKNOWN
BCL9	—		UNKNOWN
BCLAF1	—		UNKNOWN
BCMO1	—		UNKNOWN
BCOR	Microphthalmia, syndromic 1	OMIM:309800	MENDELIAN
BCOR	Microphthalmia, syndromic 2	OMIM:300166	MENDELIAN
BCOR	Oculofaciocardiodental syndrome	ORPHA:2712	UNKNOWN
BCOR	Microphthalmia, Lenz type	ORPHA:568	UNKNOWN
BCOR	Acute promyelocytic leukemia	ORPHA:520	UNKNOWN
BCORL1	Shukla-Vernon syndrome	OMIM:301029	MENDELIAN
BCR	Leukemia, acute lymphocytic, susceptibility to, 1	OMIM:613065	MENDELIAN
BCR	Leukemia, chronic myeloid	OMIM:608232	MENDELIAN
BCR	Distal 22q11.2 microdeletion syndrome	ORPHA:261330	UNKNOWN
BCR	Chronic myeloid leukemia	ORPHA:521	UNKNOWN
BCS1L	Gracile syndrome	OMIM:603358	MENDELIAN
BCS1L	Mitochondrial complex III deficiency, nuclear type 1	OMIM:124000	MENDELIAN
BCS1L	Bjornstad syndrome	OMIM:262000	MENDELIAN
BCS1L	GRACILE syndrome	ORPHA:53693	UNKNOWN
BCS1L	Björnstad syndrome	ORPHA:123	UNKNOWN
BDKRB2	—		UNKNOWN
BDNF	WAGR syndrome	ORPHA:893	UNKNOWN
BDNF	Congenital central hypoventilation syndrome	ORPHA:661	UNKNOWN
BDP1	Deafness, autosomal recessive 112	OMIM:618257	MENDELIAN
BEAN1	Spinocerebellar ataxia 31	OMIM:117210	MENDELIAN
BEAN1	Spinocerebellar ataxia type 31	ORPHA:217012	UNKNOWN
BEND2	—		UNKNOWN
BEST1	Bestrophinopathy, autosomal recessive	OMIM:611809	MENDELIAN
BEST1	Macular dystrophy, vitelliform, 2	OMIM:153700	MENDELIAN
BEST1	Vitreoretinochoroidopathy	OMIM:193220	MENDELIAN
BEST1	Retinitis pigmentosa-50	OMIM:613194	MENDELIAN
BEST1	Adult-onset foveomacular vitelliform dystrophy	ORPHA:99000	UNKNOWN
BEST1	Best vitelliform macular dystrophy	ORPHA:1243	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
BEST1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
BEST1	Isolated complete colobomatous microphthalmia	ORPHA:35612	UNKNOWN
BEX4	—		UNKNOWN
BFSP1	Cataract 33, multiple types	OMIM:611391	MENDELIAN
BFSP2	Cataract, autosomal dominant, multiple types 1	OMIM:611597	MENDELIAN
BGN	Meester-Loeys syndrome	OMIM:300989	MENDELIAN
BGN	Spondyloepimetaphyseal dysplasia, X-linked	OMIM:300106	MENDELIAN
BHLHA9	Syndactyly, mesoaxial synostotic, with phalangeal reduction	OMIM:609432	MENDELIAN
BHLHA9	Camptosynpolydactyly, complex	OMIM:607539	MENDELIAN
BHLHA9	Tibial aplasia-ectrodactyly syndrome	ORPHA:3329	UNKNOWN
BHLHA9	Gollop-Wolfgang complex	ORPHA:1986	UNKNOWN
BHLHA9	Mesoaxial synostotic syndactyly with phalangeal reduction	ORPHA:157801	UNKNOWN
BHLHE41	Short sleep, familial natural, 1	OMIM:612975	MENDELIAN
BHMT	—		UNKNOWN
BICC1	Renal dysplasia, cystic, susceptibility to	OMIM:601331	POLYGENIC
BICC1	Autosomal dominant polycystic kidney disease	ORPHA:730	UNKNOWN
BICD1	—		UNKNOWN
BICD2	Spinal muscular atrophy, lower extremity-predominant, 2, autosomaldominant	OMIM:615290	MENDELIAN
BICD2	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant	OMIM:618291	MENDELIAN
BICD2	BICD2-related autosomal dominant childhood-onset proximal spinal muscular atrophy	ORPHA:363454	UNKNOWN
BICRA	Coffin-Siris syndrome 12	OMIM:619325	MENDELIAN
BIN1	Myopathy, centronuclear, 2	OMIM:255200	MENDELIAN
BIN1	Autosomal recessive centronuclear myopathy	ORPHA:169186	UNKNOWN
BIN1	Autosomal dominant centronuclear myopathy	ORPHA:169189	UNKNOWN
BIRC3	MALT lymphoma	ORPHA:52417	UNKNOWN
BIRC5	—		UNKNOWN
BIVM	—		UNKNOWN
BLK	Maturity-onset diabetes of the young, type 11	OMIM:613375	MENDELIAN
BLK	MODY	ORPHA:552	UNKNOWN
BLK	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
BLM	Bloom syndrome	OMIM:210900	MENDELIAN
BLM	Bloom syndrome	ORPHA:125	UNKNOWN
BLMH	—		UNKNOWN
BLNK	Agammaglobulinemia 4, autosomal recessive	OMIM:613502	MENDELIAN
BLNK	Autosomal non-syndromic agammaglobulinemia	ORPHA:33110	UNKNOWN
BLOC1S3	Hermansky-Pudlak syndrome 8	OMIM:614077	MENDELIAN
BLOC1S5	Hermansky-Pudlak syndrome 11	OMIM:619172	MENDELIAN
BLOC1S6	Hermansky-Pudlak syndrome 9	OMIM:614171	MENDELIAN
BLVRA	Hyperbiliverdinemia	OMIM:614156	MENDELIAN
BMI1	—		UNKNOWN
BMP1	Osteogenesis imperfecta, type XIII	OMIM:614856	MENDELIAN
BMP10	—		UNKNOWN
BMP15	Ovarian dysgenesis 2	OMIM:300510	MENDELIAN
BMP15	46,XX gonadal dysgenesis	ORPHA:243	UNKNOWN
BMP2	Brachydactyly, type A2	OMIM:112600	MENDELIAN
BMP2	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1	OMIM:617877	MENDELIAN
BMP2	Hemochromatosis, type 1	OMIM:235200	POLYGENIC
BMP2	20p12.3 microdeletion syndrome	ORPHA:261295	UNKNOWN
BMP2	Brachydactyly type A2	ORPHA:93396	UNKNOWN
BMP2K	—		UNKNOWN
BMP3	—		UNKNOWN
BMP4	Orofacial cleft 11	OMIM:600625	MENDELIAN
BMP4	Microphthalmia, syndromic 6	OMIM:607932	MENDELIAN
BMP4	Cleft lip/palate	ORPHA:199306	UNKNOWN
BMP4	Stickler syndrome type 2	ORPHA:90654	UNKNOWN
BMP4	Microphthalmia with brain and digit anomalies	ORPHA:139471	UNKNOWN
BMP5	—		UNKNOWN
BMP7	—		UNKNOWN
BMPER	Diaphanospondylodysostosis	OMIM:608022	MENDELIAN
BMPER	Diaphanospondylodysostosis	ORPHA:66637	UNKNOWN
BMPR1A	Juvenile polyposis syndrome	OMIM:174900	MENDELIAN
BMPR1A	Polyposis syndrome, hereditary mixed, 2	OMIM:610069	MENDELIAN
BMPR1A	Generalized juvenile polyposis/juvenile polyposis coli	ORPHA:329971	UNKNOWN
BMPR1A	Juvenile polyposis of infancy	ORPHA:79076	UNKNOWN
BMPR1A	Hereditary mixed polyposis syndrome	ORPHA:157794	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
BMPR1A	Familial colorectal cancer Type X	ORPHA:440437	UNKNOWN
BMPR1B	Brachydactyly, type A2	OMIM:112600	MENDELIAN
BMPR1B	Acromesomelic dysplasia, Demirhan type	OMIM:609441	MENDELIAN
BMPR1B	Brachydactyly, type A1, D	OMIM:616849	MENDELIAN
BMPR1B	Fibular aplasia-complex brachydactyly syndrome	ORPHA:2639	UNKNOWN
BMPR1B	Brachydactyly type A2	ORPHA:93396	UNKNOWN
BMPR1B	Brachydactyly type C	ORPHA:93384	UNKNOWN
BMPR1B	Brachydactyly type A1	ORPHA:93388	UNKNOWN
BMPR1B	Acromesomelic dysplasia, Grebe type	ORPHA:2098	UNKNOWN
BMPR2	Pulmonary hypertension, primary, 1	OMIM:178600	MENDELIAN
BMPR2	Pulmonary venoocclusive disease 1	OMIM:265450	MENDELIAN
BMS1	Aplasia cutis congenita, nonsyndromic	OMIM:107600	MENDELIAN
BMS1	Aplasia cutis congenita	ORPHA:1114	UNKNOWN
BNC2	Lower urinary tract obstruction, congenital	OMIM:618612	MENDELIAN
BNC2	Posterior urethral valve	ORPHA:93110	UNKNOWN
BOC	—		UNKNOWN
BOLA1	—		UNKNOWN
BOLA2	—		UNKNOWN
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia	OMIM:614299	MENDELIAN
BOLA3	Multiple mitochondrial dysfunctions syndrome type 2	ORPHA:401874	UNKNOWN
BOLL	—		UNKNOWN
BPGM	Erythrocytosis, familial, 8	OMIM:222800	MENDELIAN
BPI	—		UNKNOWN
BPIFA1	—		UNKNOWN
BPIFA3	—		UNKNOWN
BPIFB6	—		UNKNOWN
BPY2	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
BRAF	Cardiofaciocutaneous syndrome 1	OMIM:115150	MENDELIAN
BRAF	Colorectal cancer	OMIM:114500	MENDELIAN
BRAF	Lung cancer, susceptibility to	OMIM:211980	MENDELIAN
BRAF	Leopard syndrome 3	OMIM:613707	MENDELIAN
BRAF	Noonan syndrome 7	OMIM:613706	MENDELIAN
BRAF	Melanoma, cutaneous malignant	OMIM:155600	MENDELIAN
BRAF	Noonan syndrome 1	OMIM:163950	MENDELIAN
BRAF	Cushing disease	ORPHA:96253	UNKNOWN
BRAF	Cardiofaciocutaneous syndrome	ORPHA:1340	UNKNOWN
BRAF	Craniopharyngioma	ORPHA:54595	UNKNOWN
BRAF	Syringocystadenoma papilliferum	ORPHA:840	UNKNOWN
BRAF	Large/giant congenital melanocytic nevus	ORPHA:626	UNKNOWN
BRAF	Noonan syndrome	ORPHA:648	UNKNOWN
BRAF	Noonan syndrome with multiple lentigines	ORPHA:500	UNKNOWN
BRAP	—		UNKNOWN
BRAT1	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	OMIM:618056	MENDELIAN
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal	OMIM:614498	MENDELIAN
BRCA1	Fanconi anemia, complementation group S	OMIM:617883	MENDELIAN
BRCA1	Breast-Ovarian cancer, familial, susceptibility to, 1	OMIM:604370	POLYGENIC
BRCA1	Breast cancer	OMIM:114480	MENDELIAN
BRCA1	Fanconi anemia	ORPHA:84	UNKNOWN
BRCA1	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
BRCA1	Primary peritoneal carcinoma	ORPHA:168829	UNKNOWN
BRCA1	Familial pancreatic carcinoma	ORPHA:1333	UNKNOWN
BRCA1	Cholangiocarcinoma	ORPHA:70567	UNKNOWN
BRCA2	Fanconi anemia, complementation group D1	OMIM:605724	MENDELIAN
BRCA2	Glioma susceptibility 3	OMIM:613029	POLYGENIC
BRCA2	Wilms tumor 1	OMIM:194070	MENDELIAN
BRCA2	Breast cancer	OMIM:114480	POLYGENIC
BRCA2	Pancreatic cancer, susceptibility to, 2	OMIM:613347	POLYGENIC
BRCA2	Prostate cancer	OMIM:176807	POLYGENIC
BRCA2	Breast-Ovarian cancer, familial, susceptibility to, 2	OMIM:612555	POLYGENIC
BRCA2	Medulloblastoma	OMIM:155255	POLYGENIC
BRCA2	Fanconi anemia	ORPHA:84	UNKNOWN
BRCA2	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
BRCA2	Familial pancreatic carcinoma	ORPHA:1333	UNKNOWN
BRCA2	Cholangiocarcinoma	ORPHA:70567	UNKNOWN
BRCA2	Nephroblastoma	ORPHA:654	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
BRCA2	Familial colorectal cancer Type X	ORPHA:440437	UNKNOWN
BRCC3	Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome	ORPHA:280679	UNKNOWN
BRD1	—		UNKNOWN
BRD2	—		UNKNOWN
BRD4	Cornelia de Lange syndrome 6	OMIM:620568	MENDELIAN
BRD4	Cornelia de Lange syndrome	ORPHA:199	UNKNOWN
BRD4	NUT midline carcinoma	ORPHA:443167	UNKNOWN
BRDT	Spermatogenic failure 21	OMIM:617644	MENDELIAN
BRF1	Cerebellofaciodental syndrome	OMIM:616202	MENDELIAN
BRF1	Cerebellar-facial-dental syndrome	ORPHA:444072	UNKNOWN
BRIP1	Fanconi anemia, complementation group J	OMIM:609054	MENDELIAN
BRIP1	Breast cancer	OMIM:114480	POLYGENIC
BRIP1	Fanconi anemia	ORPHA:84	UNKNOWN
BRIP1	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
BRWD1	Ciliary dyskinesia, primary, 51	OMIM:620438	MENDELIAN
BRWD1	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
BRWD3	Intellectual developmental disorder, X-linked 93	OMIM:300659	MENDELIAN
BSCL2	Lipodystrophy, congenital generalized, type 2	OMIM:269700	MENDELIAN
BSCL2	Spastic paraplegia 17	OMIM:270685	MENDELIAN
BSCL2	Neuropathy, distal hereditary motor, type VC	OMIM:619112	MENDELIAN
BSCL2	Encephalopathy, progressive, with or without lipodystrophy	OMIM:615924	MENDELIAN
BSCL2	Distal hereditary motor neuropathy type 5	ORPHA:139536	UNKNOWN
BSCL2	Progressive encephalopathy-severe neurodegeneration-lipodystrophy syndrome	ORPHA:363400	UNKNOWN
BSCL2	Autosomal dominant spastic paraplegia type 17	ORPHA:100998	UNKNOWN
BSND	Bartter syndrome, type 4A, neonatal, with sensorineural deafness	OMIM:602522	MENDELIAN
BSND	Bartter syndrome type 4	ORPHA:89938	UNKNOWN
BST1	—		UNKNOWN
BTAF1	—		UNKNOWN
BTBD9	—		UNKNOWN
BTC	—		UNKNOWN
BTD	Biotinidase deficiencymultiple carboxylase deficiency, late-onset	OMIM:253260	MENDELIAN
BTD	Biotinidase deficiency	ORPHA:79241	UNKNOWN
BTG1	—		UNKNOWN
BTK	Isolated growth hormone deficiency, type III, with agammaglobulinemia	OMIM:307200	MENDELIAN
BTK	Agammaglobulinemia, X-linked	OMIM:300755	MENDELIAN
BTK	X-linked agammaglobulinemia	ORPHA:47	UNKNOWN
BTLA	—		UNKNOWN
BTN1A1	—		UNKNOWN
BTN2A1	—		UNKNOWN
BTNL2	Sarcoidosis, susceptibility to, 2	OMIM:612387	POLYGENIC
BTNL2	Sarcoidosis	ORPHA:797	UNKNOWN
BTRC	Isolated split hand-split foot malformation	ORPHA:2440	UNKNOWN
BUB1	Microcephaly 30, primary, autosomal recessive	OMIM:620183	MENDELIAN
BUB1	Colorectal cancer	OMIM:114500	MENDELIAN
BUB1	Mosaic variegated aneuploidy syndrome	ORPHA:1052	UNKNOWN
BUB1B	Premature chromatid separation trait	OMIM:176430	MENDELIAN
BUB1B	Colorectal cancer	OMIM:114500	MENDELIAN
BUB1B	Mosaic variegated aneuploidy syndrome 1	OMIM:257300	MENDELIAN
BUB1B	Mosaic variegated aneuploidy syndrome	ORPHA:1052	UNKNOWN
BUB3	Mosaic variegated aneuploidy syndrome	ORPHA:1052	UNKNOWN
BVES	—		UNKNOWN
C10orf11	—		UNKNOWN
C10orf137	—		UNKNOWN
C10orf2	—		UNKNOWN
C11orf30	—		UNKNOWN
C11ORF40	—		UNKNOWN
C12orf10	—		UNKNOWN
C12ORF4	—		UNKNOWN
C12orf57	Temtam syndrome	OMIM:218340	MENDELIAN
C12orf65	—		UNKNOWN
C12ORF66	—		UNKNOWN
C14ORF105	—		UNKNOWN
C14ORF2	—		UNKNOWN
C14orf39	Spermatogenic failure 52	OMIM:619202	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
C14orf39	Premature ovarian failure 18	OMIM:619203	MENDELIAN
C15ORF41	—		UNKNOWN
C15ORF62	—		UNKNOWN
C16ORF58	—		UNKNOWN
C17ORF70	—		UNKNOWN
C18ORF8	—		UNKNOWN
C19orf12	Spastic paraplegia 43, autosomal recessive	OMIM:615043	MENDELIAN
C19orf12	Neurodegeneration with brain iron accumulation 4	OMIM:614298	MENDELIAN
C19ORF40	—		UNKNOWN
C1GALT1	—		UNKNOWN
C1GALT1C1	Hemolytic uremic syndrome, atypical, 8, with rhizomelic short stature	OMIM:301110	MENDELIAN
C1GALT1C1	Tn polyagglutination syndrome	OMIM:300622	MENDELIAN
C1ORF127	—		UNKNOWN
C1orf146	—		UNKNOWN
C1ORF168	—		UNKNOWN
C1QA	C1q deficiency	OMIM:613652	MENDELIAN
C1QB	C1q deficiency 2	OMIM:620321	MENDELIAN
C1QC	C1q deficiency 3	OMIM:620322	MENDELIAN
C1QTNF5	Retinal degeneration, late-onset, autosomal dominant	OMIM:605670	MENDELIAN
C1QTNF5	Late-onset retinal degeneration	ORPHA:67042	UNKNOWN
C1R	Ehlers-Danlos syndrome, periodontal type, 1	OMIM:130080	MENDELIAN
C1R	Periodontal Ehlers-Danlos syndrome	ORPHA:75392	UNKNOWN
C1S	Complement component c1s deficiency	OMIM:613783	MENDELIAN
C1S	Ehlers-Danlos syndrome, periodontal type, 2	OMIM:617174	MENDELIAN
C1S	Periodontal Ehlers-Danlos syndrome	ORPHA:75392	UNKNOWN
C2	Complement component 2 deficiency	OMIM:217000	MENDELIAN
C21ORF2	—		UNKNOWN
C21ORF59	—		UNKNOWN
C21ORF91	—		UNKNOWN
C2CD3	Orofaciodigital syndrome XIV	OMIM:615948	MENDELIAN
C2CD3	Orofaciodigital syndrome type 14	ORPHA:434179	UNKNOWN
C2CD5	—		UNKNOWN
C2ORF42	—		UNKNOWN
C2orf71	—		UNKNOWN
C2ORF80	—		UNKNOWN
C3	Hemolytic uremic syndrome, atypical, susceptibility to, 5	OMIM:612925	POLYGENIC
C3	Complement component 3 deficiency, autosomal recessive	OMIM:613779	MENDELIAN
C3AR1	—		UNKNOWN
C3ORF35	—		UNKNOWN
C3orf70	—		UNKNOWN
C4A	Complement component 4A deficiency	OMIM:614380	MENDELIAN
C4A	Behçet disease	ORPHA:117	UNKNOWN
C4A	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
C4B	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
C4BPA	—		UNKNOWN
C4ORF19	—		UNKNOWN
C4ORF26	—		UNKNOWN
C5	C5 deficiency	OMIM:609536	MENDELIAN
C5AR2	—		UNKNOWN
C5orf42	—		UNKNOWN
C6	C6 deficiency	OMIM:612446	MENDELIAN
C6orf15	—		UNKNOWN
C7	C7 deficiency	OMIM:610102	MENDELIAN
C7orf10	—		UNKNOWN
C7ORF43	—		UNKNOWN
C8A	Complement component 8 deficiency, type I	OMIM:613790	MENDELIAN
C8B	Complement component 8 deficiency, type II	OMIM:613789	MENDELIAN
C8orf37	—		UNKNOWN
C9	Macular degeneration, age-related, 15, susceptibility to	OMIM:615591	POLYGENIC
C9	Complement component 9 deficiency	OMIM:613825	MENDELIAN
C9orf72	Amyotrophic lateral sclerosis and/or frontotemporal dementia 1	OMIM:105550	MENDELIAN
CA1	—		UNKNOWN
CA12	Hyperchlorhidrosis, isolated	OMIM:143860	MENDELIAN
CA2	Osteopetrosis, autosomal recessive 3	OMIM:259730	MENDELIAN
CA2	Osteopetrosis with renal tubular acidosis	ORPHA:2785	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
CA4	Retinitis pigmentosa	ORPHA:791	UNKNOWN
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency	OMIM:615751	MENDELIAN
CA6	—		UNKNOWN
CA8	Spinocerebellar ataxia, autosomal recessive 34	OMIM:613227	MENDELIAN
CA8	Dysequilibrium syndrome	ORPHA:1766	UNKNOWN
CABIN1	—		UNKNOWN
CABP2	Deafness, autosomal recessive 93	OMIM:614899	MENDELIAN
CABP4	Cone-Rod synaptic disorder, congenital nonprogressive	OMIM:610427	MENDELIAN
CABP4	Leber congenital amaurosis	ORPHA:65	UNKNOWN
CABP4	Sleep-related hypermotor epilepsy	ORPHA:98784	UNKNOWN
CABP4	Severe early-childhood-onset retinal dystrophy	ORPHA:364055	UNKNOWN
CACHD1	Davis-Wells syndrome	OMIM:621566	MENDELIAN
CACNA1A	Spinocerebellar ataxia 6	OMIM:183086	MENDELIAN
CACNA1A	Migraine, familial hemiplegic, 1	OMIM:141500	MENDELIAN
CACNA1A	Episodic ataxia, type 2	OMIM:108500	MENDELIAN
CACNA1A	Epileptic encephalopathy, early infantile, 42	OMIM:617106	MENDELIAN
CACNA1A	Familial paroxysmal ataxia	ORPHA:97	UNKNOWN
CACNA1A	Benign paroxysmal torticollis of infancy	ORPHA:71518	UNKNOWN
CACNA1A	Spinocerebellar ataxia type 6	ORPHA:98758	UNKNOWN
CACNA1A	Lennox-Gastaut syndrome	ORPHA:2382	UNKNOWN
CACNA1A	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
CACNA1A	Familial or sporadic hemiplegic migraine	ORPHA:569	UNKNOWN
CACNA1A	Alternating hemiplegia of childhood	ORPHA:2131	UNKNOWN
CACNA1B	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements	OMIM:618497	MENDELIAN
CACNA1B	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
CACNA1C	Timothy syndrome	OMIM:601005	MENDELIAN
CACNA1C	Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures	OMIM:620029	MENDELIAN
CACNA1C	Brugada syndrome 3	OMIM:611875	MENDELIAN
CACNA1C	Long QT syndrome 8	OMIM:618447	MENDELIAN
CACNA1C	Brugada syndrome	ORPHA:130	UNKNOWN
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities	OMIM:615474	MENDELIAN
CACNA1D	Sinoatrial node dysfunction and deafness	OMIM:614896	MENDELIAN
CACNA1D	Primary hyperaldosteronism-seizures-neurological abnormalities syndrome	ORPHA:369929	UNKNOWN
CACNA1E	Developmental and epileptic encephalopathy 69	OMIM:618285	MENDELIAN
CACNA1E	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
CACNA1F	Night blindness, congenital stationary, type 2A	OMIM:300071	MENDELIAN
CACNA1F	Cone-Rod dystrophy, X-linked, 3	OMIM:300476	MENDELIAN
CACNA1F	Aland island eye disease	OMIM:300600	MENDELIAN
CACNA1F	Å...land Islands eye disease	ORPHA:178333	UNKNOWN
CACNA1F	Cone rod dystrophy	ORPHA:1872	UNKNOWN
CACNA1G	Spinocerebellar ataxia 42	OMIM:616795	MENDELIAN
CACNA1G	Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits	OMIM:618087	MENDELIAN
CACNA1G	Spinocerebellar ataxia type 42	ORPHA:458803	UNKNOWN
CACNA1H	Hyperaldosteronism, familial, type IV	OMIM:617027	MENDELIAN
CACNA1H	Childhood absence epilepsy	ORPHA:64280	UNKNOWN
CACNA1S	Congenital myopathy 18 due to dihydropyridine receptor defect	OMIM:620246	MENDELIAN
CACNA1S	Hypokalemic periodic paralysis, type 1	OMIM:170400	MENDELIAN
CACNA1S	Thyrotoxic periodic paralysis, susceptibility to, 1	OMIM:188580	POLYGENIC
CACNA1S	Malignant hyperthermia, susceptibility to, 5	OMIM:601887	POLYGENIC
CACNA1S	Thyrotoxic periodic paralysis	ORPHA:79102	UNKNOWN
CACNA1S	Hypokalemic periodic paralysis	ORPHA:681	UNKNOWN
CACNA1S	Malignant hyperthermia of anesthesia	ORPHA:423	UNKNOWN
CACNA2D1	Developmental and epileptic encephalopathy 110	OMIM:620149	MENDELIAN
CACNA2D1	Congenital short QT syndrome	ORPHA:51083	UNKNOWN
CACNA2D1	Brugada syndrome	ORPHA:130	UNKNOWN
CACNA2D1	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
CACNA2D2	Cerebellar atrophy with seizures and variable developmental delay	OMIM:618501	MENDELIAN
CACNA2D3	—		UNKNOWN
CACNA2D4	Retinal cone dystrophy 4	OMIM:610478	MENDELIAN
CACNA2D4	Cone rod dystrophy	ORPHA:1872	UNKNOWN
CACNB2	Brugada syndrome 4	OMIM:611876	MENDELIAN
CACNB2	Brugada syndrome	ORPHA:130	UNKNOWN
CACNB4	Epilepsy, idiopathic generalized, susceptibility to, 9	OMIM:607682	POLYGENIC
CACNB4	Episodic ataxia, type 5	OMIM:613855	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
CACNB4	Juvenile myoclonic epilepsy	ORPHA:307	UNKNOWN
CACNB4	Episodic ataxia type 5	ORPHA:211067	UNKNOWN
CACNG2	Intellectual developmental disorder, autosomal dominant 10	OMIM:614256	MENDELIAN
CACNG3	—		UNKNOWN
CACNG4	—		UNKNOWN
CADM1	—		UNKNOWN
CADPS2	—		UNKNOWN
CALCA	—		UNKNOWN
CALCR	Osteoporosis	OMIM:166710	POLYGENIC
CALCRL	Lymphatic malformation 8	OMIM:618773	MENDELIAN
CALHM1	—		UNKNOWN
CALM1	Long QT syndrome 14	OMIM:616247	MENDELIAN
CALM1	Ventricular tachycardia, catecholaminergic polymorphic, 4	OMIM:614916	MENDELIAN
CALM2	Long QT syndrome 15	OMIM:616249	MENDELIAN
CALM3	Long QT syndrome 16	OMIM:618782	MENDELIAN
CALR	Myelofibrosis with myeloid metaplasia, somatic	OMIM:254450	MENDELIAN
CALR	Thrombocythemia 1	OMIM:187950	MENDELIAN
CALR	Essential thrombocythemia	ORPHA:3318	UNKNOWN
CALR	Budd-Chiari syndrome	ORPHA:131	UNKNOWN
CALR	Primary myelofibrosis	ORPHA:824	UNKNOWN
CALR3	—		UNKNOWN
CAMK2A	Intellectual developmental disorder, autosomal dominant 53	OMIM:617798	MENDELIAN
CAMK2A	Intellectual developmental disorder, autosomal recessive 63	OMIM:618095	MENDELIAN
CAMK2B	Intellectual developmental disorder, autosomal dominant 54	OMIM:617799	MENDELIAN
CAMK2D	—		UNKNOWN
CAMK2G	Intellectual developmental disorder 59	OMIM:618522	MENDELIAN
CAMK4	—		UNKNOWN
CAMKK1	—		UNKNOWN
CAMKK2	—		UNKNOWN
CAMKMT	2p21 microdeletion syndrome	ORPHA:163693	UNKNOWN
CAMP	—		UNKNOWN
CAMSAP2	—		UNKNOWN
CAMTA1	Cerebellar dysfunction with variable cognitive and behavioral abnormalities	OMIM:614756	MENDELIAN
CAMTA1	Non-progressive cerebellar ataxia with intellectual disability	ORPHA:314647	UNKNOWN
CAMTA2	—		UNKNOWN
CANT1	Epiphyseal dysplasia, multiple, 7	OMIM:617719	MENDELIAN
CANT1	Desbuquois dysplasia 1	OMIM:251450	MENDELIAN
CANT1	Desbuquois syndrome	ORPHA:1425	UNKNOWN
CAPN1	Spastic paraplegia 76, autosomal recessive	OMIM:616907	MENDELIAN
CAPN1	Autosomal recessive spastic paraplegia type 76	ORPHA:488594	UNKNOWN
CAPN12	—		UNKNOWN
CAPN13	—		UNKNOWN
CAPN3	Muscular dystrophy, limb-girdle, type 2A	OMIM:253600	MENDELIAN
CAPN3	Muscular dystrophy, limb-girdle, autosomal dominant 4	OMIM:618129	MENDELIAN
CAPN3	Calpain-3-related limb-girdle muscular dystrophy R1	ORPHA:267	UNKNOWN
CAPN5	Vitreoretinopathy, neovascular inflammatory	OMIM:193235	MENDELIAN
CAPRIN1	Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline	OMIM:620636	MENDELIAN
CAPRIN1	Neurodevelopmental disorder with language impairment, autism, and attention deficit-hyperactivity disorder	OMIM:620782	MENDELIAN
CARD11	Card11 immunodeficiency	OMIM:615206	MENDELIAN
CARD11	B-cell expansion with NFkB and T-cell anergy	OMIM:616452	MENDELIAN
CARD11	Immunodeficiency 11B with atopic dermatitis	OMIM:617638	MENDELIAN
CARD14	Psoriasis 2	OMIM:602723	MENDELIAN
CARD14	Pityriasis rubra pilaris	OMIM:173200	MENDELIAN
CARD14	Pityriasis rubra pilaris	ORPHA:2897	UNKNOWN
CARD8	Inflammatory bowel disease (Crohn disease) 30	OMIM:619079	MENDELIAN
CARD9	Candidiasis, familial chronic mucocutaneous, autosomal recessive	OMIM:212050	MENDELIAN
CARS2	Combined oxidative phosphorylation deficiency 27	OMIM:616672	MENDELIAN
CARS2	Combined oxidative phosphorylation defect type 27	ORPHA:477774	UNKNOWN
CARTPT	Obesity	OMIM:601665	POLYGENIC
CASC16	—		UNKNOWN
CASC4	—		UNKNOWN
CASC5	—		UNKNOWN
CASK	Hemolytic anemia, G6PD deficient (favism)	OMIM:300908	MENDELIAN
CASK	Fg syndrome 4	OMIM:300422	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
CASK	Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia	OMIM:300749	MENDELIAN
CASK	X-linked intellectual disability, Najm type	ORPHA:163937	UNKNOWN
CASK	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
CASP1	—		UNKNOWN
CASP10	Lymphoma, non-hodgkin, familial	OMIM:605027	MENDELIAN
CASP10	Autoimmune lymphoproliferative syndrome, type IIA	OMIM:603909	MENDELIAN
CASP10	Gastric cancer, somatic	OMIM:613659	MENDELIAN
CASP10	Autoimmune lymphoproliferative syndrome	ORPHA:3261	UNKNOWN
CASP12	—		UNKNOWN
CASP2	Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly	OMIM:620653	MENDELIAN
CASP3	—		UNKNOWN
CASP5	—		UNKNOWN
CASP8	Autoimmune lymphoproliferative syndrome, type IIB	OMIM:607271	MENDELIAN
CASP8	Breast cancer	OMIM:114480	POLYGENIC
CASP8	Hepatocellular carcinoma	OMIM:114550	MENDELIAN
CASP8	Lung cancer, susceptibility to	OMIM:211980	POLYGENIC
CASP9	—		UNKNOWN
CASQ1	Myopathy, vacuolar, with CASQ1 aggregates	OMIM:616231	MENDELIAN
CASQ1	Tubular aggregate myopathy	ORPHA:2593	UNKNOWN
CASQ1	Vacuolar myopathy with sarcoplasmic reticulum protein aggregates	ORPHA:88635	UNKNOWN
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 1, with or without atrial dysfunction and/or dilated cardiomyopathy	OMIM:604772	MENDELIAN
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2	OMIM:611938	MENDELIAN
CASR	Epilepsy idiopathic generalized, susceptibility to, 8	OMIM:612899	POLYGENIC
CASR	Hyperparathyroidism, neonatal severe	OMIM:239200	MENDELIAN
CASR	Hypocalcemia, autosomal dominant 1	OMIM:601198	MENDELIAN
CASR	Hypocalciuric hypercalcemia, familial, type I	OMIM:145980	MENDELIAN
CASR	Autosomal dominant hereditary chronic pancreatitis	ORPHA:676	UNKNOWN
CASR	Autosomal dominant hypocalcemia	ORPHA:428	UNKNOWN
CASR	Neonatal severe primary hyperparathyroidism	ORPHA:417	UNKNOWN
CAST	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads	OMIM:616295	MENDELIAN
CASZ1	1p36 deletion syndrome	ORPHA:1606	UNKNOWN
CAT	Acatalasemia	OMIM:614097	MENDELIAN
CAT	Acatalasemia	ORPHA:926	UNKNOWN
CATSPER1	Spermatogenic failure 7	OMIM:612997	MENDELIAN
CATSPER2	Spermatogenic failure 7	OMIM:612997	MENDELIAN
CATSPER2	Deafness, sensorineural, and male infertility	OMIM:611102	MENDELIAN
CATSPER2	Deafness-infertility syndrome	ORPHA:94064	UNKNOWN
CATSPER3	—		UNKNOWN
CATSPER4	—		UNKNOWN
CAV1	Pulmonary hypertension, primary, 3	OMIM:615343	MENDELIAN
CAV1	Lipodystrophy, familial partial, type 7	OMIM:606721	MENDELIAN
CAV1	Lipodystrophy, congenital generalized, type 3	OMIM:612526	MENDELIAN
CAV1	Limited cutaneous systemic sclerosis	ORPHA:220402	UNKNOWN
CAV1	Diffuse cutaneous systemic sclerosis	ORPHA:220393	UNKNOWN
CAV3	Creatine phosphokinase, elevated serum	OMIM:123320	MENDELIAN
CAV3	Cardiomyopathy, familial hypertrophic 1	OMIM:192600	MENDELIAN
CAV3	Myopathy, distal, Tateyama type	OMIM:614321	MENDELIAN
CAV3	Long QT syndrome 9	OMIM:611818	MENDELIAN
CAV3	Rippling muscle disease	OMIM:606072	MENDELIAN
CAV3	Distal myopathy, Tateyama type	ORPHA:488650	UNKNOWN
CAV3	Isolated asymptomatic elevation of creatine phosphokinase	ORPHA:206599	UNKNOWN
CBFA2T2	—		UNKNOWN
CBFB	Cleidocranial dysplasia 2	OMIM:620099	MENDELIAN
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	OMIM:613563	MENDELIAN
CBL	Juvenile myelomonocytic leukemia	OMIM:607785	MENDELIAN
CBL	Aggressive systemic mastocytosis	ORPHA:98850	UNKNOWN
CBL	Noonan syndrome	ORPHA:648	UNKNOWN
CBLB	Autoimmune disease, multisystem, infantile-onset, 3	OMIM:620430	MENDELIAN
CBR1	—		UNKNOWN
CBR3	—		UNKNOWN
CBS	Homocystinuria, B6-responsive and nonresponsive types	OMIM:236200	MENDELIAN
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	ORPHA:394	UNKNOWN
CBX1	—		UNKNOWN
CBX2	46XY sex reversal 5	OMIM:613080	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
CBX2	46,XY complete gonadal dysgenesis	ORPHA:242	UNKNOWN
CBX4	—		UNKNOWN
CBY1	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
CC2D1A	Intellectual developmental disorder, autosomal recessive 3	OMIM:608443	MENDELIAN
CC2D2A	Coach syndrome 2	OMIM:619111	MENDELIAN
CC2D2A	Retinitis pigmentosa 93	OMIM:619845	MENDELIAN
CC2D2A	Joubert syndrome 9	OMIM:612285	MENDELIAN
CC2D2A	Meckel syndrome, type 6	OMIM:612284	MENDELIAN
CC2D2A	Joubert syndrome with hepatic defect	ORPHA:1454	UNKNOWN
CC2D2A	Retinitis pigmentosa	ORPHA:791	UNKNOWN
CC2D2A	Joubert syndrome with oculorenal defect	ORPHA:2318	UNKNOWN
CC2D2A	Meckel syndrome	ORPHA:564	UNKNOWN
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome	OMIM:235510	MENDELIAN
CCBE1	Hennekam syndrome	ORPHA:2136	UNKNOWN
CCDC103	—		UNKNOWN
CCDC107	—		UNKNOWN
CCDC109B	—		UNKNOWN
CCDC11	—		UNKNOWN
CCDC114	—		UNKNOWN
CCDC127	—		UNKNOWN
CCDC14	—		UNKNOWN
CCDC148	—		UNKNOWN
CCDC151	—		UNKNOWN
CCDC170	—		UNKNOWN
CCDC18	—		UNKNOWN
CCDC22	Ritscher-Schinzel syndrome 2	OMIM:300963	MENDELIAN
CCDC22	3C syndrome	ORPHA:7	UNKNOWN
CCDC28B	Bardet-Biedl syndrome 1	OMIM:209900	POLYGENIC
CCDC33	—		UNKNOWN
CCDC39	Ciliary dyskinesia, primary, 14	OMIM:613807	MENDELIAN
CCDC39	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
CCDC40	Ciliary dyskinesia, primary, 15	OMIM:613808	MENDELIAN
CCDC40	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
CCDC50	Deafness, autosomal dominant 44	OMIM:607453	MENDELIAN
CCDC62	Spermatogenic failure 67	OMIM:619803	MENDELIAN
CCDC65	—		UNKNOWN
CCDC66	—		UNKNOWN
CCDC78	Myopathy, centronuclear, 4	OMIM:614807	MENDELIAN
CCDC8	3-M syndrome 3	OMIM:614205	MENDELIAN
CCDC8	3M syndrome	ORPHA:2616	UNKNOWN
CCDC85C	—		UNKNOWN
CCDC88C	Hydrocephalus, nonsyndromic, autosomal recessive 1	OMIM:236600	MENDELIAN
CCDC88C	Spinocerebellar ataxia 40	OMIM:616053	MENDELIAN
CCDC88C	Spinocerebellar ataxia type 40	ORPHA:423275	UNKNOWN
CCDC89	—		UNKNOWN
CCDC90B	—		UNKNOWN
CCHCR1	—		UNKNOWN
CCK	—		UNKNOWN
CCKAR	—		UNKNOWN
CCKBR	—		UNKNOWN
CCL11	Asthma, susceptibility to	OMIM:600807	POLYGENIC
CCL13	—		UNKNOWN
CCL17	—		UNKNOWN
CCL22	—		UNKNOWN
CCL26	—		UNKNOWN
CCL4L1	—		UNKNOWN
CCL7	—		UNKNOWN
CCM2	Cerebral cavernous malformations 2	OMIM:603284	MENDELIAN
CCM2	Cerebral cavernous malformations 1	OMIM:116860	MENDELIAN
CCM2	Familial cerebral cavernous malformation	ORPHA:221061	UNKNOWN
CCNA1	—		UNKNOWN
CCNA2	—		UNKNOWN
CCNB1IP1	—		UNKNOWN
CCNB3	—		UNKNOWN
CCND1	Multiple myeloma	OMIM:254500	POLYGENIC

Gene_symbol	Disease_name	Disease_ID	Association_type
CCND1	Colorectal cancer	OMIM:114500	POLYGENIC
CCND1	von Hippel-Lindau syndrome	OMIM:193300	POLYGENIC
CCND1	Mantle cell lymphoma	ORPHA:52416	UNKNOWN
CCND1	Von Hippel-Lindau disease	ORPHA:892	UNKNOWN
CCND1	Multiple myeloma	ORPHA:29073	UNKNOWN
CCND2	Megalencephaly-Polymicrogyria-Polydactyly-Hydrocephalus syndrome 3	OMIM:615938	MENDELIAN
CCND2	Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome	ORPHA:83473	UNKNOWN
CCND3	—		UNKNOWN
CCNE1	—		UNKNOWN
CCNH	—		UNKNOWN
CCNK	Intellectual developmental disorder with hypertelorism and distinctive facies	OMIM:618147	MENDELIAN
CCNO	Ciliary dyskinesia, primary, 29	OMIM:615872	MENDELIAN
CCNO	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
CCNT2	—		UNKNOWN
CCR1	Behçet disease	ORPHA:117	UNKNOWN
CCR2	Cystic disease of lung	OMIM:219600	MENDELIAN
CCR3	—		UNKNOWN
CCR6	Limited cutaneous systemic sclerosis	ORPHA:220402	UNKNOWN
CCR6	Diffuse cutaneous systemic sclerosis	ORPHA:220393	UNKNOWN
CCR7	—		UNKNOWN
CCRL2	—		UNKNOWN
CCSER1	—		UNKNOWN
CCT5	Neuropathy, hereditary sensory, with spastic paraplegia, autosomal recessive	OMIM:256840	MENDELIAN
CCT5	Mutilating hereditary sensory neuropathy with spastic paraplegia	ORPHA:139578	UNKNOWN
CCT7	—		UNKNOWN
CD109	Fetal and neonatal alloimmune thrombocytopenia	ORPHA:853	UNKNOWN
CD14	—		UNKNOWN
CD151	Nephropathy with pretibial epidermolysis bullosa and deafness	OMIM:609057	MENDELIAN
CD177	—		UNKNOWN
CD19	Immunodeficiency, common variable, 3	OMIM:613493	MENDELIAN
CD19	Immunodeficiency, common variable, 2	OMIM:240500	MENDELIAN
CD1A	—		UNKNOWN
CD1E	—		UNKNOWN
CD200	—		UNKNOWN
CD22	—		UNKNOWN
CD226	—		UNKNOWN
CD24	—		UNKNOWN
CD244	Rheumatoid arthritis	OMIM:180300	POLYGENIC
CD247	Immunodeficiency 25	OMIM:610163	MENDELIAN
CD247	Oligoarticular juvenile idiopathic arthritis	ORPHA:85410	UNKNOWN
CD247	Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	ORPHA:85408	UNKNOWN
CD247	T-B+NK+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta deficiency	ORPHA:169160	UNKNOWN
CD27	Lymphoproliferative syndrome 2	OMIM:615122	MENDELIAN
CD274	Autoimmune disease, multisystem, infantile-onset, 5	OMIM:621235	MENDELIAN
CD276	—		UNKNOWN
CD2AP	Focal segmental glomerulosclerosis 3, susceptibility to	OMIM:607832	MENDELIAN
CD2AP	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect	OMIM:613646	MENDELIAN
CD36	Platelet glycoprotein IV deficiency	OMIM:608404	MENDELIAN
CD38	—		UNKNOWN
CD3D	Immunodeficiency 19	OMIM:615617	MENDELIAN
CD3D	T-B+NK+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta deficiency	ORPHA:169160	UNKNOWN
CD3E	Immunodeficiency 18	OMIM:615615	MENDELIAN
CD3E	T-B+NK+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta deficiency	ORPHA:169160	UNKNOWN
CD3EAP	—		UNKNOWN
CD3G	Immunodeficiency 17	OMIM:615607	MENDELIAN
CD4	Immunodeficiency 79	OMIM:619238	MENDELIAN
CD4	Okt4 epitope deficiency	OMIM:613949	MENDELIAN
CD4	Whipple disease	ORPHA:3452	UNKNOWN
CD40	Immunodeficiency with hyper-igm, type 3	OMIM:606843	MENDELIAN
CD40LG	Immunodeficiency, X-linked, with hyper-IgM	OMIM:308230	MENDELIAN
CD46	Hemolytic uremic syndrome, atypical, susceptibility to, 2	OMIM:612922	POLYGENIC
CD46	HELLP syndrome	ORPHA:244242	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
CD5	—		UNKNOWN
CD55	Blood group, cromer system	OMIM:613793	MENDELIAN
CD55	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy	OMIM:226300	MENDELIAN
CD58	—		UNKNOWN
CD59	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	OMIM:612300	MENDELIAN
CD72	—		UNKNOWN
CD74	—		UNKNOWN
CD79A	Agammaglobulinemia 3, autosomal recessive	OMIM:613501	MENDELIAN
CD79A	Autosomal non-syndromic agammaglobulinemia	ORPHA:33110	UNKNOWN
CD79B	Agammaglobulinemia 6, autosomal recessive	OMIM:612692	MENDELIAN
CD79B	Autosomal non-syndromic agammaglobulinemia	ORPHA:33110	UNKNOWN
CD80	—		UNKNOWN
CD81	Immunodeficiency, common variable, 6	OMIM:613496	MENDELIAN
CD82	—		UNKNOWN
CD86	—		UNKNOWN
CD8A	Cd8 deficiency, familial	OMIM:608957	MENDELIAN
CD96	C syndrome	OMIM:211750	MENDELIAN
CD96	C syndrome	ORPHA:1308	UNKNOWN
CDA	—		UNKNOWN
CDAN1	Anemia, congenital dyserythropoietic, type Ia	OMIM:224120	MENDELIAN
CDC25A	—		UNKNOWN
CDC25C	—		UNKNOWN
CDC27	—		UNKNOWN
CDC42	Takenouchi-Kosaki syndrome	OMIM:616737	MENDELIAN
CDC42	Takenouchi-Kosaki syndrome	ORPHA:487796	UNKNOWN
CDC42BPB	Chilton-Okur-Chung neurodevelopmental syndrome	OMIM:619841	MENDELIAN
CDC45	Meier-Gorlin syndrome 7	OMIM:617063	MENDELIAN
CDC45	Ear-patella-short stature syndrome	ORPHA:2554	UNKNOWN
CDC5L	—		UNKNOWN
CDC6	Meier-Gorlin syndrome 5	OMIM:613805	MENDELIAN
CDC6	Ear-patella-short stature syndrome	ORPHA:2554	UNKNOWN
CDC73	Hyperparathyroidism 1	OMIM:145000	MENDELIAN
CDC73	Hyperparathyroidism 2	OMIM:145001	MENDELIAN
CDC73	Parathyroid carcinoma	OMIM:608266	MENDELIAN
CDC73	Hyperparathyroidism-jaw tumor syndrome	ORPHA:99880	UNKNOWN
CDC73	Familial isolated hyperparathyroidism	ORPHA:99879	UNKNOWN
CDC73	Parathyroid carcinoma	ORPHA:143	UNKNOWN
CDCA7L	—		UNKNOWN
CDH1	Endometrial carcinoma, somatic	OMIM:608089	MENDELIAN
CDH1	Breast cancer	OMIM:114480	MENDELIAN
CDH1	Ovarian cancer	OMIM:167000	MENDELIAN
CDH1	Gastric cancer, hereditary diffuse	OMIM:137215	MENDELIAN
CDH1	Blepharocheilodontic syndrome 1	OMIM:119580	MENDELIAN
CDH1	Cleft lip/palate	ORPHA:199306	UNKNOWN
CDH1	Blepharo-cheilo-odontic syndrome	ORPHA:1997	UNKNOWN
CDH12	—		UNKNOWN
CDH13	—		UNKNOWN
CDH15	Intellectual developmental disorder, autosomal dominant 3	OMIM:612580	MENDELIAN
CDH18	—		UNKNOWN
CDH19	—		UNKNOWN
CDH2	Arrhythmogenic right ventricular dysplasia, familial, 14	OMIM:618920	MENDELIAN
CDH2	Agenesis of corpus callosum, cardiac, ocular, and genital syndrome	OMIM:618929	MENDELIAN
CDH2	Attention deficit-hyperactivity disorder 8	OMIM:619957	MENDELIAN
CDH20	—		UNKNOWN
CDH23	Pituitary adenoma 5, multiple types	OMIM:617540	POLYGENIC
CDH23	Usher syndrome, type ID	OMIM:601067	MENDELIAN
CDH23	Deafness, autosomal recessive 12	OMIM:601386	MENDELIAN
CDH23	Cushing disease	ORPHA:96253	UNKNOWN
CDH23	TSH-secreting pituitary adenoma	ORPHA:91347	UNKNOWN
CDH23	Prolactinoma	ORPHA:2965	UNKNOWN
CDH23	Usher syndrome type 1	ORPHA:231169	UNKNOWN
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy syndrome	OMIM:225280	MENDELIAN
CDH3	Hypotrichosis, congenital, with juvenile macular dystrophy	OMIM:601553	MENDELIAN
CDH3	Hypotrichosis with juvenile macular degeneration	ORPHA:1573	UNKNOWN
CDH3	EEM syndrome	ORPHA:1897	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
CDH5	—		UNKNOWN
CDH8	—		UNKNOWN
CDH9	—		UNKNOWN
CDHR1	Cone-rod dystrophy 15	OMIM:613660	MENDELIAN
CDHR1	Cone rod dystrophy	ORPHA:1872	UNKNOWN
CDHR1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
CDK11A	—		UNKNOWN
CDK12	—		UNKNOWN
CDK13	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder	OMIM:617360	MENDELIAN
CDK16	—		UNKNOWN
CDK19	Epileptic encephalopathy, early infantile, 87	OMIM:618916	MENDELIAN
CDK19	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
CDK2	—		UNKNOWN
CDK4	Melanoma, cutaneous malignant, susceptibility to, 3	OMIM:609048	POLYGENIC
CDK4	Microcephaly 31, primary, autosomal recessive	OMIM:621507	MENDELIAN
CDK4	Well-differentiated liposarcoma	ORPHA:99971	UNKNOWN
CDK4	Familial melanoma	ORPHA:618	UNKNOWN
CDK5	Lissencephaly 7 with cerebellar hypoplasia	OMIM:616342	MENDELIAN
CDK5R1	—		UNKNOWN
CDK5RAP2	Microcephaly 3, primary, autosomal recessive	OMIM:604804	MENDELIAN
CDK5RAP2	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
CDK5RAP3	—		UNKNOWN
CDK6	Microcephaly 12, primary, autosomal recessive	OMIM:616080	MENDELIAN
CDK6	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
CDK7	—		UNKNOWN
CDK8	Intellectual developmental disorder with hypotonia and behavioral abnormalities	OMIM:618748	MENDELIAN
CDKAL1	—		UNKNOWN
CDKL3	—		UNKNOWN
CDKL5	Developmental and epileptic encephalopathy 2	OMIM:300672	MENDELIAN
CDKL5	CDKL5-deficiency disorder	ORPHA:505652	UNKNOWN
CDKL5	Atypical Rett syndrome	ORPHA:3095	UNKNOWN
CDKL5	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
CDKL5	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
CDKN1A	—		UNKNOWN
CDKN1B	Multiple endocrine neoplasia, type IV	OMIM:610755	MENDELIAN
CDKN1B	Multiple endocrine neoplasia type 4	ORPHA:276152	UNKNOWN
CDKN1B	Multiple endocrine neoplasia type 1	ORPHA:652	UNKNOWN
CDKN1C	Beckwith-Wiedemann syndrome	OMIM:130650	MENDELIAN
CDKN1C	Intrauterine growth retardation, metaphyseal dysplasia, adrenal hypoplasiacongenita, and genital anomalies	OMIM:614732	MENDELIAN
CDKN1C	Silver-Russell syndrome due to a point mutation	ORPHA:397590	UNKNOWN
CDKN1C	Intrauterine growth restriction-short stature-early adult-onset diabetes syndrome	ORPHA:436144	UNKNOWN
CDKN1C	IMAGe syndrome	ORPHA:85173	UNKNOWN
CDKN2A	Melanoma-Astrocytoma syndrome	OMIM:155755	POLYGENIC
CDKN2A	Melanoma-Pancreatic cancer syndrome	OMIM:606719	POLYGENIC
CDKN2A	Melanoma, cutaneous malignant, susceptibility to, 2	OMIM:155601	POLYGENIC
CDKN2A	Adrenocortical carcinoma	ORPHA:1501	UNKNOWN
CDKN2A	Familial pancreatic carcinoma	ORPHA:1333	UNKNOWN
CDKN2A	Familial melanoma	ORPHA:618	UNKNOWN
CDKN2A	Li-Fraumeni syndrome	ORPHA:524	UNKNOWN
CDKN2AIP	—		UNKNOWN
CDKN2B	Familial melanoma	ORPHA:618	UNKNOWN
CDKN2B	Multiple endocrine neoplasia type 1	ORPHA:652	UNKNOWN
CDKN2B-AS1	—		UNKNOWN
CDKN2C	—		UNKNOWN
CDON	Holoprosencephaly 11	OMIM:614226	MENDELIAN
CDON	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
CDON	Pituitary stalk interruption syndrome	ORPHA:95496	UNKNOWN
CDON	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
CDON	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
CDON	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
CDON	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
CDON	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
CDSN	Hypotrichosis 2	OMIM:146520	MENDELIAN
CDSN	Peeling skin syndrome 1	OMIM:270300	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
CDSN	Hypotrichosis simplex of the scalp	ORPHA:90368	UNKNOWN
CDT1	Meier-Gorlin syndrome 4	OMIM:613804	MENDELIAN
CDT1	Ear-patella-short stature syndrome	ORPHA:2554	UNKNOWN
CDX1	—		UNKNOWN
CDX2	—		UNKNOWN
CDY1	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
CDY2A	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
CDYL	—		UNKNOWN
CEACAM16	Deafness, autosomal dominant 4B	OMIM:614614	MENDELIAN
CEACAM16	Deafness, autosomal recessive 113	OMIM:618410	MENDELIAN
CEBPA	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
CEBPE	Immunodeficiency 108 with autoinflammation	OMIM:260570	MENDELIAN
CEBPE	Specific granule deficiency	OMIM:245480	MENDELIAN
CECR1	—		UNKNOWN
CECR2	—		UNKNOWN
CECR5	—		UNKNOWN
CEL	Maturity-Onset diabetes of the young, type 8, with exocrine dysfunction	OMIM:609812	MENDELIAN
CEL	MODY	ORPHA:552	UNKNOWN
CELF2	Developmental and epileptic encephalopathy 97	OMIM:619561	MENDELIAN
CELF2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
CELF4	—		UNKNOWN
CELF6	—		UNKNOWN
CELSR1	Yellow nail syndrome	OMIM:153300	MENDELIAN
CELSR1	Lymphatic malformation 9	OMIM:619319	MENDELIAN
CELSR2	—		UNKNOWN
CELSR3	—		UNKNOWN
CENPA	—		UNKNOWN
CENPE	Microcephaly 13, primary, autosomal recessive	OMIM:616051	MENDELIAN
CENPE	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
CENPE	Seckel syndrome	ORPHA:808	UNKNOWN
CENPJ	—		UNKNOWN
CEP120	Joubert syndrome 31	OMIM:617761	MENDELIAN
CEP120	Short-Rib thoracic dysplasia 13 with or without polydactyly	OMIM:616300	MENDELIAN
CEP120	Joubert syndrome with ocular anomaly	ORPHA:220493	UNKNOWN
CEP120	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
CEP120	Jeune syndrome	ORPHA:474	UNKNOWN
CEP135	Microcephaly 8, primary, autosomal recessive	OMIM:614673	MENDELIAN
CEP135	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
CEP152	Seckel syndrome 5	OMIM:613823	MENDELIAN
CEP152	Microcephaly 9, primary, autosomal recessive	OMIM:614852	MENDELIAN
CEP152	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
CEP152	Seckel syndrome	ORPHA:808	UNKNOWN
CEP164	Nephronophthisis 15	OMIM:614845	MENDELIAN
CEP164	Senior-Loken syndrome	ORPHA:3156	UNKNOWN
CEP170B	—		UNKNOWN
CEP19	Morbid obesity and spermatogenic failure	OMIM:615703	MENDELIAN
CEP19	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
CEP250	Cone-rod dystrophy and hearing loss 2	OMIM:618358	MENDELIAN
CEP290	Leber congenital amaurosis 10	OMIM:611755	MENDELIAN
CEP290	Joubert syndrome 5	OMIM:610188	MENDELIAN
CEP290	Senior-Loken syndrome 6	OMIM:610189	MENDELIAN
CEP290	Bardet-Biedl syndrome 14	OMIM:615991	MENDELIAN
CEP290	Meckel syndrome, type 4	OMIM:611134	MENDELIAN
CEP290	Senior-Loken syndrome	ORPHA:3156	UNKNOWN
CEP290	Leber congenital amaurosis	ORPHA:65	UNKNOWN
CEP290	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
CEP290	Joubert syndrome with oculorenal defect	ORPHA:2318	UNKNOWN
CEP290	Meckel syndrome	ORPHA:564	UNKNOWN
CEP41	Joubert syndrome 15	OMIM:614464	MENDELIAN
CEP41	Joubert syndrome with ocular anomaly	ORPHA:220493	UNKNOWN
CEP41	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
CEP57	Mosaic variegated aneuploidy syndrome 2	OMIM:614114	MENDELIAN
CEP57	Mosaic variegated aneuploidy syndrome	ORPHA:1052	UNKNOWN
CEP63	Seckel syndrome 6	OMIM:614728	MENDELIAN
CEP63	Seckel syndrome	ORPHA:808	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
CEP68	—		UNKNOWN
CEP83	Nephronophthisis 18	OMIM:615862	MENDELIAN
CEP85	—		UNKNOWN
CEP85L	Lissencephaly 10	OMIM:618873	MENDELIAN
CEP85L	Posterior-predominant lissencephaly-broad flat pons and medulla-midline crossing defects syndrome	ORPHA:572013	UNKNOWN
CEP89	—		UNKNOWN
CER1	—		UNKNOWN
CERKL	Retinitis pigmentosa 26	OMIM:608380	MENDELIAN
CERKL	Retinitis pigmentosa	ORPHA:791	UNKNOWN
CERS1	Epilepsy, progressive myoclonic, 8	OMIM:616230	MENDELIAN
CERS3	Ichthyosis, congenital, autosomal recessive 9	OMIM:615023	MENDELIAN
CERS3	Congenital ichthyosiform erythroderma	ORPHA:79394	UNKNOWN
CERS6	—		UNKNOWN
CERT1	Neurodevelopmental disorder with hypotonia, speech delay, and dysmorphic facies	OMIM:616351	MENDELIAN
CES1	Drug metabolism, altered, CES1-related	OMIM:618057	MENDELIAN
CES2	—		UNKNOWN
CETN2	—		UNKNOWN
CETP	Hyperalphalipoproteinemia 1	OMIM:143470	MENDELIAN
CFAP43	Spermatogenic failure 19	OMIM:617592	MENDELIAN
CFAP43	Hydrocephalus, normal pressure, 1	OMIM:236690	MENDELIAN
CFAP44	Spermatogenic failure 20	OMIM:617593	MENDELIAN
CFAP69	Spermatogenic failure 24	OMIM:617959	MENDELIAN
CFB	Hemolytic uremic syndrome, atypical, susceptibility to, 4	OMIM:612924	POLYGENIC
CFB	Complement factor B deficiency	OMIM:615561	MENDELIAN
CFC1	Heterotaxy, visceral, 2, autosomal	OMIM:605376	MENDELIAN
CFD	Complement factor D deficiency	OMIM:613912	MENDELIAN
CFH	Complement factor H deficiency	OMIM:609814	MENDELIAN
CFH	Macular degeneration, age-related, 4	OMIM:610698	POLYGENIC
CFH	Hemolytic uremic syndrome, atypical, susceptibility to, 1	OMIM:235400	POLYGENIC
CFH	Basal laminar drusen	OMIM:126700	MENDELIAN
CFH	HELLP syndrome	ORPHA:244242	UNKNOWN
CFH	Autosomal dominant drusen	ORPHA:75376	UNKNOWN
CFHR1	Macular degeneration, age-related, 1	OMIM:603075	POLYGENIC
CFHR1	Hemolytic uremic syndrome, atypical, susceptibility to, 1	OMIM:235400	POLYGENIC
CFHR3	Macular degeneration, age-related, 1	OMIM:603075	POLYGENIC
CFHR3	Hemolytic uremic syndrome, atypical, susceptibility to, 1	OMIM:235400	POLYGENIC
CFHR4	—		UNKNOWN
CFHR5	Nephropathy due to CFHR5 deficiency	OMIM:614809	MENDELIAN
CFI	Hemolytic uremic syndrome, atypical, susceptibility to, 3	OMIM:612923	POLYGENIC
CFI	Macular degeneration, age-related, 13	OMIM:615439	POLYGENIC
CFI	Complement factor I deficiency	OMIM:610984	MENDELIAN
CFI	HELLP syndrome	ORPHA:244242	UNKNOWN
CFI	Autosomal dominant drusen	ORPHA:75376	UNKNOWN
CFL1	—		UNKNOWN
CFL2	Nemaline myopathy 7, autosomal recessive	OMIM:610687	MENDELIAN
CFL2	Typical nemaline myopathy	ORPHA:171436	UNKNOWN
CFLAR	—		UNKNOWN
CFP	Properdin deficiency, X-linked	OMIM:312060	MENDELIAN
CFTR	Pancreatitis, hereditary	OMIM:167800	POLYGENIC
CFTR	Cystic fibrosis	OMIM:219700	MENDELIAN
CFTR	Vas deferens, congenital bilateral aplasia of	OMIM:277180	MENDELIAN
CFTR	Bronchiectasis with or without elevated sweat chloride 1	OMIM:211400	POLYGENIC
CFTR	Congenital bilateral absence of vas deferens	ORPHA:48	UNKNOWN
CFTR	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
CFTR	Idiopathic bronchiectasis	ORPHA:60033	UNKNOWN
CFTR	Aquagenic palmoplantar keratoderma	ORPHA:498359	UNKNOWN
CFTR	Cystic fibrosis	ORPHA:586	UNKNOWN
CGA	—		UNKNOWN
CGB	—		UNKNOWN
CGNL1	—		UNKNOWN
CHAF1B	—		UNKNOWN
CHAMP1	Intellectual developmental disorder, autosomal dominant 40	OMIM:616579	MENDELIAN
CHAT	Myasthenic syndrome, congenital, 6, presynaptic	OMIM:254210	MENDELIAN
CHCHD10	Spinal muscular atrophy, Jokela type	OMIM:615048	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
CHCHD10	Myopathy, isolated mitochondrial, autosomal dominant	OMIM:616209	MENDELIAN
CHCHD10	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2	OMIM:615911	MENDELIAN
CHCHD10	Frontotemporal dementia with motor neuron disease	ORPHA:275872	UNKNOWN
CHCHD10	Lower motor neuron syndrome with late-adult onset	ORPHA:276435	UNKNOWN
CHCHD10	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
CHCHD10	Autosomal dominant mitochondrial myopathy with exercise intolerance	ORPHA:457050	UNKNOWN
CHD1	Pilarowski-Bjornsson syndrome	OMIM:617682	MENDELIAN
CHD1	Intellectual disability-autism-speech apraxia-craniofacial dysmorphism syndrome	ORPHA:529965	UNKNOWN
CHD1L	—		UNKNOWN
CHD2	Develomental and epileptic encephalopathy 94	OMIM:615369	MENDELIAN
CHD2	Lennox-Gastaut syndrome	ORPHA:2382	UNKNOWN
CHD2	Epilepsy with myoclonic-atonic seizures	ORPHA:1942	UNKNOWN
CHD3	Snijders Blok-Campeau syndrome	OMIM:618205	MENDELIAN
CHD4	Sifrim-Hitz-Weiss syndrome	OMIM:617159	MENDELIAN
CHD6	Hallermann-Streiff syndrome	ORPHA:2108	UNKNOWN
CHD7	Charge syndrome	OMIM:214800	MENDELIAN
CHD7	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
CHD7	Hypogonadotropic hypogonadism 5 with or without anosmia	OMIM:612370	MENDELIAN
CHD7	CHARGE syndrome	ORPHA:138	UNKNOWN
CHD7	Omenn syndrome	ORPHA:39041	UNKNOWN
CHD7	Kallmann syndrome	ORPHA:478	UNKNOWN
CHD7	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
CHD8	Intellectual developmental disorder with autism and macrocephaly	OMIM:615032	MENDELIAN
CHD8	14q11.2 microduplication syndrome	ORPHA:261229	UNKNOWN
CHDH	—		UNKNOWN
CHEK1	Oocyte/zygote/embryo maturation arrest 21	OMIM:620610	MENDELIAN
CHEK1	Female infertility due to oocyte meiotic arrest	ORPHA:488191	UNKNOWN
CHEK2	Li-Fraumeni syndrome 2	OMIM:609265	MENDELIAN
CHEK2	Osteosarcoma	OMIM:259500	MENDELIAN
CHEK2	Prostate cancer	OMIM:176807	MENDELIAN
CHEK2	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
CHEK2	Osteosarcoma	ORPHA:668	UNKNOWN
CHEK2	Li-Fraumeni syndrome	ORPHA:524	UNKNOWN
CHEK2	Familial colorectal cancer Type X	ORPHA:440437	UNKNOWN
CHFR	—		UNKNOWN
CHGA	—		UNKNOWN
CHGB	—		UNKNOWN
CHI3L1	Schizophrenia	OMIM:181500	POLYGENIC
CHI3L2	—		UNKNOWN
CHIA	—		UNKNOWN
CHIC2	Leukemia, acute myeloid	OMIM:601626	POLYGENIC
CHIT1	Chitotriosidase deficiency	OMIM:614122	MENDELIAN
CHKB	Muscular dystrophy, congenital, megaconial type	OMIM:602541	MENDELIAN
CHL1	—		UNKNOWN
CHM	Choroideremia	OMIM:303100	MENDELIAN
CHM	Choroideremia	ORPHA:180	UNKNOWN
CHMP1A	Pontocerebellar hypoplasia, type 8	OMIM:614961	MENDELIAN
CHMP2B	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7	OMIM:600795	MENDELIAN
CHMP2B	Behavioral variant of frontotemporal dementia	ORPHA:275864	UNKNOWN
CHMP2B	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
CHMP2B	Progressive non-fluent aphasia	ORPHA:100070	UNKNOWN
CHMP2B	Semantic dementia	ORPHA:100069	UNKNOWN
CHMP4B	Cataract 31, multiple types	OMIM:605387	MENDELIAN
CHN1	Duane retraction syndrome 2	OMIM:604356	MENDELIAN
CHN1	Duane retraction syndrome	ORPHA:233	UNKNOWN
CHPF	—		UNKNOWN
CHPF2	—		UNKNOWN
CHRD	—		UNKNOWN
CHRD1	Megalocornea 1, X-linked	OMIM:309300	MENDELIAN
CHRFAM7A	—		UNKNOWN
CHRM1	—		UNKNOWN
CHRM2	—		UNKNOWN
CHRM3	Prune belly syndrome	OMIM:100100	MENDELIAN
CHRM3	Prune belly syndrome	ORPHA:2970	UNKNOWN
CHRNA1	Myasthenic syndrome, congenital, 1B, fast-channel	OMIM:608930	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
CHRNA1	Myasthenic syndrome, congenital, 1A, slow-channel	OMIM:601462	MENDELIAN
CHRNA1	Multiple pterygium syndrome, Lethal type	OMIM:253290	MENDELIAN
CHRNA2	Epilepsy, nocturnal frontal lobe, 4	OMIM:610353	MENDELIAN
CHRNA2	Sleep-related hypermotor epilepsy	ORPHA:98784	UNKNOWN
CHRNA3	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT	OMIM:191800	MENDELIAN
CHRNA4	Epilepsy, nocturnal frontal lobe, type 1	OMIM:600513	MENDELIAN
CHRNA4	Sleep-related hypermotor epilepsy	ORPHA:98784	UNKNOWN
CHRNA7	Chromosome 15q13.3 microdeletion syndrome	OMIM:612001	MENDELIAN
CHRNA7	15q13.3 microdeletion syndrome	ORPHA:199318	UNKNOWN
CHRNA9	—		UNKNOWN
CHRNB1	Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	OMIM:616314	MENDELIAN
CHRNB1	Myasthenic syndrome, congenital, 2A, slow-channel	OMIM:616313	MENDELIAN
CHRNB2	Epilepsy, nocturnal frontal lobe, 3	OMIM:605375	MENDELIAN
CHRNB2	Sleep-related hypermotor epilepsy	ORPHA:98784	UNKNOWN
CHRNB3	—		UNKNOWN
CHRNB4	—		UNKNOWN
CHRND	Myasthenic syndrome, congenital, 3B, fast-channel	OMIM:616322	MENDELIAN
CHRND	Myasthenic syndrome, congenital, 3A, slow-channel	OMIM:616321	MENDELIAN
CHRND	Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency	OMIM:616323	MENDELIAN
CHRND	Multiple pterygium syndrome, Lethal type	OMIM:253290	MENDELIAN
CHRNE	Myasthenic syndrome, congenital, 4A, slow-channel	OMIM:605809	MENDELIAN
CHRNE	Myasthenic syndrome, congenital, 4B, fast-channel	OMIM:616324	MENDELIAN
CHRNE	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	OMIM:608931	MENDELIAN
CHRNG	Escobar syndrome	OMIM:265000	MENDELIAN
CHRNG	Multiple pterygium syndrome, Lethal type	OMIM:253290	MENDELIAN
CHRNG	Autosomal recessive multiple pterygium syndrome	ORPHA:2990	UNKNOWN
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1	OMIM:601776	MENDELIAN
CHST14	Musculocontractural Ehlers-Danlos syndrome	ORPHA:2953	UNKNOWN
CHST3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects	OMIM:245600	MENDELIAN
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations	OMIM:143095	MENDELIAN
CHST3	CHST3-related skeletal dysplasia	ORPHA:263463	UNKNOWN
CHST6	Macular dystrophy, corneal, 1	OMIM:217800	MENDELIAN
CHST6	Macular corneal dystrophy	ORPHA:98969	UNKNOWN
CHST7	—		UNKNOWN
CHSY1	Temtamy preaxial brachydactyly syndrome	OMIM:605282	MENDELIAN
CHSY1	Temtamy preaxial brachydactyly syndrome	ORPHA:363417	UNKNOWN
CHSY3	—		UNKNOWN
CHUK	Popliteal pterygium syndrome, Bartsocas-Papas type 2	OMIM:619339	MENDELIAN
CHUK	Fetal encasement syndrome	OMIM:613630	MENDELIAN
CIB2	Deafness, autosomal recessive 48	OMIM:609439	MENDELIAN
CIC	Intellectual developmental disorder, autosomal dominant 45	OMIM:617600	MENDELIAN
CIDEA	—		UNKNOWN
CIDEC	Lipodystrophy, familial partial, type 5	OMIM:615238	MENDELIAN
CIDEC	CIDEC-related familial partial lipodystrophy	ORPHA:435651	UNKNOWN
CIITA	Rheumatoid arthritis	OMIM:180300	POLYGENIC
CIITA	Bare lymphocyte syndrome, type II	OMIM:209920	MENDELIAN
CIITA	Immunodeficiency by defective expression of MHC class II	ORPHA:572	UNKNOWN
CIRH1A	—		UNKNOWN
CISD2	Wolfram syndrome 2	OMIM:604928	MENDELIAN
CISD2	Wolfram syndrome	ORPHA:3463	UNKNOWN
CITED2	Atrial septal defect 8	OMIM:614433	MENDELIAN
CITED2	Ventricular septal defect 2	OMIM:614431	MENDELIAN
CITED2	Atrial septal defect, sinus venosus type	ORPHA:99105	UNKNOWN
CITED2	Atrial septal defect, ostium secundum type	ORPHA:99103	UNKNOWN
CITED2	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
CIZ1	Adult-onset cervical dystonia, DYT23 type	ORPHA:420492	UNKNOWN
CKAP2L	Filippi syndrome	OMIM:272440	MENDELIAN
CKAP2L	Filippi syndrome	ORPHA:3255	UNKNOWN
CKM	—		UNKNOWN
CLASP2	—		UNKNOWN
CLCA1	—		UNKNOWN
CLCA2	—		UNKNOWN
CLCF1	Cold-Induced sweating syndrome 2	OMIM:610313	MENDELIAN
CLCF1	Crisponi syndrome	ORPHA:1545	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
CLCN1	Myopathy, congenital	OMIM:255300	MENDELIAN
CLCN1	Myotonia congenita, autosomal recessive	OMIM:255700	MENDELIAN
CLCN1	Myotonia congenita, autosomal dominant	OMIM:160800	MENDELIAN
CLCN1	Thomsen and Becker disease	ORPHA:614	UNKNOWN
CLCN2	Hyperaldosteronism, familial, type II	OMIM:605635	MENDELIAN
CLCN2	Leukoencephalopathy with ataxia	OMIM:615651	MENDELIAN
CLCN2	Epilepsy with grand mal seizures on awakening	OMIM:607628	POLYGENIC
CLCN2	Juvenile myoclonic epilepsy	ORPHA:307	UNKNOWN
CLCN2	Familial hyperaldosteronism type II	ORPHA:404	UNKNOWN
CLCN3	Neurodevelopmental disorder with hypotonia and brain abnormalities	OMIM:619512	MENDELIAN
CLCN3	Neurodevelopmental disorder with seizures and brain abnormalities	OMIM:619517	MENDELIAN
CLCN4	Raynaud-Claes syndrome	OMIM:300114	MENDELIAN
CLCN4	CLCN4-related X-linked intellectual disability syndrome	ORPHA:485350	UNKNOWN
CLCN5	Nephrolithiasis, type I	OMIM:310468	MENDELIAN
CLCN5	Dent disease 1	OMIM:300009	MENDELIAN
CLCN5	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis	OMIM:308990	MENDELIAN
CLCN5	Hypophosphatemic rickets, X-linked recessive	OMIM:300554	MENDELIAN
CLCN6	Neurodegeneration, childhood-onset, hypotonia, respiratory insufficiency and brain imaging abnormalities	OMIM:619173	MENDELIAN
CLCN7	Osteopetrosis, autosomal recessive 4	OMIM:611490	MENDELIAN
CLCN7	Osteopetrosis, autosomal dominant 2	OMIM:166600	MENDELIAN
CLCN7	Hypopigmentation, organomegaly, and delayed myelination and development	OMIM:618541	MENDELIAN
CLCN7	Albers-Schönberg osteopetrosis	ORPHA:53	UNKNOWN
CLCN7	Intermediate osteopetrosis	ORPHA:210110	UNKNOWN
CLCN7	Autosomal recessive malignant osteopetrosis	ORPHA:667	UNKNOWN
CLCNKA	Bartter syndrome, type 4B, neonatal, with sensorineural deafness	OMIM:613090	MENDELIAN
CLCNKA	Bartter syndrome type 4	ORPHA:89938	UNKNOWN
CLCNKB	Bartter syndrome, type 4B, neonatal, with sensorineural deafness	OMIM:613090	MENDELIAN
CLCNKB	Bartter syndrome, type 3	OMIM:607364	MENDELIAN
CLCNKB	Gitelman syndrome	ORPHA:358	UNKNOWN
CLCNKB	Bartter syndrome type 4	ORPHA:89938	UNKNOWN
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	OMIM:607626	MENDELIAN
CLDN1	Neonatal ichthyosis-sclerosing cholangitis syndrome	ORPHA:59303	UNKNOWN
CLDN14	Deafness, autosomal recessive 29	OMIM:614035	MENDELIAN
CLDN16	Hypomagnesemia 3, renal	OMIM:248250	MENDELIAN
CLDN19	Hypomagnesemia 5, renal, with or without ocular involvement	OMIM:248190	MENDELIAN
CLDN19	Primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement	ORPHA:2196	UNKNOWN
CLDN23	—		UNKNOWN
CLEC10A	—		UNKNOWN
CLEC11A	—		UNKNOWN
CLEC2D	—		UNKNOWN
CLEC3B	Macular dystrophy, retinal, 4	OMIM:619977	MENDELIAN
CLEC4D	—		UNKNOWN
CLEC4E	—		UNKNOWN
CLEC4M	—		UNKNOWN
CLEC7A	Candidiasis, familial, 4	OMIM:613108	MENDELIAN
CLEC7A	Chronic mucocutaneous candidiasis	ORPHA:1334	UNKNOWN
CLHC1	—		UNKNOWN
CLIC2	X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome	ORPHA:324410	UNKNOWN
CLIC5	Deafness, autosomal recessive 103	OMIM:616042	MENDELIAN
CLIP2	Williams syndrome	ORPHA:904	UNKNOWN
CLK1	—		UNKNOWN
CLK2	—		UNKNOWN
CLMP	Congenital short bowel syndrome	OMIM:615237	MENDELIAN
CLMP	Congenital short bowel syndrome	ORPHA:2301	UNKNOWN
CLN3	Retinitis pigmentosa 101	OMIM:621548	MENDELIAN
CLN3	Ceroid lipofuscinosis, neuronal, 3	OMIM:204200	MENDELIAN
CLN5	Ceroid lipofuscinosis, neuronal, 5	OMIM:256731	MENDELIAN
CLN6	Ceroid lipofuscinosis, neuronal, 6B (Kufs type)	OMIM:204300	MENDELIAN
CLN6	Ceroid lipofuscinosis, neuronal, 6	OMIM:601780	MENDELIAN
CLN8	Ceroid lipofuscinosis, neuronal, 8, northern epilepsy variant	OMIM:610003	MENDELIAN
CLN8	Ceroid lipofuscinosis, neuronal, 8	OMIM:600143	MENDELIAN
CLN8	Northern epilepsy	ORPHA:1947	UNKNOWN
CLNK	—		UNKNOWN
CLOCK	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
CLP1	Pontocerebellar hypoplasia, type 10	OMIM:615803	MENDELIAN
CLP1	Pontocerebellar hypoplasia type 10	ORPHA:411493	UNKNOWN
CLPB	3-Methylglutaconic aciduria with cataracts, neurologic involvement, and neutropenia	OMIM:616271	MENDELIAN
CLPB	3-methylglutaconic aciduria, type VIIA, autosomal dominant	OMIM:619835	MENDELIAN
CLPB	Neutropenia, severe congenital, 9, autosomal dominant	OMIM:619813	MENDELIAN
CLPB	3-methylglutaconic aciduria-neonatal cataract-neurologic involvement-congenital neutropenia syndrome	ORPHA:445038	UNKNOWN
CLPB	Autosomal dominant severe congenital neutropenia	ORPHA:486	UNKNOWN
CLPP	Perrault syndrome 3	OMIM:614129	MENDELIAN
CLPS	—		UNKNOWN
CLPTM1	—		UNKNOWN
CLPTM1L	—		UNKNOWN
CLRN1	Usher syndrome, type IIIA	OMIM:276902	MENDELIAN
CLRN1	Retinitis pigmentosa 61	OMIM:614180	MENDELIAN
CLRN1	Retinitis pigmentosa	OMIM:268000	MENDELIAN
CLRN1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
CLRN1	Usher syndrome type 3	ORPHA:231183	UNKNOWN
CLSTN1	—		UNKNOWN
CLSTN2	—		UNKNOWN
CLTCL1	Congenital insensitivity to pain with severe intellectual disability	ORPHA:453510	UNKNOWN
CLU	—		UNKNOWN
CLUL1	—		UNKNOWN
CLYBL	—		UNKNOWN
CMA1	—		UNKNOWN
CMC4	—		UNKNOWN
CMIP	—		UNKNOWN
CMPK1	—		UNKNOWN
CNBP	Dystrophia myotonica 2	OMIM:602668	MENDELIAN
CNBP	Proximal myotonic myopathy	ORPHA:606	UNKNOWN
CNDP1	—		UNKNOWN
CNDP2	—		UNKNOWN
CNGA1	Retinitis pigmentosa	OMIM:268000	MENDELIAN
CNGA1	Retinitis pigmentosa 49	OMIM:613756	MENDELIAN
CNGA1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
CNGA3	Achromatopsia 2	OMIM:216900	MENDELIAN
CNGA3	Cone rod dystrophy	ORPHA:1872	UNKNOWN
CNGA3	Achromatopsia	ORPHA:49382	UNKNOWN
CNGB1	Retinitis pigmentosa 45	OMIM:613767	MENDELIAN
CNGB1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
CNGB3	Achromatopsia 3	OMIM:262300	MENDELIAN
CNGB3	Progressive cone dystrophy	ORPHA:1871	UNKNOWN
CNGB3	Stargardt disease	ORPHA:827	UNKNOWN
CNGB3	Achromatopsia	ORPHA:49382	UNKNOWN
CNKSR1	—		UNKNOWN
CNKSR2	Intellectual developmental disorder, X-linked syndromic, Houge type	OMIM:301008	MENDELIAN
CNKSR2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
CNN2	—		UNKNOWN
CNNM2	Hypomagnesemia, seizures, and impaired intellectual development 1	OMIM:616418	MENDELIAN
CNNM2	Hypomagnesemia 6, renal	OMIM:613882	MENDELIAN
CNNM4	Jalili syndrome	OMIM:217080	MENDELIAN
CNNM4	Jalili syndrome	ORPHA:1873	UNKNOWN
CNOT1	Pancreatic agenesis-holoprosencephaly syndrome	ORPHA:556955	UNKNOWN
CNOT3	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies	OMIM:618672	MENDELIAN
CNOT4	—		UNKNOWN
CNPY3	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
CNR1	—		UNKNOWN
CNR2	—		UNKNOWN
CNTF	—		UNKNOWN
CNTLN	—		UNKNOWN
CNTN1	Myopathy, congenital, compton-north	OMIM:612540	MENDELIAN
CNTN2	Epilepsy, familial adult myoclonic, 5	OMIM:615400	MENDELIAN
CNTN2	Familial adult myoclonic epilepsy	ORPHA:86814	UNKNOWN
CNTN4	—		UNKNOWN
CNTN6	—		UNKNOWN
CNTNAP1	Lethal congenital contracture syndrome 7	OMIM:616286	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
CNTNAP1	Neuropathy, congenital hypomyelinating, 3	OMIM:618186	MENDELIAN
CNTNAP1	Hypomyelination neuropathy-arthrogryposis syndrome	ORPHA:2680	UNKNOWN
CNTNAP2	Pitt-Hopkins like syndrome 1	OMIM:610042	MENDELIAN
CNTNAP2	CNTNAP2-related developmental and epileptic encephalopathy	ORPHA:163681	UNKNOWN
CNTNAP4	—		UNKNOWN
CNTNAP5	—		UNKNOWN
COA1	—		UNKNOWN
COA5	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3	OMIM:616500	MENDELIAN
COA6	Mitochondrial complex IV deficiency, nuclear type 13	OMIM:616501	MENDELIAN
COASY	Neurodegeneration with brain iron accumulation 6	OMIM:615643	MENDELIAN
COASY	Pontocerebellar hypoplasia, type 12	OMIM:618266	MENDELIAN
COASY	COASY protein-associated neurodegeneration	ORPHA:397725	UNKNOWN
COCH	Deafness, autosomal dominant 9	OMIM:601369	MENDELIAN
COCH	Deafness, autosomal recessive 110	OMIM:618094	MENDELIAN
COG1	Congenital disorder of glycosylation, type IIg	OMIM:611209	MENDELIAN
COG1	COG1-CDG	ORPHA:263508	UNKNOWN
COG2	Congenital disorder of glycosylation, type IIq	OMIM:617395	MENDELIAN
COG2	COG2-CDG	ORPHA:435934	UNKNOWN
COG3	Congenital disorder of glycosylation, type IIbb	OMIM:620546	MENDELIAN
COG4	Congenital disorder of glycosylation, type IIj	OMIM:613489	MENDELIAN
COG4	Saul-Wilson syndrome	OMIM:618150	MENDELIAN
COG4	COG4-CDG	ORPHA:263501	UNKNOWN
COG4	Microcephalic osteodysplastic dysplasia, Saul-Wilson type	ORPHA:85172	UNKNOWN
COG5	Congenital disorder of glycosylation, type III	OMIM:613612	MENDELIAN
COG5	COG5-CDG	ORPHA:263487	UNKNOWN
COG6	Shaheen syndrome	OMIM:615328	MENDELIAN
COG6	Congenital disorder of glycosylation, type III	OMIM:614576	MENDELIAN
COG6	Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome	ORPHA:363523	UNKNOWN
COG7	Congenital disorder of glycosylation, type IIe	OMIM:608779	MENDELIAN
COG7	COG7-CDG	ORPHA:79333	UNKNOWN
COG8	Congenital disorder of glycosylation, type IIh	OMIM:611182	MENDELIAN
COG8	COG8-CDG	ORPHA:95428	UNKNOWN
COL10A1	Metaphyseal chondrodysplasia, Schmid type	OMIM:156500	MENDELIAN
COL10A1	Metaphyseal chondrodysplasia, Schmid type	ORPHA:174	UNKNOWN
COL11A1	Marshall syndrome	OMIM:154780	MENDELIAN
COL11A1	Stickler syndrome, type II	OMIM:604841	MENDELIAN
COL11A1	Fibrochondrogenesis 1	OMIM:228520	MENDELIAN
COL11A1	Deafness, autosomal dominant 37	OMIM:618533	MENDELIAN
COL11A1	Stickler syndrome type 2	ORPHA:90654	UNKNOWN
COL11A1	Fibrochondrogenesis	ORPHA:2021	UNKNOWN
COL11A1	Autosomal dominant myopia-midfacial retrusion-sensorineural hearing loss-rhizomelic dysplasia syndrome	ORPHA:440354	UNKNOWN
COL11A1	Marshall syndrome	ORPHA:560	UNKNOWN
COL11A2	Deafness, autosomal recessive 53	OMIM:609706	MENDELIAN
COL11A2	Stickler syndrome, type III	OMIM:184840	MENDELIAN
COL11A2	Fibrochondrogenesis 2	OMIM:614524	MENDELIAN
COL11A2	Otospondylomegaepiphyseal dysplasia	OMIM:215150	MENDELIAN
COL11A2	Deafness, autosomal dominant nonsyndromic sensorineural 13	OMIM:601868	MENDELIAN
COL11A2	Autosomal recessive otospondylomegaepiphyseal dysplasia	ORPHA:1427	UNKNOWN
COL11A2	Fibrochondrogenesis	ORPHA:2021	UNKNOWN
COL11A2	Autosomal dominant otospondylomegaepiphyseal dysplasia	ORPHA:166100	UNKNOWN
COL12A1	Bethlem myopathy 2	OMIM:616471	MENDELIAN
COL12A1	Ullrich congenital muscular dystrophy 2	OMIM:616470	MENDELIAN
COL12A1	Ullrich congenital muscular dystrophy	ORPHA:75840	UNKNOWN
COL12A1	Bethlem muscular dystrophy	ORPHA:610	UNKNOWN
COL12A1	Myopathic Ehlers-Danlos syndrome	ORPHA:536516	UNKNOWN
Col15A1	—		UNKNOWN
COL17A1	Epidermolysis bullosa, junctional, Non-Herlitz type	OMIM:226650	MENDELIAN
COL17A1	Epithelial recurrent erosion dystrophy	OMIM:122400	MENDELIAN
COL17A1	Epidermolysis bullosa, junctional 4, intermediate	OMIM:619787	MENDELIAN
COL17A1	Late-onset junctional epidermolysis bullosa	ORPHA:79406	UNKNOWN
COL17A1	Intermediate generalized junctional epidermolysis bullosa	ORPHA:79402	UNKNOWN
COL17A1	Localized junctional epidermolysis bullosa	ORPHA:251393	UNKNOWN
COL17A1	Epithelial recurrent erosion dystrophy	ORPHA:293381	UNKNOWN
COL18A1	Glaucoma, primary closed-angle	OMIM:618880	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
COL18A1	Knobloch syndrome, type 1	OMIM:267750	MENDELIAN
COL18A1	Knobloch syndrome	ORPHA:1571	UNKNOWN
COL1A1	Osteoporosis	OMIM:166710	POLYGENIC
COL1A1	Caffey disease	OMIM:114000	MENDELIAN
COL1A1	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1	OMIM:619115	MENDELIAN
COL1A1	Osteogenesis imperfecta, type III	OMIM:259420	MENDELIAN
COL1A1	Ehlers-Danlos syndrome, arthrochalasia type, 1	OMIM:130060	MENDELIAN
COL1A1	Osteogenesis imperfecta, type II	OMIM:166210	MENDELIAN
COL1A1	Osteogenesis imperfecta, type IV	OMIM:166220	MENDELIAN
COL1A1	Osteogenesis imperfecta, type I	OMIM:166200	MENDELIAN
COL1A1	Classical Ehlers-Danlos syndrome	ORPHA:287	UNKNOWN
COL1A1	Caffey disease	ORPHA:1310	UNKNOWN
COL1A1	Arthrochalasia Ehlers-Danlos syndrome	ORPHA:1899	UNKNOWN
COL1A1	Dermatofibrosarcoma protuberans	ORPHA:31112	UNKNOWN
COL1A2	Ehlers-Danlos syndrome, autosomal recessive, cardiac valvular form	OMIM:225320	MENDELIAN
COL1A2	Ehlers-Danlos syndrome, arthrochalasia type, 2	OMIM:617821	MENDELIAN
COL1A2	Osteoporosis	OMIM:166710	POLYGENIC
COL1A2	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2	OMIM:619120	MENDELIAN
COL1A2	Osteogenesis imperfecta, type III	OMIM:259420	MENDELIAN
COL1A2	Osteogenesis imperfecta, type II	OMIM:166210	MENDELIAN
COL1A2	Osteogenesis imperfecta, type IV	OMIM:166220	MENDELIAN
COL1A2	Arthrochalasia Ehlers-Danlos syndrome	ORPHA:1899	UNKNOWN
COL1A2	Cardiac-valvular Ehlers-Danlos syndrome	ORPHA:230851	UNKNOWN
COL21A1	—		UNKNOWN
COL25A1	Fibrosis of extraocular muscles, congenital, 5	OMIM:616219	MENDELIAN
COL25A1	Congenital fibrosis of extraocular muscles	ORPHA:45358	UNKNOWN
COL25A1	Neurogenic arthrogryposis multiplex congenita	ORPHA:1143	UNKNOWN
COL27A1	Steel syndrome	OMIM:615155	MENDELIAN
COL2A1	Spondyloepiphyseal dysplasia, Stanescu type	OMIM:616583	MENDELIAN
COL2A1	Spondylometaphyseal dysplasia, corner Fracture type	OMIM:184255	MENDELIAN
COL2A1	Spondylometaphyseal dysplasia, Algerian type	OMIM:184253	MENDELIAN
COL2A1	Spondyloepimetaphyseal dysplasia, Strudwick type	OMIM:184250	MENDELIAN
COL2A1	Czech dysplasia, Metatarsal type	OMIM:609162	MENDELIAN
COL2A1	Vitreoretinopathy with phalangeal epiphyseal dysplasia	OMIM:619248	MENDELIAN
COL2A1	Osteoarthritis with mild chondrodysplasia	OMIM:604864	MENDELIAN
COL2A1	Platyspondylic lethal skeletal dysplasia, Torrance type	OMIM:151210	MENDELIAN
COL2A1	Achondrogenesis, type II	OMIM:200610	MENDELIAN
COL2A1	Stickler syndrome, type I	OMIM:108300	MENDELIAN
COL2A1	Stickler syndrome, type I, nonsyndromic ocular	OMIM:609508	MENDELIAN
COL2A1	Avascular necrosis of femoral head, primary, 1	OMIM:608805	MENDELIAN
COL2A1	Epiphyseal dysplasia, multiple, with myopia and conductive deafness	OMIM:132450	MENDELIAN
COL2A1	Spondyloepiphyseal dysplasia congenita	OMIM:183900	MENDELIAN
COL2A1	Kniest dysplasia	OMIM:156550	MENDELIAN
COL2A1	Legg-Calve-Perthes disease	OMIM:150600	MENDELIAN
COL2A1	Spondyloperipheral dysplasia	OMIM:271700	MENDELIAN
COL2A1	Stickler syndrome type 1	ORPHA:90653	UNKNOWN
COL2A1	Autosomal dominant otospondylomegaepiphyseal dysplasia	ORPHA:166100	UNKNOWN
COL2A1	Familial avascular necrosis of femoral head	ORPHA:86820	UNKNOWN
COL2A1	Spondyloperipheral dysplasia-short ulna syndrome	ORPHA:1856	UNKNOWN
COL2A1	Legg-Calvé-Perthes disease	ORPHA:2380	UNKNOWN
COL2A1	Achondrogenesis type 2	ORPHA:93296	UNKNOWN
COL2A1	Spondylometaphyseal dysplasia, 'corner fracture' type	ORPHA:93315	UNKNOWN
COL2A1	Spondylometaphyseal dysplasia, Schmidt type	ORPHA:93316	UNKNOWN
COL2A1	Spondyloepimetaphyseal dysplasia congenita, Strudwick type	ORPHA:93346	UNKNOWN
COL2A1	Dyspondyloenchondromatosis	ORPHA:85198	UNKNOWN
COL2A1	Kniest dysplasia	ORPHA:485	UNKNOWN
COL2A1	Spondyloepiphyseal dysplasia congenita	ORPHA:94068	UNKNOWN
COL2A1	Platyspondylic dysplasia, Torrance type	ORPHA:85166	UNKNOWN
COL3A1	Polymicrogyria with or without vascular-type EDS	OMIM:618343	MENDELIAN
COL3A1	Ehlers-Danlos syndrome, Vascular type	OMIM:130050	MENDELIAN
COL3A1	Vascular Ehlers-Danlos syndrome	ORPHA:286	UNKNOWN
COL3A1	Acrogeria	ORPHA:2500	UNKNOWN
COL3A1	Familial cerebral saccular aneurysm	ORPHA:231160	UNKNOWN
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps	OMIM:611773	MENDELIAN
COL4A1	Retinal arteries, tortuosity of	OMIM:180000	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
COL4A1	Brain small vessel disease 1 with or without ocular anomalies	OMIM:175780	MENDELIAN
COL4A1	Microangiopathy and leukoencephalopathy, pontine, autosomal dominant	OMIM:618564	MENDELIAN
COL4A1	HANAC syndrome	ORPHA:73229	UNKNOWN
COL4A1	Walker-Warburg syndrome	ORPHA:899	UNKNOWN
COL4A2	Brain small vessel disease 2B, autosomal recessive	OMIM:621414	MENDELIAN
COL4A2	Brain small vessel disease 2	OMIM:614483	MENDELIAN
COL4A3	Alport syndrome 3, autosomal dominant	OMIM:104200	MENDELIAN
COL4A3	Alport syndrome, X-linked	OMIM:301050	MENDELIAN
COL4A3	Alport syndrome 3B, autosomal recessive	OMIM:620536	MENDELIAN
COL4A3	Hematuria, benign familial, 2	OMIM:620320	MENDELIAN
COL4A3	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
COL4A3	Autosomal recessive Alport syndrome	ORPHA:88919	UNKNOWN
COL4A3	Autosomal dominant Alport syndrome	ORPHA:88918	UNKNOWN
COL4A3BP	—		UNKNOWN
COL4A4	Hematuria, benign familial	OMIM:141200	MENDELIAN
COL4A4	Alport syndrome, X-linked	OMIM:301050	MENDELIAN
COL4A4	Alport syndrome 2, autosomal recessive	OMIM:203780	MENDELIAN
COL4A4	Autosomal recessive Alport syndrome	ORPHA:88919	UNKNOWN
COL4A4	Autosomal dominant Alport syndrome	ORPHA:88918	UNKNOWN
COL4A5	Alport syndrome, X-linked	OMIM:301050	MENDELIAN
COL4A5	X-linked Alport syndrome-diffuse leiomyomatosis	ORPHA:1018	UNKNOWN
COL4A5	X-linked Alport syndrome	ORPHA:88917	UNKNOWN
COL4A6	Deafness, X-linked 6	OMIM:300914	MENDELIAN
COL4A6	X-linked Alport syndrome-diffuse leiomyomatosis	ORPHA:1018	UNKNOWN
COL5A1	Fibromuscular dysplasia, multifocal	OMIM:619329	MENDELIAN
COL5A1	Ehlers-danlos syndrome, type I	OMIM:130000	MENDELIAN
COL5A1	Classical Ehlers-Danlos syndrome	ORPHA:287	UNKNOWN
COL5A2	Ehlers-Danlos syndrome, classic type, 2	OMIM:130010	MENDELIAN
COL5A2	Classical Ehlers-Danlos syndrome	ORPHA:287	UNKNOWN
COL6A1	Ullrich congenital muscular dystrophy 1	OMIM:254090	MENDELIAN
COL6A1	Bethlem myopathy 1A	OMIM:158810	MENDELIAN
COL6A1	Ullrich congenital muscular dystrophy	ORPHA:75840	UNKNOWN
COL6A1	Bethlem muscular dystrophy	ORPHA:610	UNKNOWN
COL6A2	Myosclerosis, autosomal recessive	OMIM:255600	MENDELIAN
COL6A2	Ullrich congenital muscular dystrophy 1B	OMIM:620727	MENDELIAN
COL6A2	Bethlem myopathy 1B	OMIM:620725	MENDELIAN
COL6A2	Ullrich congenital muscular dystrophy	ORPHA:75840	UNKNOWN
COL6A2	Bethlem muscular dystrophy	ORPHA:610	UNKNOWN
COL6A3	Dystonia 27	OMIM:616411	MENDELIAN
COL6A3	Bethlem myopathy 1C	OMIM:620726	MENDELIAN
COL6A3	Ullrich congenital muscular dystrophy 1C	OMIM:620728	MENDELIAN
COL6A3	Ullrich congenital muscular dystrophy	ORPHA:75840	UNKNOWN
COL6A3	Primary dystonia, DYT27 type	ORPHA:464440	UNKNOWN
COL6A3	Bethlem muscular dystrophy	ORPHA:610	UNKNOWN
COL6A4P2	—		UNKNOWN
COL6A5	—		UNKNOWN
COL6A6	—		UNKNOWN
COL7A1	Epidermolysis bullosa dystrophica, autosomal recessive	OMIM:226600	MENDELIAN
COL7A1	Epidermolysis bullosa dystrophica, pretibial	OMIM:131850	MENDELIAN
COL7A1	Epidermolysis bullosa dystrophica, autosomal dominant	OMIM:131750	MENDELIAN
COL7A1	Transient bullous dermolysis of the newborn	OMIM:131705	MENDELIAN
COL7A1	Epidermolysis bullosa with congenital localized absence of skin anddeformity of nails	OMIM:132000	MENDELIAN
COL7A1	Nail disorder, nonsyndromic congenital, 8	OMIM:607523	MENDELIAN
COL7A1	Epidermolysis bullosa pruriginosa	OMIM:604129	MENDELIAN
COL7A1	Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form	ORPHA:79408	UNKNOWN
COL7A1	Recessive dystrophic epidermolysis bullosa inversa	ORPHA:79409	UNKNOWN
COL7A1	Localized dystrophic epidermolysis bullosa, pretibial form	ORPHA:79410	UNKNOWN
COL7A1	Self-improving dystrophic epidermolysis bullosa	ORPHA:79411	UNKNOWN
COL7A1	Localized dystrophic epidermolysis bullosa, nails only	ORPHA:158676	UNKNOWN
COL7A1	Localized dystrophic epidermolysis bullosa, acral form	ORPHA:158673	UNKNOWN
COL7A1	Autosomal recessive generalized dystrophic epidermolysis bullosa, intermediate form	ORPHA:89842	UNKNOWN
COL7A1	Dystrophic epidermolysis bullosa pruriginosa	ORPHA:89843	UNKNOWN
COL7A1	Autosomal dominant generalized dystrophic epidermolysis bullosa	ORPHA:231568	UNKNOWN
COL8A2	Corneal dystrophy, posterior polymorphous, 2	OMIM:609140	MENDELIAN
COL8A2	Corneal dystrophy, fuchs endothelial, 1	OMIM:136800	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
COL8A2	Posterior polymorphous corneal dystrophy	ORPHA:98973	UNKNOWN
COL8A2	Fuchs endothelial corneal dystrophy	ORPHA:98974	UNKNOWN
COL9A1	Stickler syndrome, type IV	OMIM:614134	MENDELIAN
COL9A1	Epiphyseal dysplasia, multiple, 6	OMIM:614135	MENDELIAN
COL9A1	Autosomal recessive Stickler syndrome	ORPHA:250984	UNKNOWN
COL9A1	Multiple epiphyseal dysplasia due to collagen 9 anomaly	ORPHA:166002	UNKNOWN
COL9A2	Stickler syndrome, type V	OMIM:614284	MENDELIAN
COL9A2	Epiphyseal dysplasia, multiple, 2	OMIM:600204	MENDELIAN
COL9A2	Autosomal recessive Stickler syndrome	ORPHA:250984	UNKNOWN
COL9A2	Multiple epiphyseal dysplasia due to collagen 9 anomaly	ORPHA:166002	UNKNOWN
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy	OMIM:600969	MENDELIAN
COL9A3	Stickler syndrome, type VI	OMIM:620022	MENDELIAN
COL9A3	Autosomal recessive Stickler syndrome	ORPHA:250984	UNKNOWN
COL9A3	Multiple epiphyseal dysplasia due to collagen 9 anomaly	ORPHA:166002	UNKNOWN
COLEC11	3MC syndrome 2	OMIM:265050	MENDELIAN
COLEC11	3MC syndrome	ORPHA:293843	UNKNOWN
COLQ	Myasthenic syndrome, congenital, 5	OMIM:603034	MENDELIAN
COLQ	Synaptic congenital myasthenic syndrome	ORPHA:98915	UNKNOWN
COMMD1	—		UNKNOWN
COMP	Pseudoachondroplasia	OMIM:177170	MENDELIAN
COMP	Carpal tunnel syndrome 2	OMIM:619161	MENDELIAN
COMP	Epiphyseal dysplasia, multiple, 1	OMIM:132400	MENDELIAN
COMP	Pseudoachondroplasia	ORPHA:750	UNKNOWN
COMP	Multiple epiphyseal dysplasia type 1	ORPHA:93308	UNKNOWN
COMT	22q11.2 deletion syndrome	ORPHA:567	UNKNOWN
COPS2	—		UNKNOWN
COQ2	Multiple system atrophy 1, susceptibility to	OMIM:146500	POLYGENIC
COQ2	Coenzyme Q10 deficiency, primary, 1	OMIM:607426	MENDELIAN
COQ2	Multiple system atrophy, cerebellar type	ORPHA:227510	UNKNOWN
COQ2	Multiple system atrophy, parkinsonian type	ORPHA:98933	UNKNOWN
COQ4	Coenzyme Q10 deficiency, primary, 7	OMIM:616276	MENDELIAN
COQ4	Spastic ataxia 10, autosomal recessive	OMIM:620666	MENDELIAN
COQ5	Coenzyme Q10 deficiency, primary, 9	OMIM:619028	MENDELIAN
COQ6	Coenzyme Q10 deficiency, primary, 6	OMIM:614650	MENDELIAN
COQ6	Full schwannomatosis	ORPHA:93921	UNKNOWN
COQ9	Coenzyme Q10 deficiency, primary, 5	OMIM:614654	MENDELIAN
CORIN	Preeclampsia/eclampsia 5	OMIM:614595	MENDELIAN
CORIN	Cardiomyopathy, familial hypertrophic, 30, atrial	OMIM:620734	MENDELIAN
CORIN	Preeclampsia	ORPHA:275555	UNKNOWN
CORO1A	Immunodeficiency 8	OMIM:615401	MENDELIAN
COX10	Mitochondrial complex IV deficiency, nuclear type 3	OMIM:619046	MENDELIAN
COX10	Mitochondrial complex IV deficiency, nuclear type 1	OMIM:220110	MENDELIAN
COX14	Mitochondrial complex IV deficiency, nuclear type 10	OMIM:619053	MENDELIAN
COX15	Mitochondrial complex IV deficiency, nuclear type 6	OMIM:615119	MENDELIAN
COX15	Mitochondrial complex IV deficiency, nuclear type 1	OMIM:220110	MENDELIAN
COX20	Mitochondrial complex IV deficiency, nuclear type 11	OMIM:619054	MENDELIAN
COX4I1	Mitochondrial complex IV deficiency, nuclear type 16	OMIM:619060	MENDELIAN
COX4I2	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarialhyperostosis	OMIM:612714	MENDELIAN
COX5A	Mitochondrial complex IV deficiency, nuclear type 20	OMIM:619064	MENDELIAN
COX5B	—		UNKNOWN
COX6A1	Charcot-Marie-Tooth disease, recessive intermediate D	OMIM:616039	MENDELIAN
COX6B1	Mitochondrial complex IV deficiency, nuclear type 7	OMIM:619051	MENDELIAN
COX6B1	Mitochondrial complex IV deficiency, nuclear type 1	OMIM:220110	MENDELIAN
COX6C	—		UNKNOWN
COX7A1	—		UNKNOWN
COX7A2	—		UNKNOWN
COX7B	Linear skin defects with multiple congenital anomalies 1	OMIM:309801	MENDELIAN
COX7B	Linear skin defects with multiple congenital anomalies 2	OMIM:300887	MENDELIAN
COX7B	Microphthalmia with linear skin defects syndrome	ORPHA:2556	UNKNOWN
COX7B2	—		UNKNOWN
COX7C	—		UNKNOWN
CP	Aceruloplasminemia	OMIM:604290	MENDELIAN
CP	Aceruloplasminemia	ORPHA:48818	UNKNOWN
CPA1	Autosomal dominant hereditary chronic pancreatitis	ORPHA:676	UNKNOWN
CPA4	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
CPA6	Epilepsy, familial temporal lobe, 5	OMIM:614417	MENDELIAN
CPA6	Febrile seizures, familial, 11	OMIM:614418	MENDELIAN
CPA6	Genetic epilepsy with febrile seizure plus	ORPHA:36387	UNKNOWN
CPB2	—		UNKNOWN
CPE	Intellectual developmental disorder and hypogonadotropic hypogonadism	OMIM:619326	MENDELIAN
CPLX2	—		UNKNOWN
CPN1	Carboxypeptidase N deficiency	OMIM:212070	MENDELIAN
CPOX	Harderoporphyria	OMIM:618892	MENDELIAN
CPOX	Coproporphyria	OMIM:121300	MENDELIAN
CPOX	Hereditary coproporphyria	ORPHA:79273	UNKNOWN
CPQ	—		UNKNOWN
CPS1	Carbamoyl phosphate synthetase I deficiency, hyperammonemia due to	OMIM:237300	MENDELIAN
CPS1	Carbamoyl-phosphate synthetase 1 deficiency	ORPHA:147	UNKNOWN
CPSF7	—		UNKNOWN
CPT1A	Carnitine palmitoyltransferase I deficiency	OMIM:255120	MENDELIAN
CPT1A	Carnitine palmitoyl transferase 1A deficiency	ORPHA:156	UNKNOWN
CPT1B	—		UNKNOWN
CPT1C	Spastic paraplegia 73, autosomal dominant	OMIM:616282	MENDELIAN
CPT1C	Autosomal dominant spastic paraplegia type 73	ORPHA:444099	UNKNOWN
CPT2	Carnitine palmitoyltransferase II deficiency, myopathic, stress-induced	OMIM:255110	MENDELIAN
CPT2	Encephalopathy, acute, infection-induced, susceptibility to, 4	OMIM:614212	POLYGENIC
CPT2	Carnitine palmitoyltransferase II deficiency, lethal neonatal	OMIM:608836	MENDELIAN
CPT2	Carnitine palmitoyltransferase II deficiency, infantile	OMIM:600649	MENDELIAN
CPT2	Carnitine palmitoyl transferase II deficiency, myopathic form	ORPHA:228302	UNKNOWN
CPT2	Carnitine palmitoyl transferase II deficiency, severe infantile form	ORPHA:228305	UNKNOWN
CPT2	Carnitine palmitoyl transferase II deficiency, neonatal form	ORPHA:228308	UNKNOWN
CPXCR1	—		UNKNOWN
CPZ	—		UNKNOWN
CR2	Immunodeficiency, common variable, 7	OMIM:614699	MENDELIAN
CR2	Immunodeficiency, common variable, 2	OMIM:240500	MENDELIAN
CR2	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
CRADD	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly	OMIM:614499	MENDELIAN
CRAT	Neurodegeneration with brain iron accumulation 8	OMIM:617917	MENDELIAN
CRB1	Pigmented paravenous chorioretinal atrophy	OMIM:172870	MENDELIAN
CRB1	Retinitis pigmentosa 12	OMIM:600105	MENDELIAN
CRB1	Leber congenital amaurosis 8	OMIM:613835	MENDELIAN
CRB1	Leber congenital amaurosis	ORPHA:65	UNKNOWN
CRB1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
CRB1	Isolated complete colobomatous microphthalmia	ORPHA:35612	UNKNOWN
CRB2	Ventriculomegaly with cystic kidney disease	OMIM:219730	MENDELIAN
CRB2	Focal segmental glomerulosclerosis 9	OMIM:616220	MENDELIAN
CRB2	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
CRBN	Intellectual developmental disorder, autosomal recessive 2	OMIM:607417	MENDELIAN
CREB1	Histiocytoma, angiomatoid fibrous, somatic	OMIM:612160	MENDELIAN
CREB3	—		UNKNOWN
CREB3L1	Osteogenesis imperfecta, type XVI	OMIM:616229	MENDELIAN
CREB3L3	Hypertriglyceridemia 2	OMIM:619324	MENDELIAN
CREBBP	Rubinstein-Taybi syndrome 1	OMIM:180849	MENDELIAN
CREBBP	Menke-Hennekam syndrome 1	OMIM:618332	MENDELIAN
CREBBP	Rubinstein-Taybi syndrome due to 16p13.3 microdeletion	ORPHA:353281	UNKNOWN
CREBBP	Rubinstein-Taybi syndrome due to CREBBP mutations	ORPHA:353277	UNKNOWN
CREBBP	Menke-Hennekam syndrome	ORPHA:592574	UNKNOWN
CRELD1	Atrioventricular septal defect, partial, with heterotaxy syndrome	OMIM:606217	MENDELIAN
CRELD1	Jeffries-Lakhani neurodevelopmental syndrome	OMIM:620771	MENDELIAN
CRELD2	—		UNKNOWN
CRH	Sleep-related hypermotor epilepsy	ORPHA:98784	UNKNOWN
CRHR1	—		UNKNOWN
CRIP1	Short stature with microcephaly and distinctive facies	OMIM:615789	MENDELIAN
CRISP2	—		UNKNOWN
CRK	—		UNKNOWN
CRKL	Distal 22q11.2 microdeletion syndrome	ORPHA:261330	UNKNOWN
CRLF1	Crisponi/cold-induced sweating syndrome 1	OMIM:272430	MENDELIAN
CRLF1	Crisponi syndrome	ORPHA:1545	UNKNOWN
CRLF1	Idiopathic achalasia	ORPHA:930	UNKNOWN
CRLF2	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
CRMP1	—		UNKNOWN
CROCC	—		UNKNOWN
CRP	—		UNKNOWN
CRTAP	Osteogenesis imperfecta, type VII	OMIM:610682	MENDELIAN
CRTAP	Cole-Carpenter syndrome	ORPHA:2050	UNKNOWN
CRTC1	—		UNKNOWN
CRX	Retinitis pigmentosa	OMIM:268000	MENDELIAN
CRX	Leber congenital amaurosis 7	OMIM:613829	MENDELIAN
CRX	Cone-Rod dystrophy 2	OMIM:120970	MENDELIAN
CRX	Leber congenital amaurosis	ORPHA:65	UNKNOWN
CRX	Cone rod dystrophy	ORPHA:1872	UNKNOWN
CRX	Retinitis pigmentosa	ORPHA:791	UNKNOWN
CRY1	Delayed sleep phase disorder, susceptibility to	OMIM:614163	POLYGENIC
CRY2	—		UNKNOWN
CRYAA	Cataract 9, multiple types	OMIM:604219	MENDELIAN
CRYAA	Cataract-microcornea syndrome	ORPHA:1377	UNKNOWN
CRYAB	Cardiomyopathy, dilated, 1II	OMIM:615184	MENDELIAN
CRYAB	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related	OMIM:613869	MENDELIAN
CRYAB	Cataract 16, multiple types	OMIM:613763	MENDELIAN
CRYAB	Alpha-B crystallin-related late-onset myopathy	ORPHA:399058	UNKNOWN
CRYAB	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
CRYBA1	Cataract, congenital zonular, with sutural opacities	OMIM:600881	MENDELIAN
CRYBA2	Cataract 42	OMIM:115900	MENDELIAN
CRYBA4	Cataract 23	OMIM:610425	MENDELIAN
CRYBA4	Cataract-microcornea syndrome	ORPHA:1377	UNKNOWN
CRYBB1	Cataract 17, multiple types	OMIM:611544	MENDELIAN
CRYBB1	Cataract-microcornea syndrome	ORPHA:1377	UNKNOWN
CRYBB2	Cataract 3, multiple types	OMIM:601547	MENDELIAN
CRYBB2	Cataract-microcornea syndrome	ORPHA:1377	UNKNOWN
CRYBB3	Cataract 22, multiple types	OMIM:609741	MENDELIAN
CRYGA	—		UNKNOWN
CRYGB	Cataract, multiple types	OMIM:615188	MENDELIAN
CRYGC	Cataract 2, multiple types	OMIM:604307	MENDELIAN
CRYGC	Cataract-microcornea syndrome	ORPHA:1377	UNKNOWN
CRYGD	Cataract, crystalline aculeiform	OMIM:115700	MENDELIAN
CRYGD	Cataract-microcornea syndrome	ORPHA:1377	UNKNOWN
CRYGEP	—		UNKNOWN
CRYGS	Cataract 20, multiple types	OMIM:116100	MENDELIAN
CRYL1	—		UNKNOWN
CRYM	Deafness, autosomal dominant 40	OMIM:616357	MENDELIAN
CRYZL1	—		UNKNOWN
CSAG1	—		UNKNOWN
CSDE1	—		UNKNOWN
CSF1	—		UNKNOWN
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids	OMIM:221820	MENDELIAN
CSF1R	Brain abnormalities, neurodegeneration, and dysosteosclerosis	OMIM:618476	MENDELIAN
CSF2	—		UNKNOWN
CSF2RA	Surfactant metabolism dysfunction, pulmonary, 4	OMIM:300770	MENDELIAN
CSF2RA	Hereditary pulmonary alveolar proteinosis	ORPHA:264675	UNKNOWN
CSF2RB	Surfactant metabolism dysfunction, pulmonary, 5	OMIM:614370	MENDELIAN
CSF2RB	Hereditary pulmonary alveolar proteinosis	ORPHA:264675	UNKNOWN
CSF3R	Neutrophilia, hereditary	OMIM:162830	MENDELIAN
CSF3R	Neutropenia, severe congenital, 7, autosomal recessive	OMIM:617014	MENDELIAN
CSGALNACT1	Skeletal dysplasia, mild, with joint laxity and advanced bone age	OMIM:618870	MENDELIAN
CSGALNACT1	Desbuquois syndrome	ORPHA:1425	UNKNOWN
CSH1	—		UNKNOWN
CSMD1	—		UNKNOWN
CSMD3	—		UNKNOWN
CSNK1A1	—		UNKNOWN
CSNK1A1L	—		UNKNOWN
CSNK1D	Advanced sleep phase syndrome, familial, 2	OMIM:615224	MENDELIAN
CSNK1E	—		UNKNOWN
CSNK1G1	—		UNKNOWN
CSNK2A1	Okur-Chung neurodevelopmental syndrome	OMIM:617062	MENDELIAN
CSNK2A2	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
CSNK2A3	—		UNKNOWN
CSNK2B	Poirier-Bienvenu neurodevelopmental syndrome	OMIM:618732	MENDELIAN
CSPP1	Joubert syndrome 21	OMIM:615636	MENDELIAN
CSPP1	Joubert syndrome with Jeune asphyxiating thoracic dystrophy	ORPHA:397715	UNKNOWN
CSPP1	Meckel syndrome	ORPHA:564	UNKNOWN
CSPP1	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
CSRP3	Cardiomyopathy, familial hypertrophic, 12	OMIM:612124	MENDELIAN
CSRP3	Cardiomyopathy, dilated, 1M	OMIM:607482	MENDELIAN
CSRP3	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
CST3	Macular degeneration, age-related, 11	OMIM:611953	POLYGENIC
CST3	Leukodystrophy, adult-onset, autosomal dominant, without amyloid angiopathy	OMIM:621214	MENDELIAN
CST3	Cerebral amyloid angiopathy	OMIM:105150	MENDELIAN
CST3	ACys amyloidosis	ORPHA:100008	UNKNOWN
CST6	Ectodermal dysplasia 15, Hypohidrotic/hair type	OMIM:618535	MENDELIAN
CST9	—		UNKNOWN
CSTA	Peeling skin syndrome 4	OMIM:607936	MENDELIAN
CSTA	Acral peeling skin syndrome	ORPHA:263534	UNKNOWN
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	OMIM:254800	MENDELIAN
CSTB	Progressive myoclonic epilepsy type 1	ORPHA:308	UNKNOWN
CSTB	Autosomal recessive hypohidrotic ectodermal dysplasia	ORPHA:248	UNKNOWN
CSTF2	Intellectual developmental disorder, X-linked 113	OMIM:301116	MENDELIAN
CSTF2T	—		UNKNOWN
CT45A5	—		UNKNOWN
CTC1	Cerebroretinal microangiopathy with calcifications and cysts 1	OMIM:612199	MENDELIAN
CTC1	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
CTCF	Intellectual developmental disorder, autosomal dominant 21	OMIM:615502	MENDELIAN
CTCF	CTCF-related neurodevelopmental disorder	ORPHA:363611	UNKNOWN
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy	OMIM:604168	MENDELIAN
CTDP1	Congenital cataracts-facial dysmorphism-neuropathy syndrome	ORPHA:48431	UNKNOWN
CTF1	—		UNKNOWN
CTGF	—		UNKNOWN
CTH	Cystathioninuria	OMIM:219500	MENDELIAN
CTH	Cystathioninuria	ORPHA:212	UNKNOWN
CTHRC1	Barrett esophagus	OMIM:614266	MENDELIAN
CTLA4	Autoimmune lymphoproliferative syndrome, type V	OMIM:616100	MENDELIAN
CTLA4	SÃ©zary syndrome	ORPHA:3162	UNKNOWN
CTLA4	Classic mycosis fungoides	ORPHA:2584	UNKNOWN
CTLA4	Granulomatosis with polyangiitis	ORPHA:900	UNKNOWN
CTLA4	Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsufficiency	ORPHA:436159	UNKNOWN
CTLA4	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
CTNNA1	Macular dystrophy, butterfly-shaped pigmentary, 2	OMIM:608970	MENDELIAN
CTNNA2	Cortical dysplasia, complex, with other brain malformations 9	OMIM:618174	MENDELIAN
CTNNA3	Arrhythmogenic right ventricular dysplasia, familial, 13	OMIM:615616	MENDELIAN
CTNNB1	Neurodevelopmental disorder with spastic diplegia and visual defects	OMIM:615075	MENDELIAN
CTNNB1	Exudative vitreoretinopathy 7	OMIM:617572	MENDELIAN
CTNNB1	Colorectal cancer	OMIM:114500	MENDELIAN
CTNNB1	Ovarian cancer	OMIM:167000	MENDELIAN
CTNNB1	Hepatocellular carcinoma	OMIM:114550	MENDELIAN
CTNNB1	Medulloblastoma	OMIM:155255	MENDELIAN
CTNNB1	Pilomatrixoma, somatic	OMIM:132600	MENDELIAN
CTNNB1	Osteopathia striata-cranial sclerosis syndrome	ORPHA:2780	UNKNOWN
CTNNB1	Adrenocortical carcinoma	ORPHA:1501	UNKNOWN
CTNNB1	Pediatric hepatocellular carcinoma	ORPHA:33402	UNKNOWN
CTNNB1	Pilomatrixoma	ORPHA:91414	UNKNOWN
CTNNB1	Desmoid tumor	ORPHA:873	UNKNOWN
CTNNB1	Familial exudative vitreoretinopathy	ORPHA:891	UNKNOWN
CTNNB1	Acrofacial dysostosis, Weyers type	ORPHA:952	UNKNOWN
CTNNB1	Craniopharyngioma	ORPHA:54595	UNKNOWN
CTNNB1	Intellectual disability-eye abnormalities-microcephaly-peripheral spasticity syndrome	ORPHA:404473	UNKNOWN
CTNND1	Blepharocheilodontic syndrome 2	OMIM:617681	MENDELIAN
CTNND1	Blepharo-cheilo-odontic syndrome	ORPHA:1997	UNKNOWN
CTNND2	Monosomy 5p syndrome	ORPHA:281	UNKNOWN
CTNND2	Familial adult myoclonic epilepsy	ORPHA:86814	UNKNOWN
CTNS	Cystinosis, late-onset juvenile or adolescent nephropathic	OMIM:219900	MENDELIAN
CTNS	Cystinosis, nephropathic	OMIM:219800	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
CTNS	Cystinosis, adult nonnephropathic	OMIM:219750	MENDELIAN
CTNS	Infantile nephropathic cystinosis	ORPHA:411629	UNKNOWN
CTNS	Ocular cystinosis	ORPHA:411641	UNKNOWN
CTNS	Juvenile nephropathic cystinosis	ORPHA:411634	UNKNOWN
CTPS1	Immunodeficiency 24	OMIM:615897	MENDELIAN
CTR9	Nephroblastoma	ORPHA:654	UNKNOWN
CTRC	Pancreatitis, hereditary	OMIM:167800	POLYGENIC
CTRC	Autosomal dominant hereditary chronic pancreatitis	ORPHA:676	UNKNOWN
CTRC	Tropical pancreatitis	ORPHA:103918	UNKNOWN
CTSA	Galactosialidosis	OMIM:256540	MENDELIAN
CTSA	Brain small vessel disease 6 with leukoencephalopathy	OMIM:621394	MENDELIAN
CTSA	Galactosialidosis	ORPHA:351	UNKNOWN
CTSB	Keratolytic winter erythema	OMIM:148370	MENDELIAN
CTSB	Keratolytic winter erythema	ORPHA:50943	UNKNOWN
CTSC	Periodontitis, aggressive, 1	OMIM:170650	MENDELIAN
CTSC	Haim-Munk syndrome	OMIM:245010	MENDELIAN
CTSC	Papillon-Lefevre syndrome	OMIM:245000	MENDELIAN
CTSC	Haim-Munk syndrome	ORPHA:2342	UNKNOWN
CTSC	Papillon-Lefèvre syndrome	ORPHA:678	UNKNOWN
CTSD	Ceroid lipofuscinosis, neuronal, 10	OMIM:610127	MENDELIAN
CTSF	Ceroid lipofuscinosis, neuronal, 13	OMIM:615362	MENDELIAN
CTSG	—		UNKNOWN
CTSK	Pycnodysostosis	OMIM:265800	MENDELIAN
CTSK	Pycnodysostosis	ORPHA:763	UNKNOWN
CTSZ	—		UNKNOWN
CTTNBP2	—		UNKNOWN
CUBN	Megaloblastic anemia 1	OMIM:261100	MENDELIAN
CUBN	Proteinuria, chronic benign	OMIM:618884	MENDELIAN
CUBN	Imerslund-Gräsbeck syndrome	ORPHA:35858	UNKNOWN
CUEDC1	—		UNKNOWN
CUL2	—		UNKNOWN
CUL3	Neurodevelopmental disorder with or without autism or seizures	OMIM:619239	MENDELIAN
CUL3	Pseudohypoaldosteronism, type IIE	OMIM:614496	MENDELIAN
CUL4A	—		UNKNOWN
CUL4B	Intellectual developmental disorder, X-linked syndromic, Cabezas type	OMIM:300354	MENDELIAN
CUL4B	X-linked intellectual disability, Cabezas type	ORPHA:85293	UNKNOWN
CUL5	—		UNKNOWN
CUL7	3-M syndrome 1	OMIM:273750	MENDELIAN
CUL7	3M syndrome	ORPHA:2616	UNKNOWN
CUX1	Global developmental delay with or without impaired intellectual development	OMIM:618330	MENDELIAN
CUX2	Epileptic encephalopathy, early infantile, 67	OMIM:618141	MENDELIAN
CUX2	Lennox-Gastaut syndrome	ORPHA:2382	UNKNOWN
CWF19L1	Spinocerebellar ataxia, autosomal recessive 17	OMIM:616127	MENDELIAN
CWF19L1	Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency	ORPHA:453521	UNKNOWN
CX3CL1	—		UNKNOWN
CXCL10	—		UNKNOWN
CXCL11	—		UNKNOWN
CXCL16	—		UNKNOWN
CXCL5	—		UNKNOWN
CXCR3	—		UNKNOWN
CXCR4	Whim syndrome	OMIM:193670	MENDELIAN
CXCR4	WHIM syndrome	ORPHA:51636	UNKNOWN
CXCR6	—		UNKNOWN
CYB5A	Methemoglobinemia and ambiguous genitalia	OMIM:250790	MENDELIAN
CYB5A	46,XY difference of sex development due to isolated 17,20-lyase deficiency	ORPHA:90796	UNKNOWN
CYB5A	Autosomal recessive methemoglobinemia	ORPHA:621	UNKNOWN
CYB5R3	Methemoglobinemia due to deficiency of methemoglobin reductase	OMIM:250800	MENDELIAN
CYB5R3	Autosomal recessive methemoglobinemia	ORPHA:621	UNKNOWN
CYB5R4	—		UNKNOWN
CYBA	GRANULOMATOUS DISEASE, CHRONIC, AUTOSOMAL RECESSIVE, CYTOCHROME b-NEGATIVE	OMIM:233690	MENDELIAN
CYBA	Chronic granulomatous disease	ORPHA:379	UNKNOWN
CYBB	Chronic granulomatous disease, X-linked	OMIM:306400	MENDELIAN
CYBB	Atypical mycobacteriosis, familial, X-linked 2	OMIM:300645	MENDELIAN
CYBB	Chronic granulomatous disease	ORPHA:379	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
CYBRD1	—		UNKNOWN
CYC1	Mitochondrial complex III deficiency, nuclear type 6	OMIM:615453	MENDELIAN
CYCS	Thrombocytopenia 4	OMIM:612004	MENDELIAN
CYFIP1	—		UNKNOWN
CYLD	Frontotemporal dementia and/or amyotrophic lateral sclerosis 8	OMIM:619132	MENDELIAN
CYLD	Brooke-Spiegler syndrome	OMIM:605041	MENDELIAN
CYLD	Cylindromatosis, familial	OMIM:132700	MENDELIAN
CYLD	Trichoepithelioma, multiple familial, 1	OMIM:601606	MENDELIAN
CYLD	Familial cylindromatosis	ORPHA:211	UNKNOWN
CYLD	Familial multiple trichoepithelioma	ORPHA:867	UNKNOWN
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	OMIM:613743	MENDELIAN
CYP11A1	Inherited isolated adrenal insufficiency due to partial CYP11A1 deficiency	ORPHA:289548	UNKNOWN
CYP11A1	46,XY difference of sex development-adrenal insufficiency due to CYP11A1 deficiency	ORPHA:168558	UNKNOWN
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	OMIM:202010	MENDELIAN
CYP11B1	Aldosteronism, glucocorticoid-remediable	OMIM:103900	MENDELIAN
CYP11B1	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	ORPHA:90795	UNKNOWN
CYP11B1	Familial hyperaldosteronism type I	ORPHA:403	UNKNOWN
CYP11B2	Hypoaldosteronism, congenital, due to CMO II deficiency	OMIM:610600	MENDELIAN
CYP11B2	Corticosterone methyloxidase type I deficiency	OMIM:203400	MENDELIAN
CYP11B2	Familial hyperaldosteronism type I	ORPHA:403	UNKNOWN
CYP11B2	Early-onset familial hypoaldosteronism	ORPHA:556030	UNKNOWN
CYP17A1	Adrenal hyperplasia, congenital, due to 17-alpha-hydroxylase deficiency	OMIM:202110	MENDELIAN
CYP17A1	46,XY difference of sex development due to isolated 17,20-lyase deficiency	ORPHA:90796	UNKNOWN
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	ORPHA:90793	UNKNOWN
CYP19A1	Aromatase deficiency	OMIM:613546	MENDELIAN
CYP19A1	Aromatase deficiency	ORPHA:91	UNKNOWN
CYP19A1	Aromatase excess syndrome	ORPHA:178345	UNKNOWN
CYP1A1	—		UNKNOWN
CYP1A2	—		UNKNOWN
CYP1B1	Glaucoma 3, primary infantile, B	OMIM:600975	MENDELIAN
CYP1B1	Glaucoma 3, primary congenital, A	OMIM:231300	MENDELIAN
CYP1B1	Anterior segment dysgenesis 6, multiple subtypes	OMIM:617315	MENDELIAN
CYP1B1	Congenital glaucoma	ORPHA:98976	UNKNOWN
CYP1B1	Juvenile glaucoma	ORPHA:98977	UNKNOWN
CYP1B1	Peters anomaly	ORPHA:708	UNKNOWN
CYP21A1P	—		UNKNOWN
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	OMIM:201910	MENDELIAN
CYP24A1	Hypercalcemia, infantile, 1	OMIM:143880	MENDELIAN
CYP26A1	—		UNKNOWN
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies	OMIM:614416	MENDELIAN
CYP26C1	Focal facial dermal dysplasia 4	OMIM:614974	MENDELIAN
CYP26C1	Focal facial dermal dysplasia type IV	ORPHA:398189	UNKNOWN
CYP27A1	Cerebrotendinous xanthomatosis	OMIM:213700	MENDELIAN
CYP27A1	Cerebrotendinous xanthomatosis	ORPHA:909	UNKNOWN
CYP27B1	Vitamin D-dependent rickets, type I	OMIM:264700	MENDELIAN
CYP27B1	Hypocalcemic vitamin D-dependent rickets	ORPHA:289157	UNKNOWN
CYP2A13	—		UNKNOWN
CYP2A6	Coumarin resistance	OMIM:122700	MENDELIAN
CYP2A6	Lung cancer, susceptibility to	OMIM:211980	POLYGENIC
CYP2C18	—		UNKNOWN
CYP2C9	Coumarin resistance	OMIM:122700	MENDELIAN
CYP2D6	Drug metabolism, poor, cyp2d6-relateddrug metabolism, ultrarapid, cyp2d6-related, included	OMIM:608902	POLYGENIC
CYP2D7P1	—		UNKNOWN
CYP2E1	—		UNKNOWN
CYP2F1	—		UNKNOWN
CYP2G1P	—		UNKNOWN
CYP2J2	—		UNKNOWN
CYP2R1	Vitamin D hydroxylation-deficient rickets, type 1B	OMIM:600081	MENDELIAN
CYP2R1	Hypocalcemic vitamin D-dependent rickets	ORPHA:289157	UNKNOWN
CYP2U1	Spastic paraplegia 56, autosomal recessive	OMIM:615030	MENDELIAN
CYP2U1	Autosomal recessive spastic paraplegia type 56	ORPHA:320411	UNKNOWN
CYP2W1	—		UNKNOWN
CYP3A4	Vitamin d-dependent rickets, type 3	OMIM:619073	MENDELIAN
CYP3A43	—		UNKNOWN
CYP3A5	Hypertension, essential	OMIM:145500	POLYGENIC

Gene_symbol	Disease_name	Disease_ID	Association_type
CYP3A5P1	—		UNKNOWN
CYP3A7	—		UNKNOWN
CYP46A1	—		UNKNOWN
CYP4A11	—		UNKNOWN
CYP4A22	—		UNKNOWN
CYP4B1	—		UNKNOWN
CYP4F12	—		UNKNOWN
CYP4F2	—		UNKNOWN
CYP4F22	Ichthyosis, congenital, autosomal recessive 5	OMIM:604777	MENDELIAN
CYP4F22	Lamellar ichthyosis	ORPHA:313	UNKNOWN
CYP4F3	—		UNKNOWN
CYP4V2	Bietti crystalline corneoretinal dystrophy	OMIM:210370	MENDELIAN
CYP4V2	Bietti crystalline dystrophy	ORPHA:41751	UNKNOWN
CYP7A1	Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency	ORPHA:209902	UNKNOWN
CYP7B1	Spastic paraplegia 5A, autosomal recessive	OMIM:270800	MENDELIAN
CYP7B1	Bile acid synthesis defect, congenital, 3	OMIM:613812	MENDELIAN
CYP7B1	Autosomal recessive spastic paraplegia type 5A	ORPHA:100986	UNKNOWN
CYP7B1	Congenital bile acid synthesis defect type 3	ORPHA:79302	UNKNOWN
CYR61	—		UNKNOWN
CYS1	—		UNKNOWN
CYSLTR1	—		UNKNOWN
D2HGDH	D-2-Hydroxyglutaric aciduria 1	OMIM:600721	MENDELIAN
D2HGDH	D-2-hydroxyglutaric aciduria	ORPHA:79315	UNKNOWN
DAAM1	—		UNKNOWN
DAB1	Spinocerebellar ataxia 37	OMIM:615945	MENDELIAN
DAB1	Spinocerebellar ataxia type 37	ORPHA:363710	UNKNOWN
DAB2	—		UNKNOWN
DACH1	—		UNKNOWN
DAD1	—		UNKNOWN
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9	OMIM:616538	MENDELIAN
DAG1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9	OMIM:613818	MENDELIAN
DAG1	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	ORPHA:370997	UNKNOWN
DAG1	Alpha-dystroglycan-related limb-girdle muscular dystrophy R16	ORPHA:280333	UNKNOWN
DAG1	Walker-Warburg syndrome	ORPHA:899	UNKNOWN
DAG1	Isolated asymptomatic elevation of creatine phosphokinase	ORPHA:206599	UNKNOWN
DAO	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
DAOA	Schizophrenia	OMIM:181500	POLYGENIC
DAPK1	—		UNKNOWN
DAPK3	—		UNKNOWN
DAPP1	—		UNKNOWN
DARC	—		UNKNOWN
DARS	—		UNKNOWN
DARS2	Charcot-Marie-Tooth disease, axonal, type 2LL	OMIM:621485	MENDELIAN
DARS2	Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation	OMIM:611105	MENDELIAN
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome	ORPHA:137898	UNKNOWN
DAXX	—		UNKNOWN
DAZ1	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
DAZ1	Chromosome Y microdeletion syndrome	ORPHA:1646	UNKNOWN
DAZ2	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
DAZ2	Chromosome Y microdeletion syndrome	ORPHA:1646	UNKNOWN
DAZ3	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
DAZ3	Chromosome Y microdeletion syndrome	ORPHA:1646	UNKNOWN
DAZ4	Chromosome Y microdeletion syndrome	ORPHA:1646	UNKNOWN
DAZL	—		UNKNOWN
DBF4B	—		UNKNOWN
DBH	Orthostatic hypotension 1, due to DBH deficiency	OMIM:223360	MENDELIAN
DBH	Dopamine beta-hydroxylase deficiency	ORPHA:230	UNKNOWN
DBI	—		UNKNOWN
DBP	—		UNKNOWN
DBT	Maple syrup urine disease, type II	OMIM:620699	MENDELIAN
DCAF11	—		UNKNOWN
DCAF12L1	—		UNKNOWN
DCAF12L2	—		UNKNOWN
DCAF13	—		UNKNOWN
DCAF17	Woodhouse-Sakati syndrome	OMIM:241080	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
DCAF17	Woodhouse-Sakati syndrome	ORPHA:3464	UNKNOWN
DCAF4	—		UNKNOWN
DCAF8	Giant axonal neuropathy, autosomal dominant	OMIM:610100	MENDELIAN
DCAKD	—		UNKNOWN
DCBLD1	—		UNKNOWN
DCBLD2	—		UNKNOWN
DCC	Gaze palsy, familial horizontal, with progressive scoliosis, 2	OMIM:617542	MENDELIAN
DCC	Esophageal cancer, somatic	OMIM:133239	MENDELIAN
DCC	Colorectal cancer	OMIM:114500	MENDELIAN
DCC	Mirror movements 1 and/or agenesis of the corpus callosum	OMIM:157600	MENDELIAN
DCC	Horizontal gaze palsy with progressive scoliosis	ORPHA:2744	UNKNOWN
DCC	Familial congenital mirror movements	ORPHA:238722	UNKNOWN
DCC	Kallmann syndrome	ORPHA:478	UNKNOWN
DCDC2	Deafness, autosomal recessive 66	OMIM:610212	MENDELIAN
DCDC2	Nephronophthisis 19	OMIM:616217	MENDELIAN
DCDC2	Sclerosing cholangitis, neonatal	OMIM:617394	MENDELIAN
DCDC2	Senior-Boichis syndrome	ORPHA:84081	UNKNOWN
DCHS1	Van Maldergem syndrome 1	OMIM:601390	MENDELIAN
DCHS1	Mitral valve prolapse, myxomatous 2	OMIM:607829	MENDELIAN
DCHS1	Cerebrofacioarticular syndrome	ORPHA:314679	UNKNOWN
DCK	—		UNKNOWN
DCLK1	—		UNKNOWN
DCLRE1A	—		UNKNOWN
DCLRE1B	Dyskeratosis congenita, autosomal recessive 8	OMIM:620133	MENDELIAN
DCLRE1C	Omenn syndrome	OMIM:603554	MENDELIAN
DCLRE1C	Severe combined immunodeficiency with sensitivity to ionizing radiation	OMIM:602450	MENDELIAN
DCLRE1C	T-B-NK+ severe combined immunodeficiency due to DCLRE1C deficiency	ORPHA:275	UNKNOWN
DCLRE1C	Omenn syndrome	ORPHA:39041	UNKNOWN
DCN	Corneal dystrophy, congenital stromal	OMIM:610048	MENDELIAN
DCP1B	—		UNKNOWN
DCPS	Al-Raqad syndrome	OMIM:616459	MENDELIAN
DCTD	—		UNKNOWN
DCTN1	Amyotrophic lateral sclerosis 1	OMIM:105400	POLYGENIC
DCTN1	Neuronopathy, distal hereditary motor, type VIIB	OMIM:607641	MENDELIAN
DCTN1	Perry syndrome	OMIM:168605	MENDELIAN
DCTN1	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
DCTN1	Perry syndrome	ORPHA:178509	UNKNOWN
DCUN1D1	—		UNKNOWN
DCX	Lissencephaly, X-linked, 1	OMIM:300067	MENDELIAN
DCX	Lissencephaly type 1 due to doublecortin gene mutation	ORPHA:2148	UNKNOWN
DCXR	Pentosuria	OMIM:260800	MENDELIAN
DCXR	Pentosuria	ORPHA:2843	UNKNOWN
DDAH1	—		UNKNOWN
DDAH2	—		UNKNOWN
DDB1	White-Kernohan syndrome	OMIM:619426	MENDELIAN
DDB2	Xeroderma pigmentosum, complementation group E	OMIM:278740	MENDELIAN
DDB2	Xeroderma pigmentosum	ORPHA:910	UNKNOWN
DDC	Aromatic L-amino acid decarboxylase deficiency	OMIM:608643	MENDELIAN
DDC	Aromatic L-amino acid decarboxylase deficiency	ORPHA:35708	UNKNOWN
DDHD1	Spastic paraplegia 28, autosomal recessive	OMIM:609340	MENDELIAN
DDHD1	Autosomal recessive spastic paraplegia type 28	ORPHA:101008	UNKNOWN
DDHD2	Spastic paraplegia 54, autosomal recessive	OMIM:615033	MENDELIAN
DDHD2	Autosomal recessive spastic paraplegia type 54	ORPHA:320380	UNKNOWN
DDIT3	Myxoid/round cell liposarcoma	ORPHA:99967	UNKNOWN
DDOST	Congenital disorder of glycosylation, type IR	OMIM:614507	MENDELIAN
DDOST	DDOST-CDG	ORPHA:300536	UNKNOWN
DDR1	—		UNKNOWN
DDR2	Warburg-Cinotti syndrome	OMIM:618175	MENDELIAN
DDR2	Spondylometaphyseal dysplasia, short Limb-Hand type	OMIM:271665	MENDELIAN
DDR GK1	Spondyloepimetaphyseal dysplasia, Shohat type	OMIM:602557	MENDELIAN
DDR GK1	Spondyloepimetaphyseal dysplasia, Shohat type	ORPHA:93352	UNKNOWN
DDX10	—		UNKNOWN
DDX11	Warsaw breakage syndrome	OMIM:613398	MENDELIAN
DDX20	—		UNKNOWN
DDX23	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
DDX25	—		UNKNOWN
DDX39B	—		UNKNOWN
DDX3X	Intellectual developmental disorder, X-linked, syndrome, snijders Blok type	OMIM:300958	MENDELIAN
DDX3X	Toriello-Carey syndrome	ORPHA:3338	UNKNOWN
DDX3X	X-linked intellectual disability-hypotonia-movement disorder syndrome	ORPHA:457260	UNKNOWN
DDX3Y	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
DDX3Y	Chromosome Y microdeletion syndrome	ORPHA:1646	UNKNOWN
DDX41	Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to	OMIM:616871	POLYGENIC
DDX47	—		UNKNOWN
DDX5	—		UNKNOWN
DDX53	—		UNKNOWN
DDX58	—		UNKNOWN
DDX59	Orofaciodigital syndrome V	OMIM:174300	MENDELIAN
DDX59	Orofaciodigital syndrome type 5	ORPHA:2919	UNKNOWN
DEAF1	Vulto-van Silfout-de Vries syndrome	OMIM:615828	MENDELIAN
DEAF1	Dyskinesia, seizures, and intellectual developmental disorder	OMIM:617171	MENDELIAN
DEAF1	Intellectual disability-epilepsy-extrapyrmidal syndrome	ORPHA:468620	UNKNOWN
DEC1	—		UNKNOWN
DECR1	—		UNKNOWN
DEF6	Immunodeficiency 87 and autoimmunity	OMIM:619573	MENDELIAN
DEFB1	—		UNKNOWN
DEFB124	—		UNKNOWN
DEFB126	—		UNKNOWN
DEFB4A	—		UNKNOWN
DENND1A	—		UNKNOWN
DENND2B	—		UNKNOWN
DENND5A	Epileptic encephalopathy, early infantile, 49	OMIM:617281	MENDELIAN
DENR	—		UNKNOWN
DEPDC5	Epilepsy, familial focal, with variable foci	OMIM:604364	MENDELIAN
DEPDC5	Developmental and epileptic encephalopathy 111	OMIM:620504	MENDELIAN
DEPDC5	Sleep-related hypermotor epilepsy	ORPHA:98784	UNKNOWN
DEPDC5	Familial focal epilepsy with variable foci	ORPHA:98820	UNKNOWN
DEPDC5	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
DES	Cardiomyopathy, dilated, 1l	OMIM:604765	MENDELIAN
DES	Myopathy, myofibrillar, 1	OMIM:601419	MENDELIAN
DES	Scapuloperoneal syndrome, neurogenic, Kaeser type	OMIM:181400	MENDELIAN
DES	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
DES	Desminopathy	ORPHA:98909	UNKNOWN
DESI2	—		UNKNOWN
DFNA5	—		UNKNOWN
DFNB31	—		UNKNOWN
DFNB59	—		UNKNOWN
DGAT1	Diarrhea 7, protein-losing Enteropathy type	OMIM:615863	MENDELIAN
DGAT2L6	—		UNKNOWN
DGCR14	—		UNKNOWN
DGCR2	Velocardiofacial syndrome	OMIM:192430	MENDELIAN
DGCR5	—		UNKNOWN
DGCR6	Velocardiofacial syndrome	OMIM:192430	MENDELIAN
DGCR8	Velocardiofacial syndrome	OMIM:192430	MENDELIAN
DGKD	—		UNKNOWN
DGKE	Nephrotic syndrome, type 7	OMIM:615008	MENDELIAN
DGKE	Immunoglobulin-mediated membranoproliferative glomerulonephritis	ORPHA:329903	UNKNOWN
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	OMIM:251880	MENDELIAN
DGUOK	Portal hypertension, noncirrhotic	OMIM:617068	MENDELIAN
DGUOK	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4	OMIM:617070	MENDELIAN
DGUOK	Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency	ORPHA:329314	UNKNOWN
DHCR24	Desmosterolosis	OMIM:602398	MENDELIAN
DHCR24	Desmosterolosis	ORPHA:35107	UNKNOWN
DHCR7	Smith-Lemli-Opitz syndrome	OMIM:270400	MENDELIAN
DHCR7	Smith-Lemli-Opitz syndrome	ORPHA:818	UNKNOWN
DHDDS	Developmental delay and seizures with or without movement abnormalities	OMIM:617836	MENDELIAN
DHDDS	Congenital disorder of glycosylation, type 1bb	OMIM:621567	MENDELIAN
DHDDS	Retinitis pigmentosa 59	OMIM:613861	MENDELIAN
DHDDS	Retinitis pigmentosa	ORPHA:791	UNKNOWN
DHDDS	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency	OMIM:613839	MENDELIAN
DHH	46XY gonadal dysgenesis with minifascicular neuropathy	OMIM:607080	MENDELIAN
DHH	46,xy sex reversal 7	OMIM:233420	MENDELIAN
DHH	46,XY complete gonadal dysgenesis	ORPHA:242	UNKNOWN
DHH	46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome	ORPHA:168563	UNKNOWN
DHODH	Postaxial acrofacial dysostosis	OMIM:263750	MENDELIAN
DHODH	Postaxial acrofacial dysostosis	ORPHA:246	UNKNOWN
DHRS4L1	—		UNKNOWN
DHTKD1	Charcot-Marie-Tooth disease, axonal, type 2Q	OMIM:615025	MENDELIAN
DHTKD1	Alpha-aminoadipic and alpha-ketoadipic aciduria	OMIM:204750	MENDELIAN
DHTKD1	2-aminoadipic 2-oxoadipic aciduria	ORPHA:79154	UNKNOWN
DHX15	—		UNKNOWN
DHX16	Neuromuscular disease and ocular or auditory anomalies with or without seizures	OMIM:618733	MENDELIAN
DHX30	Neurodevelopmental disorder with severe motor impairment and absent language	OMIM:617804	MENDELIAN
DHX36	—		UNKNOWN
DHX37	46, XY sex reversal 11	OMIM:273250	MENDELIAN
DHX37	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies	OMIM:618731	MENDELIAN
DHX37	46,XY partial gonadal dysgenesis	ORPHA:251510	UNKNOWN
DHX37	46,XY complete gonadal dysgenesis	ORPHA:242	UNKNOWN
DHX37	Testicular regression syndrome	ORPHA:983	UNKNOWN
DHX38	Retinitis pigmentosa 84	OMIM:618220	MENDELIAN
DHX38	Retinitis pigmentosa	ORPHA:791	UNKNOWN
DHX9	Intellectual developmental disorder, autosomal dominant 75	OMIM:620988	MENDELIAN
DIABLO	Deafness, autosomal dominant 64	OMIM:614152	MENDELIAN
DIAPH1	Deafness, autosomal dominant 1	OMIM:124900	MENDELIAN
DIAPH1	Seizures, cortical blindness, and microcephaly syndrome	OMIM:616632	MENDELIAN
DIAPH1	Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome	ORPHA:477814	UNKNOWN
DIAPH1	DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome	ORPHA:494444	UNKNOWN
DIAPH1	Moyamoya disease	ORPHA:2573	UNKNOWN
DIAPH2	Premature ovarian failure 2A	OMIM:300511	MENDELIAN
DIAPH3	Auditory neuropathy, autosomal dominant, 1	OMIM:609129	MENDELIAN
DICER1	Global developmental delay, lung cysts, overgrowth, and wilms tumor	OMIM:618272	MENDELIAN
DICER1	Pleuropulmonary blastoma	OMIM:601200	MENDELIAN
DICER1	Goiter, multinodular 1	OMIM:138800	MENDELIAN
DICER1	Rhabdomyosarcoma, embryonal, 2	OMIM:180295	MENDELIAN
DICER1	Familial multinodular goiter	ORPHA:276399	UNKNOWN
DIO1	Thyroid hormone metabolism, abnormal, 2	OMIM:619855	MENDELIAN
DIO2	—		UNKNOWN
DIP2A	—		UNKNOWN
DIP2B	Intellectual developmental disorder, autosomal dominant, FRA12A type	OMIM:136630	MENDELIAN
DIP2C	—		UNKNOWN
DIRC2	—		UNKNOWN
DIS3	—		UNKNOWN
DIS3L2	Perlman syndrome	OMIM:267000	MENDELIAN
DIS3L2	Perlman syndrome	ORPHA:2849	UNKNOWN
DIS3L2	Nephroblastoma	ORPHA:654	UNKNOWN
DISC2	Schizophrenia	OMIM:181500	MENDELIAN
DISP1	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
DISP1	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
DISP1	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
DISP1	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
DISP1	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
DISP1	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
DKC1	Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1	OMIM:301108	MENDELIAN
DKC1	Dyskeratosis congenita, X-linked	OMIM:305000	MENDELIAN
DKC1	Hoyeraal-Hreidarsson syndrome	ORPHA:3322	UNKNOWN
DKC1	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
DKK1	Arnold-Chiari malformation type I	ORPHA:268882	UNKNOWN
DKK1	Idiopathic juvenile osteoporosis	ORPHA:85193	UNKNOWN
DKK2	—		UNKNOWN
DKK3	—		UNKNOWN
DKK4	—		UNKNOWN
DLAT	Pyruvate dehydrogenase E2 deficiency	OMIM:245348	MENDELIAN
DLAT	Pyruvate dehydrogenase E2 deficiency	ORPHA:79244	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
DLC1	Colorectal cancer	OMIM:114500	MENDELIAN
DLD	Dihydrolipoamide dehydrogenase deficiency	OMIM:246900	MENDELIAN
DLD	Pyruvate dehydrogenase E3 deficiency	ORPHA:2394	UNKNOWN
DLEC1	Squamous cell carcinoma of the esophagus	ORPHA:99977	UNKNOWN
DLG1	Cleft lip/palate	ORPHA:199306	UNKNOWN
DLG2	—		UNKNOWN
DLG3	Intellectual developmental disorder, X-linked 90	OMIM:300850	MENDELIAN
DLG4	Intellectual developmental disorder 62	OMIM:618793	MENDELIAN
DLG5	Yuksel-Vogel-Bauser syndrome	OMIM:620703	MENDELIAN
DLGAP2	—		UNKNOWN
DLGAP3	—		UNKNOWN
DLL1	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures	OMIM:618709	MENDELIAN
DLL1	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
DLL1	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
DLL1	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
DLL1	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
DLL1	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
DLL1	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
DLL3	Spondylocostal dysostosis, autosomal recessive 1	OMIM:277300	MENDELIAN
DLL3	Autosomal recessive spondylocostal dysostosis	ORPHA:2311	UNKNOWN
DLL4	Adams-Oliver syndrome 6	OMIM:616589	MENDELIAN
DLL4	Adams-Oliver syndrome	ORPHA:974	UNKNOWN
DLL4	Aplasia cutis congenita	ORPHA:1114	UNKNOWN
DLST	Paragangliomas 7	OMIM:618475	MENDELIAN
DLST	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
DLX3	Trichodentoosseous syndrome	OMIM:190320	MENDELIAN
DLX3	Amelogenesis imperfecta, type IV	OMIM:104510	MENDELIAN
DLX3	Tricho-dento-osseous syndrome	ORPHA:3352	UNKNOWN
DLX5	Split-Hand/foot malformation 1 with sensorineural hearing loss	OMIM:220600	MENDELIAN
DLX5	Split-Hand/foot malformation 1	OMIM:183600	MENDELIAN
DLX5	Isolated split hand-split foot malformation	ORPHA:2440	UNKNOWN
DLX6	Isolated split hand-split foot malformation	ORPHA:2440	UNKNOWN
DMBT1	—		UNKNOWN
DMBX1	—		UNKNOWN
DMC1	—		UNKNOWN
DMD	Cardiomyopathy, dilated, 3B	OMIM:302045	MENDELIAN
DMD	Muscular dystrophy, Becker type	OMIM:300376	MENDELIAN
DMD	Duchenne muscular dystrophy	OMIM:310200	MENDELIAN
DMD	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
DMD	Becker muscular dystrophy	ORPHA:98895	UNKNOWN
DMD	Duchenne muscular dystrophy	ORPHA:98896	UNKNOWN
DMD	Symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers	ORPHA:206546	UNKNOWN
DMGDH	Dimethylglycine dehydrogenase deficiency	OMIM:605850	MENDELIAN
DMGDH	Dimethylglycine dehydrogenase deficiency	ORPHA:243343	UNKNOWN
DMP1	Hypophosphatemic rickets, autosomal recessive	OMIM:241520	MENDELIAN
DMP1	Autosomal recessive hypophosphatemic rickets	ORPHA:289176	UNKNOWN
DMPK	Myotonic dystrophy 1	OMIM:160900	MENDELIAN
DMPK	Congenital-onset Steinert myotonic dystrophy	ORPHA:589821	UNKNOWN
DMRT1	46,XY complete gonadal dysgenesis	ORPHA:242	UNKNOWN
DMXL1	—		UNKNOWN
DMXL2	Deafness, autosomal dominant 71	OMIM:617605	MENDELIAN
DMXL2	Polyendocrine-polyneuropathy syndrome	OMIM:616113	MENDELIAN
DMXL2	Epileptic encephalopathy, early infantile, 81	OMIM:618663	MENDELIAN
DMXL2	Polyendocrine-polyneuropathy syndrome	ORPHA:453533	UNKNOWN
DMXL2	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
DNA2	Progressive external ophthalmoplegia with mitochondrial DNA deletions,autosomal dominant, 6	OMIM:615156	MENDELIAN
DNA2	Seckel syndrome 8	OMIM:615807	MENDELIAN
DNA2	Rothmund-Thomson syndrome, type 4	OMIM:620819	MENDELIAN
DNA2	DNA2-related mitochondrial DNA deletion syndrome	ORPHA:352470	UNKNOWN
DNA2	Seckel syndrome	ORPHA:808	UNKNOWN
DNAAF1	Ciliary dyskinesia, primary, 13	OMIM:613193	MENDELIAN
DNAAF1	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DNAAF2	Ciliary dyskinesia, primary, 10	OMIM:612518	MENDELIAN
DNAAF2	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DNAAF3	Ciliary dyskinesia, primary, 2	OMIM:606763	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
DNAAF3	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DNAH1	Spermatogenic failure 18	OMIM:617576	MENDELIAN
DNAH1	Ciliary dyskinesia, primary, 37	OMIM:617577	MENDELIAN
DNAH1	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DNAH11	Ciliary dyskinesia, primary, 7	OMIM:611884	MENDELIAN
DNAH11	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DNAH17	Spermatogenic failure 39	OMIM:618643	MENDELIAN
DNAH3	—		UNKNOWN
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus	OMIM:608644	MENDELIAN
DNAH5	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DNAH6	—		UNKNOWN
DNAH7	Ciliary dyskinesia, primary, 50	OMIM:620356	MENDELIAN
DNAH7	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DNAH9	Ciliary dyskinesia, primary, 40	OMIM:618300	MENDELIAN
DNAH9	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DNAI1	Ciliary dyskinesia, primary, 1	OMIM:244400	MENDELIAN
DNAI1	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus	OMIM:612444	MENDELIAN
DNAI2	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DNAJA4	—		UNKNOWN
DNAJB2	Spinal muscular atrophy, distal, autosomal recessive, 5	OMIM:614881	MENDELIAN
DNAJB6	Muscular dystrophy, limb-girdle, type 1E	OMIM:603511	MENDELIAN
DNAJB6	DNAJB6-related limb-girdle muscular dystrophy D1	ORPHA:34516	UNKNOWN
DNAJC13	Hereditary late-onset Parkinson disease	ORPHA:411602	UNKNOWN
DNAJC19	3-methylglutaconic aciduria, type V	OMIM:610198	MENDELIAN
DNAJC19	Dilated cardiomyopathy with ataxia	ORPHA:66634	UNKNOWN
DNAJC3	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus	OMIM:616192	MENDELIAN
DNAJC3	Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome	ORPHA:445062	UNKNOWN
DNAJC30	Leber hereditary optic neuropathy	ORPHA:104	UNKNOWN
DNAJC30	Williams syndrome	ORPHA:904	UNKNOWN
DNAJC5	Ceroid lipofuscinosis, neuronal, 4B, autosomal dominant	OMIM:162350	MENDELIAN
DNAJC6	Parkinson disease 19a, juvenile-onset	OMIM:615528	MENDELIAN
DNAJC6	Young-onset Parkinson disease	ORPHA:2828	UNKNOWN
DNAJC6	Atypical juvenile parkinsonism	ORPHA:391411	UNKNOWN
DNAL1	Ciliary dyskinesia, primary, 16	OMIM:614017	MENDELIAN
DNAL1	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DNAL4	Mirror movements 3	OMIM:616059	MENDELIAN
DNAL4	Familial congenital mirror movements	ORPHA:238722	UNKNOWN
DNASE1	Systemic lupus erythematosus	OMIM:152700	POLYGENIC
DNASE1	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
DNASE1L3	Systemic lupus erythematosus 16	OMIM:614420	MENDELIAN
DNASE1L3	Hypocomplementemic urticarial vasculitis	ORPHA:36412	UNKNOWN
DNASE2	Autoinflammatory-pancytopenia syndrome	OMIM:619858	MENDELIAN
DND1	—		UNKNOWN
DNM1	Epileptic encephalopathy, early infantile, 31	OMIM:616346	MENDELIAN
DNM1	Developmental and epileptic encephalopathy 31B, autosomal recessive	OMIM:620352	MENDELIAN
DNM1	Lennox-Gastaut syndrome	ORPHA:2382	UNKNOWN
DNM1	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
DNM1L	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1	OMIM:614388	MENDELIAN
DNM1L	Optic atrophy 5	OMIM:610708	MENDELIAN
DNM1L	DNM1L-related encephalopathy due to mitochondrial and peroxisomal fission defect	ORPHA:330050	UNKNOWN
DNM1L	Autosomal dominant optic atrophy, classic form	ORPHA:98673	UNKNOWN
DNM2	Lethal congenital contracture syndrome 5	OMIM:615368	MENDELIAN
DNM2	Centronuclear myopathy 1	OMIM:160150	MENDELIAN
DNM2	Charcot-Marie-Tooth disease, dominant intermediate B	OMIM:606482	MENDELIAN
DNM2	Autosomal dominant centronuclear myopathy	ORPHA:169189	UNKNOWN
DNMT1	Neuropathy, hereditary sensory, type IE	OMIM:614116	MENDELIAN
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant	OMIM:604121	MENDELIAN
DNMT1	Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome	ORPHA:314404	UNKNOWN
DNMT3A	Tatton-Brown-Rahman syndrome	OMIM:615879	MENDELIAN
DNMT3A	Heyn-Sproul-Jackson syndrome	OMIM:618724	MENDELIAN
DNMT3A	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
DNMT3A	Tatton-Brown-Rahman syndrome	ORPHA:404443	UNKNOWN
DNMT3A	Sporadic pheochromocytoma/secreting paraganglioma	ORPHA:276621	UNKNOWN
DNMT3B	Facioscapulohumeral muscular dystrophy 4, digenic	OMIM:619478	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
DNMT3B	Immunodeficiency-Centromeric instability-facial anomalies syndrome	OMIM:242860	MENDELIAN
DNMT3B	Facioscapulohumeral dystrophy	ORPHA:269	UNKNOWN
DNMT3B	ICF syndrome	ORPHA:2268	UNKNOWN
DNMT3L	—		UNKNOWN
DNTT	—		UNKNOWN
DOC2A	—		UNKNOWN
DOCK3	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia	OMIM:618292	MENDELIAN
DOCK4	—		UNKNOWN
DOCK6	Adams-Oliver syndrome 2	OMIM:614219	MENDELIAN
DOCK6	Adams-Oliver syndrome	ORPHA:974	UNKNOWN
DOCK7	Developmental and epileptic encephalopathy 23	OMIM:615859	MENDELIAN
DOCK7	Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome	ORPHA:411986	UNKNOWN
DOCK8	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections	OMIM:243700	MENDELIAN
DOCK8	Combined immunodeficiency due to DOCK8 deficiency	ORPHA:217390	UNKNOWN
DOCK9	—		UNKNOWN
DOK1	—		UNKNOWN
DOK2	—		UNKNOWN
DOK5	—		UNKNOWN
DOK7	Myasthenia, limb-girdle, familial	OMIM:254300	MENDELIAN
DOK7	Fetal akinesia deformation sequence 3	OMIM:618389	MENDELIAN
DOK7	Fetal akinesia deformation sequence	ORPHA:994	UNKNOWN
DOLK	Congenital disorder of glycosylation, type Im	OMIM:610768	MENDELIAN
DOLK	DK1-CDG	ORPHA:91131	UNKNOWN
DOLK	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
DOLPP1	—		UNKNOWN
DOT1L	Nil-Deshwar neurodevelopmental syndrome	OMIM:621265	MENDELIAN
DPAGT1	Congenital disorder of glycosylation, type Ij	OMIM:608093	MENDELIAN
DPAGT1	Myasthenic syndrome, congenital, 13, with tubular aggregates	OMIM:614750	MENDELIAN
DPAGT1	Congenital myasthenic syndrome with glycosylation defect	ORPHA:353327	UNKNOWN
DPAGT1	DPAGT1-CDG	ORPHA:86309	UNKNOWN
DPCD	—		UNKNOWN
DPH1	Developmental delay with short stature, dysmorphic features, and sparse hair	OMIM:616901	MENDELIAN
DPH1	Craniofacial dysplasia-short stature-ectodermal anomalies-intellectual disability syndrome	ORPHA:459061	UNKNOWN
DPM1	Congenital disorder of glycosylation, type IE	OMIM:608799	MENDELIAN
DPM1	DPM1-CDG	ORPHA:79322	UNKNOWN
DPM2	Congenital disorder of glycosylation, type Iu	OMIM:615042	MENDELIAN
DPM2	Congenital muscular dystrophy with intellectual disability and severe epilepsy	ORPHA:329178	UNKNOWN
DPM3	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15	OMIM:612937	MENDELIAN
DPM3	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15	OMIM:618992	MENDELIAN
DPM3	DPM3-CDG	ORPHA:263494	UNKNOWN
DPP10	—		UNKNOWN
DPP3	—		UNKNOWN
DPP4	—		UNKNOWN
DPP6	Ventricular fibrillation, paroxysmal familial, 2	OMIM:612956	POLYGENIC
DPP6	Intellectual developmental disorder, autosomal dominant 33	OMIM:616311	MENDELIAN
DPP6	Autosomal dominant primary microcephaly	ORPHA:2514	UNKNOWN
DPY19L2	Spermatogenic failure 9	OMIM:613958	MENDELIAN
DPYD	Dihydropyrimidine dehydrogenase deficiency	OMIM:274270	MENDELIAN
DPYD	1p21.3 microdeletion syndrome	ORPHA:293948	UNKNOWN
DPYD	Dihydropyrimidine dehydrogenase deficiency	ORPHA:1675	UNKNOWN
DPYS	Dihydropyrimidinuria	OMIM:222748	MENDELIAN
DPYS	Dihydropyrimidinuria	ORPHA:38874	UNKNOWN
DRC1	Ciliary dyskinesia, primary, 21	OMIM:615294	MENDELIAN
DRC1	Spermatogenic failure 80	OMIM:620222	MENDELIAN
DRC1	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
DRD1	—		UNKNOWN
DRD2	Myoclonus-dystonia syndrome	ORPHA:36899	UNKNOWN
DRD3	Tremor, hereditary essential, 1	OMIM:190300	POLYGENIC
DRD3	Schizophrenia	OMIM:181500	POLYGENIC
DRD4	Attention deficit-hyperactivity disorder	OMIM:143465	POLYGENIC
DRD5	Blepharospasm, benign essential, susceptibility to	OMIM:606798	POLYGENIC
DRD5	Attention deficit-hyperactivity disorder	OMIM:143465	POLYGENIC
DROSHA	—		UNKNOWN
DRP2	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
DSC2	Arrhythmogenic right ventricular dysplasia, familial, 11	OMIM:610476	MENDELIAN
DSC3	Hypotrichosis and recurrent skin vesicles	OMIM:613102	MENDELIAN
DSCAM	—		UNKNOWN
DSCR8	—		UNKNOWN
DSE	Ehlers-Danlos syndrome, musculocontractural type, 2	OMIM:615539	MENDELIAN
DSE	Musculocontractural Ehlers-Danlos syndrome	ORPHA:2953	UNKNOWN
DSG1	Keratosis palmoplantaris striata I	OMIM:148700	MENDELIAN
DSG1	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper-ige	OMIM:615508	MENDELIAN
DSG1	Striate palmoplantar keratoderma	ORPHA:50942	UNKNOWN
DSG2	Cardiomyopathy, dilated, 1BB	OMIM:612877	MENDELIAN
DSG2	Arrhythmogenic right ventricular dysplasia, familial, 10	OMIM:610193	MENDELIAN
DSG2	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
DSG3	Blistering, acantholytic, of oral and laryngeal mucosa	OMIM:619226	MENDELIAN
DSG4	Hypotrichosis 6	OMIM:607903	MENDELIAN
DSG4	Hypotrichosis simplex	ORPHA:55654	UNKNOWN
DSG4	Monilethrix	ORPHA:573	UNKNOWN
DSP	Keratosis palmoplantaris striata II	OMIM:612908	MENDELIAN
DSP	Cardiomyopathy, dilated, with woolly hair and keratoderma	OMIM:605676	MENDELIAN
DSP	Epidermolysis bullosa, lethal acantholytic	OMIM:609638	MENDELIAN
DSP	Cardiomyopathy, dilated, with woolly hair, keratoderma, and tooth agenesis	OMIM:615821	MENDELIAN
DSP	Arrhythmogenic right ventricular dysplasia, familial, 8	OMIM:607450	MENDELIAN
DSP	Lethal acantholytic erosive disorder	ORPHA:158687	UNKNOWN
DSP	Idiopathic pulmonary fibrosis	ORPHA:2032	UNKNOWN
DSP	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
DSP	Carvajal syndrome	ORPHA:65282	UNKNOWN
DSP	Striate palmoplantar keratoderma	ORPHA:50942	UNKNOWN
DSPP	Deafness, autosomal dominant 39, with dentinogenesis imperfecta 1	OMIM:605594	MENDELIAN
DSPP	Dentinogenesis imperfecta, shields type III	OMIM:125500	MENDELIAN
DSPP	Dentin dysplasia, type II	OMIM:125420	MENDELIAN
DSPP	Dentinogenesis imperfecta 1	OMIM:125490	MENDELIAN
DST	Lethal congenital contracture syndrome 12	OMIM:621511	MENDELIAN
DST	Congenital myopathy 29 with contractures	OMIM:621510	MENDELIAN
DST	Epidermolysis bullosa simplex, autosomal recessive 2	OMIM:615425	MENDELIAN
DST	Neuropathy, hereditary sensory and autonomic, type VI	OMIM:614653	MENDELIAN
DSTYK	Spastic paraplegia 23	OMIM:270750	MENDELIAN
DSTYK	Congenital anomalies of kidney and urinary tract, susceptibility to	OMIM:610805	MENDELIAN
DSTYK	Autosomal recessive spastic paraplegia type 23	ORPHA:101003	UNKNOWN
DTHD1	—		UNKNOWN
DTNA	Left ventricular noncompaction 1	OMIM:604169	MENDELIAN
DTNA	Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis 2	OMIM:620971	MENDELIAN
DTNB	—		UNKNOWN
DTNBP1	Hermansky-Pudlak syndrome 7	OMIM:614076	MENDELIAN
DUOX1	—		UNKNOWN
DUOX2	Thyroid dysmorphogenesis 6	OMIM:607200	MENDELIAN
DUOX2	Genetic transient congenital hypothyroidism	ORPHA:226316	UNKNOWN
DUOX2	Familial thyroid dysmorphogenesis	ORPHA:95716	UNKNOWN
DUOXA2	Thyroid morphogenesis, genetic defect in, 5	OMIM:274900	MENDELIAN
DUOXA2	Familial thyroid dysmorphogenesis	ORPHA:95716	UNKNOWN
DUSP15	—		UNKNOWN
DUSP22	—		UNKNOWN
DUSP23	—		UNKNOWN
DUSP3	—		UNKNOWN
DUSP6	Hypogonadotropic hypogonadism 19 with or without anosmia	OMIM:615269	MENDELIAN
DUSP6	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
DUSP6	Kallmann syndrome	ORPHA:478	UNKNOWN
DUSP6	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
DUT	Bone marrow failure and diabetes mellitus syndrome	OMIM:620044	MENDELIAN
DUX4	Facioscapulohumeral dystrophy	ORPHA:269	UNKNOWN
DUX4L3	—		UNKNOWN
DVL1	Robinow syndrome, autosomal dominant 1	OMIM:180700	MENDELIAN
DVL1	Robinow syndrome, autosomal dominant 2	OMIM:616331	MENDELIAN
DVL1	Autosomal dominant Robinow syndrome	ORPHA:3107	UNKNOWN
DVL2	—		UNKNOWN
DXO	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
DYM	Smith-Mccort dysplasia 1	OMIM:607326	MENDELIAN
DYM	Dyggve-Melchior-Clausen disease	OMIM:223800	MENDELIAN
DYM	Dyggve-Melchior-Clausen disease	ORPHA:239	UNKNOWN
DYNAP	—		UNKNOWN
DYNC1H1	Charcot-marie-tooth disease, axonal, type 2O	OMIM:614228	MENDELIAN
DYNC1H1	Spinal muscular atrophy, lower extremity-predominant, 1, autosomal dominant	OMIM:158600	MENDELIAN
DYNC1H1	Cortical dysplasia, complex, with other brain malformations 13	OMIM:614563	MENDELIAN
DYNC1I1	—		UNKNOWN
DYNC2H1	Asphyxiating thoracic dystrophy 3	OMIM:613091	MENDELIAN
DYNC2H1	Short rib-polydactyly syndrome, Verma-Naumoff type	ORPHA:93271	UNKNOWN
DYNC2H1	Jeune syndrome	ORPHA:474	UNKNOWN
DYRK1A	Intellectual developmental disorder, autosomal dominant 7	OMIM:614104	MENDELIAN
DYRK1A	Intellectual disability syndrome due to a DYRK1A point mutation	ORPHA:464311	UNKNOWN
DYRK1A	DYRK1A-related intellectual disability syndrome due to 21q22.13q22.2 microdeletion	ORPHA:268261	UNKNOWN
DYRK1B	Abdominal obesity-metabolic syndrome 3	OMIM:615812	MENDELIAN
DYSF	Miyoshi muscular dystrophy 1	OMIM:254130	MENDELIAN
DYSF	Muscular dystrophy, limb-girdle, type 2B	OMIM:253601	MENDELIAN
DYSF	Myopathy, distal, with anterior tibial onset	OMIM:606768	MENDELIAN
DYSF	Distal myopathy with anterior tibial onset	ORPHA:178400	UNKNOWN
DYSF	Dysferlin-related limb-girdle muscular dystrophy R2	ORPHA:268	UNKNOWN
DYSF	Miyoshi myopathy	ORPHA:45448	UNKNOWN
DYX1C1	—		UNKNOWN
DYX1C1-CCPG1	—		UNKNOWN
E2F1	—		UNKNOWN
E2F3	—		UNKNOWN
E2F4	—		UNKNOWN
E2F5	—		UNKNOWN
EARS2	Combined oxidative phosphorylation deficiency 12	OMIM:614924	MENDELIAN
EBAG9	—		UNKNOWN
EBF3	Hypotonia, ataxia, and delayed development syndrome	OMIM:617330	MENDELIAN
EBF3	Distal deletion 10q syndrome	ORPHA:96148	UNKNOWN
EBF4	—		UNKNOWN
EBP	Mend syndrome	OMIM:300960	MENDELIAN
EBP	Chondrodysplasia punctata 2, X-linked dominant	OMIM:302960	MENDELIAN
EBP	MEND syndrome	ORPHA:401973	UNKNOWN
EBP	X-linked dominant chondrodysplasia punctata	ORPHA:35173	UNKNOWN
ECE1	Hypertension, essential	OMIM:145500	POLYGENIC
ECE1	Hirschsprung disease, cardiac defects, and autonomic dysfunction	OMIM:613870	MENDELIAN
ECE1	Hirschsprung disease	ORPHA:388	UNKNOWN
ECE2	—		UNKNOWN
ECEL1	Arthrogryposis, distal, type 5D	OMIM:615065	MENDELIAN
ECHS1	Mitochondrial short-chain enoyl-coa hydratase 1 deficiency	OMIM:616277	MENDELIAN
ECI1	—		UNKNOWN
ECM1	Urbach-Wiethe disease	OMIM:247100	MENDELIAN
ECM1	Lipoid proteinosis	ORPHA:530	UNKNOWN
ECM2	—		UNKNOWN
ECSIT	—		UNKNOWN
EDA	Ectodermal dysplasia 1, hypohidrotic, X-linked	OMIM:305100	MENDELIAN
EDA	Tooth agenesis, selective, X-linked, 1	OMIM:313500	MENDELIAN
EDA	X-linked hypohidrotic ectodermal dysplasia	ORPHA:181	UNKNOWN
EDA	Oligodontia	ORPHA:99798	UNKNOWN
EDA2R	X-linked hypohidrotic ectodermal dysplasia	ORPHA:181	UNKNOWN
EDAR	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant	OMIM:129490	MENDELIAN
EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive	OMIM:224900	MENDELIAN
EDAR	Autosomal recessive hypohidrotic ectodermal dysplasia	ORPHA:248	UNKNOWN
EDAR	Autosomal dominant hypohidrotic ectodermal dysplasia	ORPHA:1810	UNKNOWN
EDARADD	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant	OMIM:129490	MENDELIAN
EDARADD	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive	OMIM:224900	MENDELIAN
EDARADD	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant	OMIM:614940	MENDELIAN
EDARADD	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive	OMIM:614941	MENDELIAN
EDARADD	Autosomal recessive hypohidrotic ectodermal dysplasia	ORPHA:248	UNKNOWN
EDARADD	Autosomal dominant hypohidrotic ectodermal dysplasia	ORPHA:1810	UNKNOWN
EDARADD	Oligodontia	ORPHA:99798	UNKNOWN
EDN1	Question mark ears, isolated	OMIM:612798	MENDELIAN
EDN1	Auriculocondylar syndrome 3	OMIM:615706	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
EDN1	Auriculocondylar syndrome	ORPHA:137888	UNKNOWN
EDN2	—		UNKNOWN
EDN3	Waardenburg syndrome, type 4B	OMIM:613265	MENDELIAN
EDN3	Hirschsprung disease, susceptibility to, 4	OMIM:613712	POLYGENIC
EDN3	Hirschsprung disease	ORPHA:388	UNKNOWN
EDN3	Waardenburg-Shah syndrome	ORPHA:897	UNKNOWN
EDN3	Congenital central hypoventilation syndrome	ORPHA:661	UNKNOWN
EDNRA	Mandibulofacial dysostosis with alopecia	OMIM:616367	MENDELIAN
EDNRA	Migraine with or without aura, susceptibility to, 1	OMIM:157300	POLYGENIC
EDNRA	Cystic fibrosis	ORPHA:586	UNKNOWN
EDNRB	Hirschsprung disease, susceptibility to, 2	OMIM:600155	POLYGENIC
EDNRB	Waardenburg-Shah syndrome	OMIM:277580	MENDELIAN
EDNRB	ABCD syndrome	OMIM:600501	MENDELIAN
EDNRB	Hirschsprung disease	ORPHA:388	UNKNOWN
EDNRB	Waardenburg-Shah syndrome	ORPHA:897	UNKNOWN
EDNRB	Waardenburg syndrome type 2	ORPHA:895	UNKNOWN
EED	Cohen-Gibson syndrome	OMIM:617561	MENDELIAN
EEF1A2	Developmental and epileptic encephalopathy 33	OMIM:616409	MENDELIAN
EEF1A2	Intellectual developmental disorder, autosomal dominant 38	OMIM:616393	MENDELIAN
EEF1A2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
EEF2	Spinocerebellar ataxia 26	OMIM:609306	MENDELIAN
EEF2	Spinocerebellar ataxia type 26	ORPHA:101112	UNKNOWN
EEF2K	—		UNKNOWN
EFCAB13	—		UNKNOWN
EFCAB5	—		UNKNOWN
EFEMP1	Doyne honeycomb retinal dystrophy	OMIM:126600	MENDELIAN
EFEMP1	Glaucoma 1, open angle, H	OMIM:611276	MENDELIAN
EFEMP1	Cutis laxa, autosomal recessive, type ID	OMIM:620780	MENDELIAN
EFEMP1	Autosomal dominant drusen	ORPHA:75376	UNKNOWN
EFEMP1	Juvenile glaucoma	ORPHA:98977	UNKNOWN
EFEMP2	Cutis laxa, autosomal recessive, type IB	OMIM:614437	MENDELIAN
EFEMP2	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
EFEMP2	Autosomal recessive cutis laxa type 1	ORPHA:90349	UNKNOWN
EFHC1	Epilepsy, juvenile absence	OMIM:607631	POLYGENIC
EFHC1	Myoclonic epilepsy, juvenile, susceptibility to, 1	OMIM:254770	POLYGENIC
EFHC1	Juvenile myoclonic epilepsy	ORPHA:307	UNKNOWN
EFHC1	Juvenile absence epilepsy	ORPHA:1941	UNKNOWN
EFHC2	—		UNKNOWN
EFNA5	—		UNKNOWN
EFNB1	Craniofrontonasal syndrome	OMIM:304110	MENDELIAN
EFNB1	Craniofrontonasal dysplasia	ORPHA:1520	UNKNOWN
EFR3A	—		UNKNOWN
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type	OMIM:610536	MENDELIAN
EFTUD2	Mandibulofacial dysostosis-microcephaly syndrome	ORPHA:79113	UNKNOWN
EGF	Hypomagnesemia 4, renal	OMIM:611718	MENDELIAN
EGFL7	—		UNKNOWN
EGFR	Lung cancer, susceptibility to	OMIM:211980	MENDELIAN
EGFR	Inflammatory skin and bowel disease, neonatal, 2	OMIM:616069	MENDELIAN
EGLN1	Erythrocytosis, familial, 3	OMIM:609820	MENDELIAN
EGR2	Charcot-Marie-Tooth disease, demyelinating, type 1D	OMIM:607678	MENDELIAN
EGR2	Hypertrophic neuropathy of dejerine-sottas	OMIM:145900	MENDELIAN
EGR2	Neuropathy, congenital hypomyelinating, 1, autosomal recessive	OMIM:605253	MENDELIAN
EGR3	—		UNKNOWN
EHD2	—		UNKNOWN
EHHADH	Fanconi renotubular syndrome 3	OMIM:615605	MENDELIAN
EHHADH	Primary Fanconi renotubular syndrome	ORPHA:3337	UNKNOWN
EHMT1	Kleefstra syndrome 1	OMIM:610253	MENDELIAN
EHMT1	Kleefstra syndrome due to a point mutation	ORPHA:261652	UNKNOWN
EHMT1	Kleefstra syndrome due to 9q34 microdeletion	ORPHA:96147	UNKNOWN
EHMT2	—		UNKNOWN
EIF2AK3	Epiphyseal dysplasia, multiple, with early-onset diabetes mellitus	OMIM:226980	MENDELIAN
EIF2AK3	Wolcott-Rallison syndrome	ORPHA:1667	UNKNOWN
EIF2AK4	Pulmonary venoocclusive disease 2	OMIM:234810	MENDELIAN
EIF2AK4	Pulmonary capillary hemangiomatosis	ORPHA:199241	UNKNOWN
EIF2B1	Leukoencephalopathy with vanishing white matter	OMIM:603896	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
EIF2B2	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure	OMIM:620312	MENDELIAN
EIF2B3	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure	OMIM:620313	MENDELIAN
EIF2B4	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure	OMIM:620314	MENDELIAN
EIF2B5	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure	OMIM:620315	MENDELIAN
EIF2D	—		UNKNOWN
EIF2S3	Mehmo syndrome	OMIM:300148	MENDELIAN
EIF2S3	MEHMO syndrome	ORPHA:85282	UNKNOWN
EIF3G	—		UNKNOWN
EIF3H	—		UNKNOWN
EIF4A2	Neurodevelopmental disorder with hypotonia and speech delay, with or without seizures	OMIM:620455	MENDELIAN
EIF4A3	Robin sequence with cleft mandible and limb anomalies	OMIM:268305	MENDELIAN
EIF4A3	Richieri Costa-Pereira syndrome	ORPHA:3102	UNKNOWN
EIF4B	—		UNKNOWN
EIF4G1	Parkinson disease 18	OMIM:614251	POLYGENIC
EIF4G1	Hereditary late-onset Parkinson disease	ORPHA:411602	UNKNOWN
EIF4H	Williams syndrome	ORPHA:904	UNKNOWN
ELAC2	Combined oxidative phosphorylation deficiency 17	OMIM:615440	MENDELIAN
ELANE	Cyclic neutropenia	OMIM:162800	MENDELIAN
ELANE	Neutropenia, severe congenital, 1, autosomal dominant	OMIM:202700	MENDELIAN
ELANE	Cyclic neutropenia	ORPHA:2686	UNKNOWN
ELANE	Autosomal dominant severe congenital neutropenia	ORPHA:486	UNKNOWN
ELAVL2	—		UNKNOWN
ELAVL3	—		UNKNOWN
ELAVL4	—		UNKNOWN
ELF3	—		UNKNOWN
ELF4	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2	OMIM:301074	MENDELIAN
ELK1	—		UNKNOWN
ELK3	—		UNKNOWN
ELMOD2	—		UNKNOWN
ELMOD3	Deafness, autosomal recessive 88	OMIM:615429	MENDELIAN
ELMOD3	Deafness, autosomal dominant 81	OMIM:619500	MENDELIAN
ELN	Supravalvular aortic stenosis	OMIM:185500	MENDELIAN
ELN	Cutis laxa, autosomal dominant 1	OMIM:123700	MENDELIAN
ELN	Williams-Beuren syndrome	OMIM:194050	MENDELIAN
ELN	Supravalvular aortic stenosis	ORPHA:3193	UNKNOWN
ELN	Williams syndrome	ORPHA:904	UNKNOWN
ELN	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
ELN	Autosomal dominant cutis laxa	ORPHA:90348	UNKNOWN
ELOVL4	Spinocerebellar ataxia 34	OMIM:133190	MENDELIAN
ELOVL4	Stargardt disease 3	OMIM:600110	MENDELIAN
ELOVL4	Ichthyosis, spastic quadriplegia, and impaired intellectual development	OMIM:614457	MENDELIAN
ELOVL4	Stargardt disease	ORPHA:827	UNKNOWN
ELOVL4	Spinocerebellar ataxia type 34	ORPHA:1955	UNKNOWN
ELOVL5	Spinocerebellar ataxia 38	OMIM:615957	MENDELIAN
ELOVL5	Spinocerebellar ataxia type 38	ORPHA:423296	UNKNOWN
ELP2	Intellectual developmental disorder, autosomal recessive 58	OMIM:617270	MENDELIAN
ELP4	Aniridia 2	OMIM:617141	MENDELIAN
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation	OMIM:616875	MENDELIAN
EMC1	Global developmental delay-visual anomalies-progressive cerebellar atrophy-truncal hypotonia syndrome	ORPHA:480898	UNKNOWN
EMC4	—		UNKNOWN
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked	OMIM:310300	MENDELIAN
EMD	Cardiomyopathy, dilated, 3C	OMIM:301163	MENDELIAN
EMD	X-linked Emery-Dreifuss muscular dystrophy	ORPHA:98863	UNKNOWN
EME1	—		UNKNOWN
EME2	—		UNKNOWN
EMG1	Bowen-Conradi syndrome	OMIM:211180	MENDELIAN
EMG1	Bowen-Conradi syndrome	ORPHA:1270	UNKNOWN
EML1	Band heterotopia	OMIM:600348	MENDELIAN
EML4	—		UNKNOWN
EMP2	Familial idiopathic steroid-resistant nephrotic syndrome	OMIM:615861	MENDELIAN
EMP2	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
EMX1	—		UNKNOWN
EMX2	Schizencephaly	OMIM:269160	MENDELIAN
EMX2	Kallmann syndrome	ORPHA:478	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
EMX2	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
EN2	—		UNKNOWN
ENAM	Amelogenesis imperfecta, type IB	OMIM:104500	MENDELIAN
ENAM	Amelogenesis imperfecta, type IC	OMIM:204650	MENDELIAN
ENDOV	—		UNKNOWN
ENG	Telangiectasia, hereditary hemorrhagic, type 1	OMIM:187300	MENDELIAN
ENG	Generalized juvenile polyposis/juvenile polyposis coli	ORPHA:329971	UNKNOWN
ENG	Hereditary hemorrhagic telangiectasia	ORPHA:774	UNKNOWN
ENG	Familial cerebral saccular aneurysm	ORPHA:231160	UNKNOWN
ENO1	—		UNKNOWN
ENO3	Glycogen storage disease XIII	OMIM:612932	MENDELIAN
ENO3	Glycogen storage disease due to muscle beta-enolase deficiency	ORPHA:99849	UNKNOWN
ENOX1	—		UNKNOWN
ENPP1	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
ENPP1	Cole disease	OMIM:615522	MENDELIAN
ENPP1	Hypophosphatemic rickets, autosomal recessive, 2	OMIM:613312	MENDELIAN
ENPP1	Arterial calcification, generalized, of infancy, 1	OMIM:208000	MENDELIAN
ENPP1	Obesity	OMIM:601665	POLYGENIC
ENPP1	Pseudoxanthoma elasticum	ORPHA:758	UNKNOWN
ENPP1	Autosomal recessive hypophosphatemic rickets	ORPHA:289176	UNKNOWN
ENPP1	Generalized arterial calcification of infancy	ORPHA:51608	UNKNOWN
ENSA	—		UNKNOWN
ENTPD1	Spastic paraplegia 64, autosomal recessive	OMIM:615683	MENDELIAN
ENTPD1	Autosomal recessive spastic paraplegia type 64	ORPHA:401810	UNKNOWN
EOGT	Adams-Oliver syndrome 4	OMIM:615297	MENDELIAN
EOGT	Adams-Oliver syndrome	ORPHA:974	UNKNOWN
EOMES	Microcephaly-polymicrogyria-corpus callosum agenesis syndrome	ORPHA:171703	UNKNOWN
EP300	Rubinstein-Taybi syndrome 1	OMIM:180849	MENDELIAN
EP300	Menke-Hennekam syndrome 2	OMIM:618333	MENDELIAN
EP300	Colorectal cancer	OMIM:114500	MENDELIAN
EP300	Rubinstein-Taybi syndrome 2	OMIM:613684	MENDELIAN
EP300	Rubinstein-Taybi syndrome due to EP300 haploinsufficiency	ORPHA:353284	UNKNOWN
EPAS1	Erythrocytosis, familial, 4	OMIM:611783	MENDELIAN
EPAS1	Sporadic pheochromocytoma/secretory paraganglioma	ORPHA:276621	UNKNOWN
EPB41	Elliptocytosis 1	OMIM:611804	MENDELIAN
EPB41	Hereditary elliptocytosis	ORPHA:288	UNKNOWN
EPB41L1	Chromosome 20q11-q12 deletion syndrome	OMIM:614257	MENDELIAN
EPB41L2	—		UNKNOWN
EPB41L4A	—		UNKNOWN
EPB42	Spherocytosis, type 5	OMIM:612690	MENDELIAN
EPB42	Hereditary spherocytosis	ORPHA:822	UNKNOWN
EPC2	—		UNKNOWN
EPCAM	Diarrhea 5, with tufting enteropathy, congenital	OMIM:613217	MENDELIAN
EPCAM	Colorectal cancer, hereditary nonpolyposis, type 8	OMIM:613244	MENDELIAN
EPCAM	Lynch syndrome	ORPHA:144	UNKNOWN
EPCAM	Congenital tufting enteropathy	ORPHA:92050	UNKNOWN
EPG5	Neurodevelopmental disorder with parkinsonism or other movement abnormalities	OMIM:621506	MENDELIAN
EPG5	Vici syndrome	OMIM:242840	MENDELIAN
EPG5	Vici syndrome	ORPHA:1493	UNKNOWN
EPHA2	Cataract, posterior polar, 1 ctpa cataract, congenital total, included	OMIM:116600	MENDELIAN
EPHA3	—		UNKNOWN
EPHA4	—		UNKNOWN
EPHA5	—		UNKNOWN
EPHA6	—		UNKNOWN
EPHA7	—		UNKNOWN
EPHB1	—		UNKNOWN
EPHB2	Prostate cancer/brain cancer susceptibility	OMIM:603688	POLYGENIC
EPHB2	Bleeding disorder, platelet-type, 22	OMIM:618462	MENDELIAN
EPHB4	Capillary malformation-arteriovenous malformation 2	OMIM:618196	MENDELIAN
EPHB4	Lymphatic malformation 7	OMIM:617300	MENDELIAN
EPHB4	Vein of Galen malformation	ORPHA:1053	UNKNOWN
EPHB4	Meige disease	ORPHA:90186	UNKNOWN
EPHB6	—		UNKNOWN
EPHX2	Hypercholesterolemia, familial, 1	OMIM:143890	POLYGENIC
EPM2A	Myoclonic epilepsy of Lafora	OMIM:254780	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
EPM2A	Lafora disease	ORPHA:501	UNKNOWN
EPO	Erythrocytosis, familial, 5	OMIM:617907	MENDELIAN
EPO	Diamond-Blackfan anemia-like	OMIM:617911	MENDELIAN
EPOR	Erythrocytosis, familial, 1	OMIM:133100	MENDELIAN
EPOR	Primary familial and congenital erythrocytosis	ORPHA:90042	UNKNOWN
EPS8	Deafness, autosomal recessive 102	OMIM:615974	MENDELIAN
EPX	Eosinophil peroxidase deficiency	OMIM:261500	MENDELIAN
ERAP1	Behçet disease	ORPHA:117	UNKNOWN
ERAP2	—		UNKNOWN
ERBB2	Glioma susceptibility 1	OMIM:137800	MENDELIAN
ERBB2	Visceral neuropathy, familial, 2, autosomal recessive	OMIM:619465	MENDELIAN
ERBB2	Ovarian cancer	OMIM:167000	MENDELIAN
ERBB2	Lung cancer, susceptibility to	OMIM:211980	MENDELIAN
ERBB2	Gastric cancer, somatic	OMIM:613659	MENDELIAN
ERBB2	Hirschsprung disease	ORPHA:388	UNKNOWN
ERBB2	Adenocarcinoma of the oesophagus and oesophagogastric junction	ORPHA:99976	UNKNOWN
ERBB3	Visceral neuropathy, familial, autosomal recessive	OMIM:243180	MENDELIAN
ERBB3	Erythroleukemia, familial, susceptibility to	OMIM:133180	POLYGENIC
ERBB3	Lethal congenital contracture syndrome 2	OMIM:607598	MENDELIAN
ERBB3	Hirschsprung disease	ORPHA:388	UNKNOWN
ERBB4	Amyotrophic lateral sclerosis 19	OMIM:615515	MENDELIAN
ERBB4	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
ERCC1	Cerebrooculofacioskeletal syndrome 4	OMIM:610758	MENDELIAN
ERCC1	COFS syndrome	ORPHA:1466	UNKNOWN
ERCC1	Cockayne syndrome type 2	ORPHA:90322	UNKNOWN
ERCC2	Cerebrooculofacioskeletal syndrome 2	OMIM:610756	MENDELIAN
ERCC2	Xeroderma pigmentosum, complementation group D	OMIM:278730	MENDELIAN
ERCC2	Trichothiodystrophy 1, photosensitive	OMIM:601675	MENDELIAN
ERCC2	COFS syndrome	ORPHA:1466	UNKNOWN
ERCC2	Trichothiodystrophy	ORPHA:33364	UNKNOWN
ERCC2	Xeroderma pigmentosum-Cockayne syndrome complex	ORPHA:220295	UNKNOWN
ERCC2	Xeroderma pigmentosum	ORPHA:910	UNKNOWN
ERCC3	Trichothiodystrophy 2, photosensitive	OMIM:616390	MENDELIAN
ERCC3	Xeroderma pigmentosum, complementation group B	OMIM:610651	MENDELIAN
ERCC3	Trichothiodystrophy	ORPHA:33364	UNKNOWN
ERCC3	Xeroderma pigmentosum-Cockayne syndrome complex	ORPHA:220295	UNKNOWN
ERCC3	Xeroderma pigmentosum	ORPHA:910	UNKNOWN
ERCC4	Fanconi anemia, complementation group Q	OMIM:615272	MENDELIAN
ERCC4	XFE progeroid syndrome	OMIM:610965	MENDELIAN
ERCC4	Xeroderma pigmentosum, complementation group F	OMIM:278760	MENDELIAN
ERCC4	Fanconi anemia	ORPHA:84	UNKNOWN
ERCC4	Xeroderma pigmentosum-Cockayne syndrome complex	ORPHA:220295	UNKNOWN
ERCC4	Xeroderma pigmentosum	ORPHA:910	UNKNOWN
ERCC4	Cockayne syndrome type 1	ORPHA:90321	UNKNOWN
ERCC5	Cerebrooculofacioskeletal syndrome 3	OMIM:616570	MENDELIAN
ERCC5	Xeroderma pigmentosum, complementation group G	OMIM:278780	MENDELIAN
ERCC5	COFS syndrome	ORPHA:1466	UNKNOWN
ERCC5	Xeroderma pigmentosum-Cockayne syndrome complex	ORPHA:220295	UNKNOWN
ERCC5	Xeroderma pigmentosum	ORPHA:910	UNKNOWN
ERCC6	Premature ovarian failure 11	OMIM:616946	MENDELIAN
ERCC6	Cockayne syndrome, type B	OMIM:133540	MENDELIAN
ERCC6	De Sanctis-Cacchione syndrome	OMIM:278800	MENDELIAN
ERCC6	UV-sensitive syndrome 1	OMIM:600630	MENDELIAN
ERCC6	Lung cancer, susceptibility to	OMIM:211980	POLYGENIC
ERCC6	Cerebrooculofacioskeletal syndrome 1	OMIM:214150	MENDELIAN
ERCC6	COFS syndrome	ORPHA:1466	UNKNOWN
ERCC6	Cockayne syndrome type 3	ORPHA:90324	UNKNOWN
ERCC6	Cockayne syndrome type 1	ORPHA:90321	UNKNOWN
ERCC6	Cockayne syndrome type 2	ORPHA:90322	UNKNOWN
ERCC6L2	Bone marrow failure syndrome 2	OMIM:615715	MENDELIAN
ERCC8	Cockayne syndrome, type A	OMIM:216400	MENDELIAN
ERCC8	UV-sensitive syndrome 2	OMIM:614621	MENDELIAN
ERCC8	Cockayne syndrome type 3	ORPHA:90324	UNKNOWN
ERCC8	Cockayne syndrome type 1	ORPHA:90321	UNKNOWN
ERCC8	Cockayne syndrome type 2	ORPHA:90322	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
ERF	Craniosynostosis 4	OMIM:600775	MENDELIAN
ERF	Chitayat syndrome	OMIM:617180	MENDELIAN
ERG	Lymphatic malformation 14	OMIM:620602	MENDELIAN
ERLIN1	Spastic paraplegia 62, autosomal recessive	OMIM:615681	MENDELIAN
ERLIN1	Autosomal recessive spastic paraplegia type 62	ORPHA:401785	UNKNOWN
ERLIN1	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
ERLIN2	Spastic paraplegia 18A, autosomal dominant	OMIM:620512	MENDELIAN
ERLIN2	Spastic paraplegia 18, autosomal recessive	OMIM:611225	MENDELIAN
ERLIN2	Autosomal spastic paraplegia type 18	ORPHA:209951	UNKNOWN
ERLIN2	Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome	ORPHA:280384	UNKNOWN
ERLIN2	Juvenile primary lateral sclerosis	ORPHA:247604	UNKNOWN
ERMARD	Periventricular nodular heterotopia 6	OMIM:615544	MENDELIAN
ERMARD	6q terminal deletion syndrome	ORPHA:75857	UNKNOWN
ERMARD	Periventricular nodular heterotopia	ORPHA:98892	UNKNOWN
ERRFI1	—		UNKNOWN
ESAM	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity	OMIM:620371	MENDELIAN
ESCO2	Roberts-SC phocomelia syndrome	OMIM:268300	MENDELIAN
ESCO2	Cleft lip/palate with abnormal thumbs and microcephaly	OMIM:216100	MENDELIAN
ESCO2	Roberts syndrome	ORPHA:3103	UNKNOWN
ESCO2	Juberg-Hayward syndrome	ORPHA:2319	UNKNOWN
ESPL1	—		UNKNOWN
ESPN	Deafness, autosomal recessive 36, with or without vestibular involvement	OMIM:609006	MENDELIAN
ESPN	Deafness, autosomal dominant 91	OMIM:621556	MENDELIAN
ESPN	Usher syndrome, type 1M	OMIM:618632	MENDELIAN
ESPN	Usher syndrome type 1	ORPHA:231169	UNKNOWN
ESR1	Migraine with or without aura, susceptibility to, 1	OMIM:157300	POLYGENIC
ESR1	Estrogen resistance	OMIM:615363	MENDELIAN
ESR1	Breast cancer	OMIM:114480	MENDELIAN
ESR1	Estrogen resistance syndrome	ORPHA:785	UNKNOWN
ESR2	Ovarian dysgenesis 8	OMIM:618187	MENDELIAN
ESRRB	Deafness, autosomal recessive 35	OMIM:608565	MENDELIAN
ESX1	—		UNKNOWN
ETFA	Multiple acyl-CoA dehydrogenation deficiency	OMIM:231680	MENDELIAN
ETFB	Multiple acyl-CoA dehydrogenation deficiency	OMIM:231680	MENDELIAN
ETFDH	Multiple acyl-CoA dehydrogenation deficiency	OMIM:231680	MENDELIAN
ETHE1	Encephalopathy, ethylmalonic	OMIM:602473	MENDELIAN
ETHE1	Ethylmalonic encephalopathy	ORPHA:51188	UNKNOWN
ETNPPL	—		UNKNOWN
ETS1	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
ETV5	—		UNKNOWN
ETV6	Thrombocytopenia 5	OMIM:616216	MENDELIAN
ETV6	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
EVC	Ellis-Van creveld syndrome	OMIM:225500	MENDELIAN
EVC	Weyers acrofacial dysostosis	OMIM:193530	MENDELIAN
EVC	Ellis-Van Creveld syndrome	ORPHA:289	UNKNOWN
EVC	Acrofacial dysostosis, Weyers type	ORPHA:952	UNKNOWN
EVC2	Ellis-Van creveld syndrome	OMIM:225500	MENDELIAN
EVC2	Weyers acrofacial dysostosis	OMIM:193530	MENDELIAN
EVC2	Ellis-Van Creveld syndrome	ORPHA:289	UNKNOWN
EVC2	Acrofacial dysostosis, Weyers type	ORPHA:952	UNKNOWN
EVI5	—		UNKNOWN
EWSR1	Ewing sarcoma	OMIM:612219	MENDELIAN
EWSR1	Desmoplastic small round cell tumor	ORPHA:83469	UNKNOWN
EXO1	—		UNKNOWN
EXO5	—		UNKNOWN
EXOC1	—		UNKNOWN
EXOC4	—		UNKNOWN
EXOC6B	Spondyloepimetaphyseal dysplasia with joint laxity, type 3	OMIM:618395	MENDELIAN
EXOC8	Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy	OMIM:619076	MENDELIAN
EXOSC3	Pontocerebellar hypoplasia, type 1B	OMIM:614678	MENDELIAN
EXOSC3	Pontocerebellar hypoplasia type 1	ORPHA:2254	UNKNOWN
EXOSC5	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects	OMIM:619576	MENDELIAN
EXOSC8	Pontocerebellar hypoplasia, type 1C	OMIM:616081	MENDELIAN
EXOSC8	Pontocerebellar hypoplasia type 1	ORPHA:2254	UNKNOWN
EXPH5	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive	OMIM:615028	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
EXT1	Chondrosarcoma	OMIM:215300	MENDELIAN
EXT1	Exostoses, multiple, type I	OMIM:133700	MENDELIAN
EXT1	Multiple osteochondromas	ORPHA:321	UNKNOWN
EXT1	Trichorhinophalangeal syndrome type 2	ORPHA:502	UNKNOWN
EXT2	Seizures, scoliosis, and macrocephaly/microcephaly syndrome	OMIM:616682	MENDELIAN
EXT2	Exostoses, multiple, type II	OMIM:133701	MENDELIAN
EXT2	Multiple osteochondromas	ORPHA:321	UNKNOWN
EXT2	Potocki-Shaffer syndrome	ORPHA:52022	UNKNOWN
EXT2	Seizures-scoliosis-macrocephaly syndrome	ORPHA:466926	UNKNOWN
EXTL1	—		UNKNOWN
EXTL2	—		UNKNOWN
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities	OMIM:617425	MENDELIAN
EXTL3	Skeletal dysplasia-T-cell immunodeficiency-developmental delay syndrome	ORPHA:508533	UNKNOWN
EYA1	Otofaciocervical syndrome	OMIM:166780	MENDELIAN
EYA1	Branchiootic syndrome 1	OMIM:602588	MENDELIAN
EYA1	Branchiootorenal syndrome 1	OMIM:113650	MENDELIAN
EYA1	Branchiootic syndrome	ORPHA:52429	UNKNOWN
EYA1	BOR syndrome	ORPHA:107	UNKNOWN
EYA4	Deafness, autosomal dominant nonsyndromic sensorineural 10	OMIM:601316	MENDELIAN
EYA4	Cardiomyopathy, dilated, 1J	OMIM:605362	MENDELIAN
EYA4	Sensorineural deafness with dilated cardiomyopathy	ORPHA:217622	UNKNOWN
EYS	Retinitis pigmentosa 25	OMIM:602772	MENDELIAN
EYS	Retinitis pigmentosa	ORPHA:791	UNKNOWN
EZH2	Weaver syndrome	OMIM:277590	MENDELIAN
EZH2	Weaver syndrome	ORPHA:3447	UNKNOWN
F10	Factor X deficiency	OMIM:227600	MENDELIAN
F10	Congenital factor X deficiency	ORPHA:328	UNKNOWN
F11	Factor XI deficiency	OMIM:612416	MENDELIAN
F11	Congenital factor XI deficiency	ORPHA:329	UNKNOWN
F12	Angioedema, hereditary, 3	OMIM:610618	MENDELIAN
F12	Factor XII deficiency	OMIM:234000	MENDELIAN
F12	Congenital factor XII deficiency	ORPHA:330	UNKNOWN
F13A1	Thrombophiliavenous thromboembolism, included	OMIM:188050	POLYGENIC
F13A1	Factor XIII, A subunit, deficiency of	OMIM:613225	MENDELIAN
F13A1	Congenital factor XIII deficiency	ORPHA:331	UNKNOWN
F13B	Factor XIIB deficiency	OMIM:613235	MENDELIAN
F13B	Congenital factor XIII deficiency	ORPHA:331	UNKNOWN
F2	Thrombophiliavenous thromboembolism, included	OMIM:188050	MENDELIAN
F2	Pregnancy loss, recurrent, susceptibility to, 2	OMIM:614390	POLYGENIC
F2	Ischemic stroke, susceptibility to	OMIM:601367	POLYGENIC
F2	Congenital prothrombin deficiency	OMIM:613679	MENDELIAN
F2	Congenital factor II deficiency	ORPHA:325	UNKNOWN
F2R	—		UNKNOWN
F2RL1	—		UNKNOWN
F3	—		UNKNOWN
F5	Thrombophilia due to deficiency of activated protein C cofactor	OMIM:188055	MENDELIAN
F5	Budd-Chiari syndrome	OMIM:600880	POLYGENIC
F5	Pregnancy loss, recurrent, susceptibility to, 1	OMIM:614389	POLYGENIC
F5	Ischemic stroke, susceptibility to	OMIM:601367	POLYGENIC
F5	Factor V deficiency	OMIM:227400	MENDELIAN
F5	Congenital factor V deficiency	ORPHA:326	UNKNOWN
F5	Budd-Chiari syndrome	ORPHA:131	UNKNOWN
F7	Factor VII deficiency	OMIM:227500	MENDELIAN
F7	Congenital factor VII deficiency	ORPHA:327	UNKNOWN
F8	Thrombophilia 13, X-linked, due to factor VIII defect	OMIM:301071	MENDELIAN
F8	Hemophilia A	OMIM:306700	MENDELIAN
F8	Bleeding disorder in hemophilia A carriers	ORPHA:177926	UNKNOWN
F8	Moderate hemophilia A	ORPHA:169805	UNKNOWN
F8	Severe hemophilia A	ORPHA:169802	UNKNOWN
F9	Hemophilia B	OMIM:306900	MENDELIAN
F9	Thrombophilia, X-linked, due to factor IX defect	OMIM:300807	MENDELIAN
FA2H	Spastic paraplegia 35, autosomal recessive	OMIM:612319	MENDELIAN
FA2H	Autosomal recessive spastic paraplegia type 35	ORPHA:171629	UNKNOWN
FA2H	Fatty acid hydroxylase-associated neurodegeneration	ORPHA:329308	UNKNOWN
FAAH2	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
FABP1	—		UNKNOWN
FABP2	—		UNKNOWN
FABP3	—		UNKNOWN
FABP4	—		UNKNOWN
FABP6	—		UNKNOWN
FABP7	—		UNKNOWN
FADD	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations	OMIM:613759	MENDELIAN
FADD	FADD-related immunodeficiency	ORPHA:306550	UNKNOWN
FADS2	—		UNKNOWN
FAH	Tyrosinemia, type I	OMIM:276700	MENDELIAN
FAH	Tyrosinemia type 1	ORPHA:882	UNKNOWN
FAM104A	—		UNKNOWN
FAM111A	Kenny-caffey syndrome, type 2	OMIM:127000	MENDELIAN
FAM111A	Gracile bone dysplasia	OMIM:602361	MENDELIAN
FAM111A	Autosomal dominant Kenny-Caffey syndrome	ORPHA:93325	UNKNOWN
FAM111B	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis	OMIM:615704	MENDELIAN
FAM120A	—		UNKNOWN
FAM120AOS	—		UNKNOWN
FAM120B	—		UNKNOWN
FAM123B	—		UNKNOWN
FAM126A	—		UNKNOWN
FAM134B	—		UNKNOWN
FAM136A	—		UNKNOWN
FAM161A	Retinitis pigmentosa 28	OMIM:606068	MENDELIAN
FAM161A	Retinitis pigmentosa	ORPHA:791	UNKNOWN
FAM175A	—		UNKNOWN
FAM175B	—		UNKNOWN
FAM177A1	Neurodevelopmental disorder with white matter abnormalities and gait disturbance	OMIM:621152	MENDELIAN
FAM189B	—		UNKNOWN
FAM205A	—		UNKNOWN
FAM20A	Amelogenesis imperfecta, type IG (enamel-renal syndrome)	OMIM:204690	MENDELIAN
FAM20A	Enamel-renal syndrome	ORPHA:1031	UNKNOWN
FAM20C	Raine syndrome	OMIM:259775	MENDELIAN
FAM20C	Osteosclerotic bone dysplasia	ORPHA:1832	UNKNOWN
FAM3C	—		UNKNOWN
FAM46C	—		UNKNOWN
FAM47B	—		UNKNOWN
FAM47C	—		UNKNOWN
FAM58A	—		UNKNOWN
FAM63B	—		UNKNOWN
FAM64A	—		UNKNOWN
FAM65B	—		UNKNOWN
FAM71A	—		UNKNOWN
FAM81B	—		UNKNOWN
FAM83H	—		UNKNOWN
FAM8A1	—		UNKNOWN
FAM91A1	—		UNKNOWN
FAM9C	—		UNKNOWN
FAN1	Interstitial nephritis, karyomegalic	OMIM:614817	MENDELIAN
FANCA	Fanconi anemia	OMIM:227650	MENDELIAN
FANCA	Fanconi anemia	ORPHA:84	UNKNOWN
FANCB	Fanconi anemia, complementation group B	OMIM:300514	MENDELIAN
FANCB	Fanconi anemia	ORPHA:84	UNKNOWN
FANCB	VACTERL with hydrocephalus	ORPHA:3412	UNKNOWN
FANCC	Fanconi anemia, complementation group C	OMIM:227645	MENDELIAN
FANCC	Fanconi anemia	ORPHA:84	UNKNOWN
FANCD2	Fanconi anemia, complementation group D2	OMIM:227646	MENDELIAN
FANCD2	Fanconi anemia	ORPHA:84	UNKNOWN
FANCE	Fanconi anemia, complementation group E	OMIM:600901	MENDELIAN
FANCE	Fanconi anemia	ORPHA:84	UNKNOWN
FANCF	Fanconi anemia, complementation group F	OMIM:603467	MENDELIAN
FANCF	Fanconi anemia	ORPHA:84	UNKNOWN
FANCG	Fanconi anemia, complementation group G	OMIM:614082	MENDELIAN
FANCG	Fanconi anemia	ORPHA:84	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
FANCI	Fanconi anemia, complementation group I	OMIM:609053	MENDELIAN
FANCI	Fanconi anemia	ORPHA:84	UNKNOWN
FANCL	Fanconi anemia, complementation group L	OMIM:614083	MENDELIAN
FANCL	Fanconi anemia	ORPHA:84	UNKNOWN
FANCM	Spermatogenic failure 28	OMIM:618086	MENDELIAN
FANCM	Premature ovarian failure 15	OMIM:618096	MENDELIAN
FANCM	Fanconi anemia	ORPHA:84	UNKNOWN
FANCM	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
FAR1	Cataracts, spastic paraparesis, and speech delay	OMIM:619338	MENDELIAN
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder	OMIM:616154	MENDELIAN
FAR1	Fatty acyl-CoA reductase 1 deficiency	ORPHA:438178	UNKNOWN
FARS2	Spastic paraplegia 77, autosomal recessive	OMIM:617046	MENDELIAN
FARS2	Combined oxidative phosphorylation deficiency 14	OMIM:614946	MENDELIAN
FARS2	Autosomal recessive spastic paraplegia type 77	ORPHA:466722	UNKNOWN
FAS	Autoimmune lymphoproliferative syndrome	OMIM:601859	MENDELIAN
FAS	Autoimmune lymphoproliferative syndrome	ORPHA:3261	UNKNOWN
FAS	Behçet disease	ORPHA:117	UNKNOWN
FAS	Vogt-Koyanagi-Harada disease	ORPHA:3437	UNKNOWN
FASLG	Lung cancer, susceptibility to	OMIM:211980	POLYGENIC
FASLG	Autoimmune lymphoproliferative syndrome	OMIM:601859	MENDELIAN
FASLG	Autoimmune lymphoproliferative syndrome	ORPHA:3261	UNKNOWN
FASN	—		UNKNOWN
FASTKD2	Combined oxidative phosphorylation deficiency 44	OMIM:618855	MENDELIAN
FASTKD2	Mitochondrial complex IV deficiency, nuclear type 1	OMIM:220110	MENDELIAN
FAT1	—		UNKNOWN
FAT2	Spinocerebellar ataxia 45	OMIM:617769	MENDELIAN
FAT3	—		UNKNOWN
FAT4	Van maldergem syndrome 2	OMIM:615546	MENDELIAN
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2	OMIM:616006	MENDELIAN
FAT4	Cerebrofacioarticular syndrome	ORPHA:314679	UNKNOWN
FAT4	Hennekam syndrome	ORPHA:2136	UNKNOWN
FATE1	—		UNKNOWN
FBLIM1	—		UNKNOWN
FBLN1	FBLN1-related developmental delay-central nervous system anomaly-syndactyly syndrome	ORPHA:404451	UNKNOWN
FBLN2	—		UNKNOWN
FBLN5	Macular degeneration, age-related, 3	OMIM:608895	MENDELIAN
FBLN5	Cutis laxa, autosomal dominant 2	OMIM:614434	MENDELIAN
FBLN5	Charcot-Marie-Tooth disease, demyelinating, type 1H	OMIM:619764	MENDELIAN
FBLN5	Cutis laxa, autosomal recessive, type 1A	OMIM:219100	MENDELIAN
FBLN5	Autosomal dominant cutis laxa	ORPHA:90348	UNKNOWN
FBLN5	Autosomal recessive cutis laxa type 1	ORPHA:90349	UNKNOWN
FBN1	Mass syndrome	OMIM:604308	MENDELIAN
FBN1	Marfan syndrome	OMIM:154700	MENDELIAN
FBN1	Ectopia lentis, familial	OMIM:129600	MENDELIAN
FBN1	Marfan lipodystrophy syndrome	OMIM:616914	MENDELIAN
FBN1	Geleophysic dysplasia 2	OMIM:614185	MENDELIAN
FBN1	Stiff skin syndrome	OMIM:184900	MENDELIAN
FBN1	Weill-Marchesani syndrome 2, dominant	OMIM:608328	MENDELIAN
FBN1	Acromicric dysplasia	OMIM:102370	MENDELIAN
FBN1	Stiff skin syndrome	ORPHA:2833	UNKNOWN
FBN1	Acromicric dysplasia	ORPHA:969	UNKNOWN
FBN1	Geleophysic dysplasia	ORPHA:2623	UNKNOWN
FBN1	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
FBN1	Isolated ectopia lentis	ORPHA:1885	UNKNOWN
FBN1	Neonatal Marfan syndrome	ORPHA:284979	UNKNOWN
FBN1	Shprintzen-Goldberg syndrome	ORPHA:2462	UNKNOWN
FBN1	Glaucoma-ectopia lentis-microspherophakia-stiff joints-short stature syndrome	ORPHA:2084	UNKNOWN
FBN1	Weill-Marchesani syndrome	ORPHA:3449	UNKNOWN
FBN2	Contractural arachnodactyly, congenital	OMIM:121050	MENDELIAN
FBN2	Macular degeneration, early-onset	OMIM:616118	MENDELIAN
FBN2	Congenital contractural arachnodactyly	ORPHA:115	UNKNOWN
FBN3	—		UNKNOWN
FBP1	Fructose-1,6-bisphosphatase deficiency	OMIM:229700	MENDELIAN
FBP1	Fructose-1,6-bisphosphatase deficiency	ORPHA:348	UNKNOWN
FBRSL1	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	OMIM:615471	MENDELIAN
FBXL6	—		UNKNOWN
FBXO10	—		UNKNOWN
FBXO11	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities	OMIM:618089	MENDELIAN
FBXO15	—		UNKNOWN
FBXO18	—		UNKNOWN
FBXO25	—		UNKNOWN
FBXO28	Developmental and epileptic encephalopathy 100	OMIM:619777	MENDELIAN
FBXO28	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
FBXO31	Intellectual developmental disorder, autosomal recessive 45	OMIM:615979	MENDELIAN
FBXO38	Neuronopathy, distal hereditary motor, type IID	OMIM:615575	MENDELIAN
FBXO7	Parkinson disease 15	OMIM:260300	MENDELIAN
FBXO7	Parkinsonian-pyramidal syndrome	ORPHA:171695	UNKNOWN
FBXW11	Neurodevelopmental, jaw, eye, and digital syndrome	OMIM:618914	MENDELIAN
FBXW4	Isolated split hand-split foot malformation	ORPHA:2440	UNKNOWN
FBXW7	Developmental delay, hypotonia, and impaired language	OMIM:620012	MENDELIAN
FCAR	—		UNKNOWN
FCER1A	—		UNKNOWN
FCER1G	—		UNKNOWN
FCER2	—		UNKNOWN
FCGR1A	—		UNKNOWN
FCGR2A	Cystic fibrosis	OMIM:219700	POLYGENIC
FCGR2A	Systemic lupus erythematosus	OMIM:152700	POLYGENIC
FCGR2B	Systemic lupus erythematosus	OMIM:152700	POLYGENIC
FCGR2B	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
FCGR2C	Immune thrombocytopenia	ORPHA:3002	UNKNOWN
FCGR3A	Immunodeficiency 20	OMIM:615707	MENDELIAN
FCGR3B	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
FCGR3B	Neonatal alloimmune neutropenia	ORPHA:464370	UNKNOWN
FCGRT	—		UNKNOWN
FCN1	—		UNKNOWN
FCN2	—		UNKNOWN
FCN3	Immunodeficiency due to ficolin 3 deficiency	OMIM:613860	MENDELIAN
FCRL3	—		UNKNOWN
FCRL6	—		UNKNOWN
FDFT1	Squalene synthase deficiency	OMIM:618156	MENDELIAN
FDX1L	—		UNKNOWN
FECH	Protoporphyria, erythropoietic, 1	OMIM:177000	MENDELIAN
FECH	Autosomal erythropoietic protoporphyria	ORPHA:79278	UNKNOWN
FEM1A	—		UNKNOWN
FEM1B	Neurodevelopmental disorder with behavioral, ear, and skeletal abnormalities	OMIM:621263	MENDELIAN
FEN1	—		UNKNOWN
FERMT1	Kindler syndrome	OMIM:173650	MENDELIAN
FERMT1	Kindler epidermolysis bullosa	ORPHA:2908	UNKNOWN
FERMT3	Leukocyte adhesion deficiency, type III	OMIM:612840	MENDELIAN
FEV	—		UNKNOWN
FEZF1	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
FEZF1	Hypogonadotropic hypogonadism 22 with or without anosmia	OMIM:616030	MENDELIAN
FEZF1	Kallmann syndrome	ORPHA:478	UNKNOWN
FEZF2	—		UNKNOWN
FFAR1	—		UNKNOWN
FGA	Congenital dysfibrinogenemia	OMIM:616004	MENDELIAN
FGA	Afibrinogenemia, congenitalhypofibrinogenemia, congenital, included	OMIM:202400	MENDELIAN
FGA	Amyloidosis, familial visceral	OMIM:105200	MENDELIAN
FGA	Familial hypofibrinogenemia	ORPHA:101041	UNKNOWN
FGA	Familial afibrinogenemia	ORPHA:98880	UNKNOWN
FGA	Familial dysfibrinogenemia	ORPHA:98881	UNKNOWN
FGB	Congenital dysfibrinogenemia	OMIM:616004	MENDELIAN
FGB	Afibrinogenemia, congenitalhypofibrinogenemia, congenital, included	OMIM:202400	MENDELIAN
FGB	Familial hypofibrinogenemia	ORPHA:101041	UNKNOWN
FGB	Familial afibrinogenemia	ORPHA:98880	UNKNOWN
FGB	Familial dysfibrinogenemia	ORPHA:98881	UNKNOWN
FGD1	Aarskog-Scott syndrome	OMIM:305400	MENDELIAN
FGD1	Aarskog-Scott syndrome	ORPHA:915	UNKNOWN
FGD3	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
FGD4	Charcot-marie-tooth disease, type 4H	OMIM:609311	MENDELIAN
FGF1	—		UNKNOWN
FGF10	Aplasia of lacrimal and salivary glands	OMIM:180920	MENDELIAN
FGF10	Lacrimeoauriculodentodigital syndrome-3	OMIM:620193	MENDELIAN
FGF10	Lacrimeoauriculodentodigital syndrome	ORPHA:2363	UNKNOWN
FGF13	Developmental and epileptic encephalopathy 90	OMIM:301058	MENDELIAN
FGF13	Intellectual developmental disorder, X-linked 110	OMIM:301095	MENDELIAN
FGF13	Genetic epilepsy with febrile seizure plus	ORPHA:36387	UNKNOWN
FGF14	Spinocerebellar ataxia 27B, late-onset	OMIM:620174	MENDELIAN
FGF14	Nystagmus 4, congenital, autosomal dominant	OMIM:193003	MENDELIAN
FGF14	Spinocerebellar ataxia type 27A	ORPHA:98764	UNKNOWN
FGF16	Metacarpal 4-5 fusion	OMIM:309630	MENDELIAN
FGF17	Hypogonadotropic hypogonadism 20 with or without anosmia	OMIM:615270	MENDELIAN
FGF17	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
FGF17	Kallmann syndrome	ORPHA:478	UNKNOWN
FGF17	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
FGF19	—		UNKNOWN
FGF2	—		UNKNOWN
FGF20	Renal hypodysplasia/aplasia 2	OMIM:615721	MENDELIAN
FGF20	Renal agenesis, bilateral	ORPHA:1848	UNKNOWN
FGF22	—		UNKNOWN
FGF23	Tumoral calcinosis, hyperphosphatemic, familial, 2	OMIM:617993	MENDELIAN
FGF23	Hypophosphatemic rickets, autosomal dominant	OMIM:193100	MENDELIAN
FGF23	Autosomal dominant hypophosphatemic rickets	ORPHA:89937	UNKNOWN
FGF3	Deafness, congenital, with inner ear agenesis, microtia, and microdontia	OMIM:610706	MENDELIAN
FGF3	Otodental syndrome	ORPHA:2791	UNKNOWN
FGF3	Deafness with labyrinthine aplasia, microtia, and microdontia	ORPHA:90024	UNKNOWN
FGF4	Short-rib thoracic dysplasia 22 without polydactyly	OMIM:621260	MENDELIAN
FGF5	Trichomegaly	OMIM:190330	MENDELIAN
FGF6	—		UNKNOWN
FGF7	—		UNKNOWN
FGF8	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
FGF8	Hypogonadotropic hypogonadism 6 with or without anosmia	OMIM:612702	MENDELIAN
FGF8	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
FGF8	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
FGF8	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
FGF8	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
FGF8	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
FGF8	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
FGF8	Kallmann syndrome	ORPHA:478	UNKNOWN
FGF8	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
FGF9	Multiple synostoses syndrome 3	OMIM:612961	MENDELIAN
FGF9	Multiple synostoses syndrome	ORPHA:3237	UNKNOWN
FGFBP1	—		UNKNOWN
FGFR1	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
FGFR1	Encephalocraniocutaneous lipomatosis	OMIM:613001	MENDELIAN
FGFR1	Trigonocephaly 1	OMIM:190440	MENDELIAN
FGFR1	Hypogonadotropic hypogonadism 2 with or without anosmia	OMIM:147950	MENDELIAN
FGFR1	Hartsfield syndrome	OMIM:615465	MENDELIAN
FGFR1	Osteoglophonic dysplasia	OMIM:166250	MENDELIAN
FGFR1	Jackson-Weiss syndrome	OMIM:123150	MENDELIAN
FGFR1	Pfeiffer syndrome	OMIM:101600	MENDELIAN
FGFR1	Septo-optic dysplasia spectrum	ORPHA:3157	UNKNOWN
FGFR1	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
FGFR1	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
FGFR1	Non-syndromic metopic craniosynostosis	ORPHA:3366	UNKNOWN
FGFR1	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
FGFR1	Osteoglosphonic dysplasia	ORPHA:2645	UNKNOWN
FGFR1	Encephalocraniocutaneous lipomatosis	ORPHA:2396	UNKNOWN
FGFR1	Pfeiffer syndrome type 1	ORPHA:93258	UNKNOWN
FGFR1	Hartsfield syndrome	ORPHA:2117	UNKNOWN
FGFR1	Kallmann syndrome	ORPHA:478	UNKNOWN
FGFR1	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
FGFR1	Oligodontia	ORPHA:99798	UNKNOWN
FGFR1OP	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis	OMIM:207410	MENDELIAN
FGFR2	Crouzon syndrome	OMIM:123500	MENDELIAN
FGFR2	Beare-Stevenson cutis gyrata syndrome	OMIM:123790	MENDELIAN
FGFR2	Familial scaphocephaly syndrome, Mcgillivray type	OMIM:609579	MENDELIAN
FGFR2	Bent bone dysplasia syndrome	OMIM:614592	MENDELIAN
FGFR2	Apert syndrome	OMIM:101200	MENDELIAN
FGFR2	Saethre-Chotzen syndrome	OMIM:101400	MENDELIAN
FGFR2	Lacrimoauriculodentodigital syndrome-1	OMIM:149730	MENDELIAN
FGFR2	Gastric cancer, somatic	OMIM:613659	MENDELIAN
FGFR2	Jackson-Weiss syndrome	OMIM:123150	MENDELIAN
FGFR2	Pfeiffer syndrome	OMIM:101600	MENDELIAN
FGFR2	Apert syndrome	ORPHA:87	UNKNOWN
FGFR2	Crouzon syndrome	ORPHA:207	UNKNOWN
FGFR2	Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome	ORPHA:1555	UNKNOWN
FGFR2	Jackson-Weiss syndrome	ORPHA:1540	UNKNOWN
FGFR2	FGFR2-related bent bone dysplasia	ORPHA:313855	UNKNOWN
FGFR2	Familial scaphocephaly syndrome, McGillivray type	ORPHA:168624	UNKNOWN
FGFR2	Saethre-Chotzen syndrome	ORPHA:794	UNKNOWN
FGFR2	Pfeiffer syndrome type 3	ORPHA:93260	UNKNOWN
FGFR2	Pfeiffer syndrome type 1	ORPHA:93258	UNKNOWN
FGFR2	Pfeiffer syndrome type 2	ORPHA:93259	UNKNOWN
FGFR2	Lacrimoauriculodentodigital syndrome	ORPHA:2363	UNKNOWN
FGFR3	Hypochondroplasia	OMIM:146000	MENDELIAN
FGFR3	Epidermal nevus, somatic	OMIM:162900	MENDELIAN
FGFR3	Achondroplasia, severe, with developmental delay and acanthosis nigricans	OMIM:616482	MENDELIAN
FGFR3	Achondroplasia	OMIM:100800	MENDELIAN
FGFR3	Lacrimoauriculodentodigital syndrome-2	OMIM:620192	MENDELIAN
FGFR3	Testicular tumor, somatic	OMIM:273300	MENDELIAN
FGFR3	Camptodactyly, tall stature, and hearing loss syndrome	OMIM:610474	MENDELIAN
FGFR3	Cervical cancer	OMIM:603956	MENDELIAN
FGFR3	Bladder cancer	OMIM:109800	MENDELIAN
FGFR3	Thanatophoric dysplasia, type II	OMIM:187601	MENDELIAN
FGFR3	Thanatophoric dysplasia, type I	OMIM:187600	MENDELIAN
FGFR3	Colorectal cancer	OMIM:114500	MENDELIAN
FGFR3	Muenke syndrome	OMIM:602849	MENDELIAN
FGFR3	Crouzon syndrome with acanthosis nigricans	OMIM:612247	MENDELIAN
FGFR3	Achondroplasia	ORPHA:15	UNKNOWN
FGFR3	Non-syndromic bicoronal craniosynostosis	ORPHA:35099	UNKNOWN
FGFR3	Muenke syndrome	ORPHA:53271	UNKNOWN
FGFR3	Thanatophoric dysplasia type 1	ORPHA:1860	UNKNOWN
FGFR3	Saethre-Chotzen syndrome	ORPHA:794	UNKNOWN
FGFR3	Thanatophoric dysplasia type 2	ORPHA:93274	UNKNOWN
FGFR3	Crouzon syndrome-acanthosis nigricans syndrome	ORPHA:93262	UNKNOWN
FGFR3	Lacrimoauriculodentodigital syndrome	ORPHA:2363	UNKNOWN
FGFR3	Hypochondroplasia	ORPHA:429	UNKNOWN
FGFR3	Camptodactyly-tall stature-scoliosis-hearing loss syndrome	ORPHA:85164	UNKNOWN
FGFR3	Severe achondroplasia-developmental delay-acanthosis nigricans syndrome	ORPHA:85165	UNKNOWN
FGFR4	—		UNKNOWN
FGFRL1	Wolf-Hirschhorn syndrome	OMIM:194190	MENDELIAN
FGG	Congenital dysfibrinogenemia	OMIM:616004	MENDELIAN
FGG	Afibrinogenemia, congenitalhypofibrinogenemia, congenital, included	OMIM:202400	MENDELIAN
FGG	Familial hypofibrinogenemia	ORPHA:101041	UNKNOWN
FGG	Familial afibrinogenemia	ORPHA:98880	UNKNOWN
FGG	Familial dysfibrinogenemia	ORPHA:98881	UNKNOWN
FH	Hereditary leiomyomatosis and renal cell cancer	OMIM:150800	MENDELIAN
FH	Fumarase deficiency	OMIM:606812	MENDELIAN
FH	Fumaric aciduria	ORPHA:24	UNKNOWN
FH	Hereditary leiomyomatosis and renal cell cancer	ORPHA:523	UNKNOWN
FH	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
FHL1	Uruguay faciocardiomusculoskeletal syndrome	OMIM:300280	MENDELIAN
FHL1	Myopathy, reducing body, X-linked, childhood-onset	OMIM:300718	MENDELIAN
FHL1	Myopathy, reducing body, X-linked, early-onset, severe	OMIM:300717	MENDELIAN
FHL1	Myopathy, X-linked, with postural muscle atrophy	OMIM:300696	MENDELIAN
FHL1	Scapuloperoneal myopathy, X-linked dominant	OMIM:300695	MENDELIAN
FHL1	X-linked Emery-Dreifuss muscular dystrophy	ORPHA:98863	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
FHL2	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
FHOD3	Cardiomyopathy, familial hypertrophic, 28	OMIM:619402	MENDELIAN
FIG4	Polymicrogyria, bilateral temporooccipital	OMIM:612691	MENDELIAN
FIG4	Yunis-Varon syndrome	OMIM:216340	MENDELIAN
FIG4	Charcot-Marie-Tooth disease, type 4J	OMIM:611228	MENDELIAN
FIG4	Amyotrophic lateral sclerosis 11	OMIM:612577	MENDELIAN
FIG4	Bilateral parasagittal parieto-occipital polymicrogyria	ORPHA:208441	UNKNOWN
FIG4	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
FIG4	Yunis-Varon syndrome	ORPHA:3472	UNKNOWN
FIGLA	Premature ovarian failure 6	OMIM:612310	MENDELIAN
FIGLA	46,XX gonadal dysgenesis	ORPHA:243	UNKNOWN
FIGN	—		UNKNOWN
FIP1L1	Acute promyelocytic leukemia	ORPHA:520	UNKNOWN
FKBP10	Bruck syndrome 1	OMIM:259450	MENDELIAN
FKBP10	Osteogenesis imperfecta, type XI	OMIM:610968	MENDELIAN
FKBP10	Bruck syndrome	ORPHA:2771	UNKNOWN
FKBP10	Kuskokwim syndrome	ORPHA:1149	UNKNOWN
FKBP14	Ehlers-Danlos syndrome, kyphoscoliotic type, 2	OMIM:614557	MENDELIAN
FKBP14	Kyphoscoliotic Ehlers-Danlos syndrome due to FKBP22 deficiency	ORPHA:300179	UNKNOWN
FKBP1A	—		UNKNOWN
FKBP4	—		UNKNOWN
FKBP5	Major depressive disorder	OMIM:608516	POLYGENIC
FKBP6	Spermatogenic failure 77	OMIM:620103	MENDELIAN
FKBP6	Williams syndrome	ORPHA:904	UNKNOWN
FKBP8	—		UNKNOWN
FKBPL	—		UNKNOWN
FKRP	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	OMIM:607155	MENDELIAN
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	OMIM:236670	MENDELIAN
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	OMIM:613153	MENDELIAN
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5	OMIM:606612	MENDELIAN
FKRP	FKRP-related limb-girdle muscular dystrophy R9	ORPHA:34515	UNKNOWN
FKRP	Congenital muscular dystrophy with intellectual disability	ORPHA:370968	UNKNOWN
FKRP	Congenital muscular dystrophy without intellectual disability	ORPHA:370980	UNKNOWN
FKRP	Congenital muscular dystrophy with cerebellar involvement	ORPHA:370959	UNKNOWN
FKRP	Walker-Warburg syndrome	ORPHA:899	UNKNOWN
FKRP	Muscle-eye-brain disease	ORPHA:588	UNKNOWN
FKTN	Cardiomyopathy, dilated, 1X	OMIM:611615	MENDELIAN
FKTN	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4	OMIM:611588	MENDELIAN
FKTN	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	OMIM:236670	MENDELIAN
FKTN	Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4	OMIM:613152	MENDELIAN
FKTN	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	OMIM:253800	MENDELIAN
FKTN	Congenital muscular dystrophy without intellectual disability	ORPHA:370980	UNKNOWN
FKTN	Congenital muscular dystrophy, Fukuyama type	ORPHA:272	UNKNOWN
FKTN	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
FKTN	Walker-Warburg syndrome	ORPHA:899	UNKNOWN
FKTN	Muscle-eye-brain disease	ORPHA:588	UNKNOWN
FLCN	Pneumothorax, primary spontaneous	OMIM:173600	MENDELIAN
FLCN	Potocki-Lupski syndrome	OMIM:610883	MENDELIAN
FLCN	Colorectal cancer	OMIM:114500	MENDELIAN
FLCN	Renal cell carcinoma, nonpapillary	OMIM:144700	MENDELIAN
FLCN	Birt-Hogg-Dube syndrome	OMIM:135150	MENDELIAN
FLCN	Birt-Hogg-DubÃ© syndrome	ORPHA:122	UNKNOWN
FLCN	Familial spontaneous pneumothorax	ORPHA:2903	UNKNOWN
FLG	Dermatitis, atopic, susceptibility to, 2	OMIM:605803	POLYGENIC
FLG	Ichthyosis vulgaris	OMIM:146700	MENDELIAN
FLG	Recessive X-linked ichthyosis	ORPHA:461	UNKNOWN
FLI1	Bleeding disorder, platelet-type, 21	OMIM:617443	MENDELIAN
FLI1	Peripheral primitive neuroectodermal tumor	ORPHA:370348	UNKNOWN
FLI1	Paris-Trousseau thrombocytopenia	ORPHA:851	UNKNOWN
FLI1	Jacobsen syndrome	ORPHA:2308	UNKNOWN
FLJ22792	—		UNKNOWN
FLNA	Melnick-Needles syndrome	OMIM:309350	MENDELIAN
FLNA	Heterotopia, periventricular, X-linked dominant	OMIM:300049	MENDELIAN
FLNA	Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked	OMIM:300048	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
FLNA	Cardiac valvular dysplasia, X-linked	OMIM:314400	MENDELIAN
FLNA	Fg syndrome 2	OMIM:300321	MENDELIAN
FLNA	Terminal osseous dysplasia	OMIM:300244	MENDELIAN
FLNA	Frontometaphyseal dysplasia	OMIM:305620	MENDELIAN
FLNA	Otopalatodigital syndrome, type II	OMIM:304120	MENDELIAN
FLNA	Otopalatodigital syndrome, type I	OMIM:311300	MENDELIAN
FLNA	Otopalatodigital syndrome type 1	ORPHA:90650	UNKNOWN
FLNA	Otopalatodigital syndrome type 2	ORPHA:90652	UNKNOWN
FLNA	FLNA-related X-linked myxomatous valvular dysplasia	ORPHA:555877	UNKNOWN
FLNA	Neuronal intestinal pseudoobstruction	ORPHA:99811	UNKNOWN
FLNA	Terminal osseous dysplasia-pigmentary defects syndrome	ORPHA:88630	UNKNOWN
FLNA	Periventricular nodular heterotopia	ORPHA:98892	UNKNOWN
FLNA	Frontometaphyseal dysplasia	ORPHA:1826	UNKNOWN
FLNA	X-linked Ehlers-Danlos syndrome	ORPHA:75497	UNKNOWN
FLNA	Congenital short bowel syndrome	ORPHA:2301	UNKNOWN
FLNA	Melnick-Needles syndrome	ORPHA:2484	UNKNOWN
FLNB	Spondylcarpotarsal synostosis syndrome	OMIM:272460	MENDELIAN
FLNB	Atelosteogenesis, type III	OMIM:108721	MENDELIAN
FLNB	Atelosteogenesis, type I	OMIM:108720	MENDELIAN
FLNB	Larsen syndrome	OMIM:150250	MENDELIAN
FLNB	Boomerang dysplasia	OMIM:112310	MENDELIAN
FLNB	Spondylcarpotarsal synostosis	ORPHA:3275	UNKNOWN
FLNB	Boomerang dysplasia	ORPHA:1263	UNKNOWN
FLNB	Atelosteogenesis type III	ORPHA:56305	UNKNOWN
FLNB	Atelosteogenesis type I	ORPHA:1190	UNKNOWN
FLNB	Larsen syndrome	ORPHA:503	UNKNOWN
FLNC	Myopathy, distal, 4	OMIM:614065	MENDELIAN
FLNC	Filaminopathy, autosomal dominant	OMIM:609524	MENDELIAN
FLNC	Cardiomyopathy, familial hypertrophic, 26	OMIM:617047	MENDELIAN
FLNC	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
FLNC	FLNC-related handgrip and calf weakness-distal myopathy	ORPHA:63273	UNKNOWN
FLNC	Familial isolated restrictive cardiomyopathy	ORPHA:75249	UNKNOWN
FLNC	Muscle filaminopathy	ORPHA:171445	UNKNOWN
FLOT1	—		UNKNOWN
FLRT1	Spastic paraplegia-optic atrophy-neuropathy syndrome	ORPHA:320406	UNKNOWN
FLRT3	Hypogonadotropic hypogonadism 21 with or without anosmia	OMIM:615271	MENDELIAN
FLRT3	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
FLRT3	Kallmann syndrome	ORPHA:478	UNKNOWN
FLT1	Preeclampsia	ORPHA:275555	UNKNOWN
FLT3	Leukemia, acute lymphocytic, susceptibility to, 1	OMIM:613065	MENDELIAN
FLT3	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
FLT4	Lymphatic malformation 1	OMIM:153100	MENDELIAN
FLT4	Hemangioma, capillary infantile	OMIM:602089	MENDELIAN
FLT4	Congenital heart defects, multiple types, 7	OMIM:618780	MENDELIAN
FLT4	Milroy disease	ORPHA:79452	UNKNOWN
FLT4	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
FLVCR1	Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia	OMIM:621060	MENDELIAN
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa	OMIM:609033	MENDELIAN
FLVCR1	Posterior column ataxia-retinitis pigmentosa syndrome	ORPHA:88628	UNKNOWN
FLVCR2	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	OMIM:225790	MENDELIAN
FMN1	—		UNKNOWN
FMN2	Intellectual developmental disorder, autosomal recessive 47	OMIM:616193	MENDELIAN
FMO1	—		UNKNOWN
FMO2	—		UNKNOWN
FMO3	Trimethylaminuria	OMIM:602079	MENDELIAN
FMO3	Severe primary trimethylaminuria	ORPHA:468726	UNKNOWN
FMO4	—		UNKNOWN
FMO5	—		UNKNOWN
FMO6P	—		UNKNOWN
FMOD	—		UNKNOWN
FMR1	Fragile X tremor/ataxia syndrome	OMIM:300623	MENDELIAN
FMR1	Fragile X syndrome	OMIM:300624	MENDELIAN
FMR1	Premature ovarian failure 1	OMIM:311360	MENDELIAN
FMR1	Xq27.3q28 duplication syndrome	ORPHA:261483	UNKNOWN
FMR1	Fragile X syndrome	ORPHA:908	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
FMR1	Fragile X-associated tremor/ataxia syndrome	ORPHA:93256	UNKNOWN
FMR1-AS1	—		UNKNOWN
FN1	Spondylometaphyseal dysplasia, corner Fracture type	OMIM:184255	MENDELIAN
FN1	Glomerulopathy with fibronectin deposits 2	OMIM:601894	MENDELIAN
FN1	Fibronectin glomerulopathy	ORPHA:84090	UNKNOWN
FN1	Spondylometaphyseal dysplasia, 'corner fracture' type	ORPHA:93315	UNKNOWN
FN3K	—		UNKNOWN
FNBP1	—		UNKNOWN
FNBP4	—		UNKNOWN
FNDC7	—		UNKNOWN
FOLH1	—		UNKNOWN
FOLR1	Neurodegeneration due to cerebral folate transport deficiency	OMIM:613068	MENDELIAN
FOXA1	—		UNKNOWN
FOXA2	Combined pituitary hormone deficiencies, genetic forms	ORPHA:95494	UNKNOWN
FOXA3	—		UNKNOWN
FOXC1	Axenfeld-Rieger syndrome, type 3	OMIM:602482	MENDELIAN
FOXC1	Anterior segment dysgenesis 3	OMIM:601631	MENDELIAN
FOXC1	Isolated aniridia	ORPHA:250923	UNKNOWN
FOXC1	Axenfeld-Rieger syndrome	ORPHA:782	UNKNOWN
FOXC1	Peters anomaly	ORPHA:708	UNKNOWN
FOXC2	Lymphedema-Distichiasis syndrome	OMIM:153400	MENDELIAN
FOXC2	Lymphedema-distichiasis syndrome	ORPHA:33001	UNKNOWN
FOXD1	—		UNKNOWN
FOXD3	Autoimmune disease, susceptibility to, 1	OMIM:607836	POLYGENIC
FOXD4	—		UNKNOWN
FOXE1	Thyroid cancer, nonmedullary, 4	OMIM:616534	POLYGENIC
FOXE1	Bamforth-Lazarus syndrome	OMIM:241850	MENDELIAN
FOXE1	Bamforth-Lazarus syndrome	ORPHA:1226	UNKNOWN
FOXE1	Athyreosis	ORPHA:95713	UNKNOWN
FOXE1	Familial papillary or follicular thyroid carcinoma	ORPHA:319487	UNKNOWN
FOXE3	Cataract 34, multiple types	OMIM:612968	MENDELIAN
FOXE3	Anterior segment dysgenesis 2	OMIM:610256	MENDELIAN
FOXE3	Aortic aneurysm, familial thoracic 11, susceptibility to	OMIM:617349	POLYGENIC
FOXE3	Congenital primary aphakia	ORPHA:83461	UNKNOWN
FOXE3	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
FOXE3	Peters anomaly	ORPHA:708	UNKNOWN
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins	OMIM:265380	MENDELIAN
FOXF1	Congenital alveolar capillary dysplasia	ORPHA:210122	UNKNOWN
FOXF2	—		UNKNOWN
FOXG1	Rett syndrome, congenital variant	OMIM:613454	MENDELIAN
FOXG1	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
FOXG1	FOXG1 syndrome due to 14q12 microdeletion	ORPHA:261144	UNKNOWN
FOXH1	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
FOXH1	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
FOXH1	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
FOXH1	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
FOXH1	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
FOXH1	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
FOXI1	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	OMIM:600791	MENDELIAN
FOXI1	Pendred syndrome	OMIM:274600	MENDELIAN
FOXI1	Pendred syndrome	ORPHA:705	UNKNOWN
FOXJ1	Ciliary dyskinesia, primary, 43	OMIM:618699	MENDELIAN
FOXJ1	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
FOXK1	—		UNKNOWN
FOXL1	Otosclerosis 11	OMIM:620576	MENDELIAN
FOXL2	Blepharophimosis, epicanthus inversus, and ptosis	OMIM:110100	MENDELIAN
FOXL2	Premature ovarian failure 3	OMIM:608996	MENDELIAN
FOXL2	Blepharophimosis-ptosis-epicanthus inversus syndrome plus	ORPHA:572333	UNKNOWN
FOXN1	—		UNKNOWN
FOXN1	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant	OMIM:618806	MENDELIAN
FOXN1	Immune defect due to absence of thymus	OMIM:242700	MENDELIAN
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	OMIM:601705	MENDELIAN
FOXN1	T-cell immunodeficiency with thymic aplasia	ORPHA:83471	UNKNOWN
FOXN1	T-B+NK+ severe combined immunodeficiency due to FOXN1 deficiency	ORPHA:169095	UNKNOWN
FOXO1	Rhabdomyosarcoma 2, alveolar	OMIM:268220	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
FOXP1	Intellectual developmental disorder with language impairment with or without autistic features	OMIM:613670	MENDELIAN
FOXP1	MALT lymphoma	ORPHA:52417	UNKNOWN
FOXP1	FOXP1 Syndrome	ORPHA:391372	UNKNOWN
FOXP2	Speech-language disorder-1	OMIM:602081	MENDELIAN
FOXP2	Isolated childhood apraxia of speech	ORPHA:209908	UNKNOWN
FOXP2	7q31 microdeletion syndrome	ORPHA:251061	UNKNOWN
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	OMIM:304790	MENDELIAN
FOXP3	Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome	ORPHA:37042	UNKNOWN
FOXP4	—		UNKNOWN
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19	OMIM:618241	MENDELIAN
FOXRED1	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
FPGS	—		UNKNOWN
FPR2	—		UNKNOWN
FRA10AC1	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities	OMIM:620113	MENDELIAN
FRAS1	Fraser syndrome	OMIM:219000	MENDELIAN
FRAS1	Fraser syndrome	ORPHA:2052	UNKNOWN
FREM1	Manitoba oculotrichoanal syndrome	OMIM:248450	MENDELIAN
FREM1	Bifid nose with or without anorectal and renal anomalies	OMIM:608980	MENDELIAN
FREM1	Trigonocephaly 2	OMIM:614485	MENDELIAN
FREM1	Oculotrichoanal syndrome	ORPHA:2717	UNKNOWN
FREM1	Non-syndromic metopic craniosynostosis	ORPHA:3366	UNKNOWN
FREM1	BNAR syndrome	ORPHA:217266	UNKNOWN
FREM2	Fraser syndrome 2	OMIM:617666	MENDELIAN
FREM2	Cryptophthalmos, unilateral or bilateral, isolated	OMIM:123570	MENDELIAN
FREM2	Fraser syndrome	ORPHA:2052	UNKNOWN
FREM3	—		UNKNOWN
FRG1	Facioscapulohumeral muscular dystrophy 1	OMIM:158900	MENDELIAN
FRG1	Facioscapulohumeral dystrophy	ORPHA:269	UNKNOWN
FRK	—		UNKNOWN
FRMD5	Neurodevelopmental disorder with eye movement abnormalities and ataxia	OMIM:620094	MENDELIAN
FRMD6	—		UNKNOWN
FRMD7	Nystagmus 1, congenital, X-linked	OMIM:310700	MENDELIAN
FRMPD4	Intellectual developmental disorder, X-linked 104	OMIM:300983	MENDELIAN
FRRS1L	Epileptic encephalopathy, early infantile, 37	OMIM:616981	MENDELIAN
FRS2	—		UNKNOWN
FRY	—		UNKNOWN
FRYL	Pan-Chung-Bellen syndrome	OMIM:621049	MENDELIAN
FRZB	Osteoarthritis susceptibility 1	OMIM:165720	POLYGENIC
FSBP	—		UNKNOWN
FSCB	—		UNKNOWN
FSCN2	Retinitis pigmentosa 30	OMIM:607921	MENDELIAN
FSCN2	Retinitis pigmentosa	ORPHA:791	UNKNOWN
FSHB	Hypogonadotropic hypogonadism 24 without anosmia	OMIM:229070	MENDELIAN
FSHB	Isolated follicle stimulating hormone deficiency	ORPHA:52901	UNKNOWN
FSHR	Ovarian hyperstimulation syndrome	OMIM:608115	MENDELIAN
FSHR	Ovarian dysgenesis 1	OMIM:233300	MENDELIAN
FSHR	46,XX gonadal dysgenesis	ORPHA:243	UNKNOWN
FSHR	Ovarian hyperstimulation syndrome	ORPHA:64739	UNKNOWN
FST	—		UNKNOWN
FSTL1	—		UNKNOWN
FSTL4	—		UNKNOWN
FSTL5	—		UNKNOWN
FTCD	Formiminotransferase deficiency	OMIM:229100	MENDELIAN
FTCD	Formiminoglutamic aciduria	ORPHA:51208	UNKNOWN
FTH1	Hemochromatosis, type 5	OMIM:615517	MENDELIAN
FTH1	Neurodegeneration with brain iron accumulation 9	OMIM:620669	MENDELIAN
FTHL17	—		UNKNOWN
FTL	Hyperferritinemia with or without cataract	OMIM:600886	MENDELIAN
FTL	L-ferritin deficiency, dominant and recessive	OMIM:615604	MENDELIAN
FTL	Neurodegeneration with brain iron accumulation 3	OMIM:606159	MENDELIAN
FTL	Hereditary hyperferritinemia-cataract syndrome	ORPHA:163	UNKNOWN
FTL	Neuroferritinopathy	ORPHA:157846	UNKNOWN
FTL	Genetic hyperferritinemia without iron overload	ORPHA:254704	UNKNOWN
FTO	Growth retardation, developmental delay, coarse facies, and earlydeath	OMIM:612938	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
FTO	Obesity, susceptibility to, BMIQ14	OMIM:612460	POLYGENIC
FTSJ1	Intellectual developmental disorder, X-linked 9	OMIM:309549	MENDELIAN
FUBP1	—		UNKNOWN
FUCA1	Fucosidosis	OMIM:230000	MENDELIAN
FUCA1	Fucosidosis	ORPHA:349	UNKNOWN
FURIN	—		UNKNOWN
FUS	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia	OMIM:608030	MENDELIAN
FUS	Tremor, hereditary essential, 4	OMIM:614782	MENDELIAN
FUS	Myxoid/round cell liposarcoma	ORPHA:99967	UNKNOWN
FUS	Frontotemporal dementia with motor neuron disease	ORPHA:275872	UNKNOWN
FUS	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
FUS	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
FUT1	Bombay phenotype	OMIM:616754	MENDELIAN
FUT2	Bombay phenotype	OMIM:616754	MENDELIAN
FUT7	—		UNKNOWN
FUT8	Congenital disorder of glycosylation with defective fucosylation 1	OMIM:618005	MENDELIAN
FUZ	Neural tube defects, susceptibility to	OMIM:182940	POLYGENIC
FUZ	Caudal regression syndrome	ORPHA:3027	UNKNOWN
FXN	Friedreich ataxia 2	OMIM:601992	MENDELIAN
FXN	Friedreich ataxia 1	OMIM:229300	MENDELIAN
FXN	Friedreich ataxia	ORPHA:95	UNKNOWN
FXYD2	Hypomagnesemia 2, renal	OMIM:154020	MENDELIAN
FXYD6	—		UNKNOWN
FYCO1	Cataract, autosomal recessive congenital 2	OMIM:610019	MENDELIAN
FYN	—		UNKNOWN
FZD1	—		UNKNOWN
FZD3	—		UNKNOWN
FZD4	Exudative vitreoretinopathy 1	OMIM:133780	MENDELIAN
FZD4	Familial exudative vitreoretinopathy	ORPHA:891	UNKNOWN
FZD4	Retinopathy of prematurity	ORPHA:90050	UNKNOWN
FZD4	Persistent hyperplastic primary vitreous	ORPHA:91495	UNKNOWN
FZD6	Nail disorder, nonsyndromic congenital, 1	OMIM:161050	MENDELIAN
FZD9	—		UNKNOWN
FZR1	Developmental and epileptic encephalopathy 109	OMIM:620145	MENDELIAN
FZR1	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
G6PC	—		UNKNOWN
G6PC2	—		UNKNOWN
G6PC3	Neutropenia, severe congenital, 4, autosomal recessive	OMIM:612541	MENDELIAN
G6PD	Hemolytic anemia, G6PD deficient (favism)	OMIM:300908	MENDELIAN
GAA	Glycogen storage disease II	OMIM:232300	MENDELIAN
GAA	Pompe disease, late-onset	OMIM:621314	MENDELIAN
GAA	Glycogen storage disease due to acid maltase deficiency, infantile onset	ORPHA:308552	UNKNOWN
GAB2	—		UNKNOWN
GABARAPL1	—		UNKNOWN
GABBR1	Neurodevelopmental disorder with language delay and variable cognitive abnormalities	OMIM:620502	MENDELIAN
GABBR2	Neurodevelopmental disorder with poor language and loss of hand skills	OMIM:617903	MENDELIAN
GABBR2	Epileptic encephalopathy, early infantile, 59	OMIM:617904	MENDELIAN
GABBR2	Atypical Rett syndrome	ORPHA:3095	UNKNOWN
GABBR2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
GABRA1	Developmental and epileptic encephalopathy 19	OMIM:615744	MENDELIAN
GABRA1	Juvenile myoclonic epilepsy	ORPHA:307	UNKNOWN
GABRA1	Childhood absence epilepsy	ORPHA:64280	UNKNOWN
GABRA1	Dravet syndrome	ORPHA:33069	UNKNOWN
GABRA2	Alcohol dependence	OMIM:103780	POLYGENIC
GABRA2	Epileptic encephalopathy, early infantile, 78	OMIM:618557	MENDELIAN
GABRA2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
GABRA3	Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features	OMIM:301091	MENDELIAN
GABRA3	Thyrotoxic periodic paralysis	ORPHA:79102	UNKNOWN
GABRA5	Epileptic encephalopathy, early infantile, 79	OMIM:618559	MENDELIAN
GABRA5	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
GABRA6	—		UNKNOWN
GABRB2	Epileptic encephalopathy, infantile or early childhood, 2	OMIM:617829	MENDELIAN
GABRB2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
GABRB3	Epileptic encephalopathy, early infantile, 43	OMIM:617113	MENDELIAN
GABRB3	Epilepsy, childhood absence, susceptibility to, 5	OMIM:612269	POLYGENIC

Gene_symbol	Disease_name	Disease_ID	Association_type
GABRB3	Lennox-Gastaut syndrome	ORPHA:2382	UNKNOWN
GABRB3	Childhood absence epilepsy	ORPHA:64280	UNKNOWN
GABRD	Epilepsy, idiopathic generalized, 10	OMIM:613060	POLYGENIC
GABRD	Juvenile myoclonic epilepsy	ORPHA:307	UNKNOWN
GABRD	Genetic epilepsy with febrile seizure plus	ORPHA:36387	UNKNOWN
GABRD	1p36 deletion syndrome	ORPHA:1606	UNKNOWN
GABRG1	—		UNKNOWN
GABRG2	Epileptic encephalopathy, early infantile, 74	OMIM:618396	MENDELIAN
GABRG2	Febrile seizures, familial, 8	OMIM:607681	MENDELIAN
GABRG2	Genetic epilepsy with febrile seizure plus	ORPHA:36387	UNKNOWN
GABRG2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
GABRG2	Self-limited epilepsy with centrotemporal spikes	ORPHA:1945	UNKNOWN
GABRG2	Childhood absence epilepsy	ORPHA:64280	UNKNOWN
GABRG2	Dravet syndrome	ORPHA:33069	UNKNOWN
GABRG3	—		UNKNOWN
GABRR2	—		UNKNOWN
GAD1	Developmental and epileptic encephalopathy 89	OMIM:619124	MENDELIAN
GAD2	—		UNKNOWN
GADD45A	—		UNKNOWN
GADD45B	—		UNKNOWN
GAGE8	—		UNKNOWN
GAK	—		UNKNOWN
GAL3ST1	—		UNKNOWN
GAL3ST2	—		UNKNOWN
GAL3ST3	—		UNKNOWN
GAL3ST4	—		UNKNOWN
GALC	Krabbe disease	OMIM:245200	MENDELIAN
GALC	Late-infantile/juvenile Krabbe disease	ORPHA:206443	UNKNOWN
GALC	Adult Krabbe disease	ORPHA:206448	UNKNOWN
GALC	Infantile Krabbe disease	ORPHA:206436	UNKNOWN
GALE	Galactose epimerase deficiency	OMIM:230350	MENDELIAN
GALE	Thrombocytopenia 13, syndromic	OMIM:620776	MENDELIAN
GALK1	Galactokinase deficiency with cataracts	OMIM:230200	MENDELIAN
GALK1	Galactokinase deficiency	ORPHA:79237	UNKNOWN
GALNS	Mucopolysaccharidosis IVA	OMIM:253000	MENDELIAN
GALNT11	—		UNKNOWN
GALNT13	—		UNKNOWN
GALNT14	—		UNKNOWN
GALNT18	—		UNKNOWN
GALNT2	Congenital disorder of glycosylation, type II t	OMIM:618885	MENDELIAN
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 1	OMIM:211900	MENDELIAN
GALNT5	—		UNKNOWN
GALNT6	—		UNKNOWN
GALNT7	—		UNKNOWN
GALNT8	—		UNKNOWN
GALNT9	—		UNKNOWN
GALNTL5	—		UNKNOWN
GALNTL6	—		UNKNOWN
GALP	—		UNKNOWN
GALT	Galactosemia	OMIM:230400	MENDELIAN
GALT	Classic galactosemia	ORPHA:79239	UNKNOWN
GAMT	Cerebral creatine deficiency syndrome 2	OMIM:612736	MENDELIAN
GAMT	Guanidinoacetate methyltransferase deficiency	ORPHA:382	UNKNOWN
GAN	Giant axonal neuropathy 1, autosomal recessive	OMIM:256850	MENDELIAN
GAN	Giant axonal neuropathy	ORPHA:643	UNKNOWN
GAP43	—		UNKNOWN
GARS	—		UNKNOWN
GAS1	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
GAS1	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
GAS1	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
GAS1	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
GAS1	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
GAS1	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
GAS2L2	Ciliary dyskinesia, primary, 41	OMIM:618449	MENDELIAN
GAS2L2	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
GAS6	—		UNKNOWN
GATA1	Thrombocytopenia with beta-thalassemia, X-linked	OMIM:314050	MENDELIAN
GATA1	Myeloproliferative syndrome, transient	OMIM:159595	MENDELIAN
GATA1	Hemolytic anemia due to elevated adenosine deaminase	OMIM:301083	MENDELIAN
GATA1	Thrombocytopenia, X-linked, with or without dyserythropoietic anemia	OMIM:300367	MENDELIAN
GATA1	Anemia, X-linked, with or without neutropenia and/or platelet abnormalities	OMIM:300835	MENDELIAN
GATA1	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
GATA1	Thrombocytopenia with congenital dyserythropoietic anemia	ORPHA:67044	UNKNOWN
GATA1	Beta-thalassemia-X-linked thrombocytopenia syndrome	ORPHA:231393	UNKNOWN
GATA1	Congenital erythropoietic porphyria	ORPHA:79277	UNKNOWN
GATA2	Myelodysplastic syndrome	OMIM:614286	POLYGENIC
GATA2	Immunodeficiency 21	OMIM:614172	MENDELIAN
GATA2	Lymphedema, primary, with myelodysplasia	OMIM:614038	MENDELIAN
GATA2	Leukemia, acute myeloid	OMIM:601626	POLYGENIC
GATA2	GATA2 deficiency spectrum	ORPHA:228423	UNKNOWN
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia	OMIM:146255	MENDELIAN
GATA3	Hypoparathyroidism-sensorineural deafness-renal disease syndrome	ORPHA:2237	UNKNOWN
GATA4	Testicular anomalies with or without congenital heart disease	OMIM:615542	MENDELIAN
GATA4	Tetralogy of Fallot	OMIM:187500	MENDELIAN
GATA4	Ventricular septal defect 1	OMIM:614429	MENDELIAN
GATA4	Atrioventricular septal defect 4	OMIM:614430	MENDELIAN
GATA4	Atrial septal defect 2	OMIM:607941	MENDELIAN
GATA4	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
GATA4	46,XY partial gonadal dysgenesis	ORPHA:251510	UNKNOWN
GATA4	Atrial septal defect, ostium secundum type	ORPHA:99103	UNKNOWN
GATA4	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
GATA4	8p23.1 microdeletion syndrome	ORPHA:251071	UNKNOWN
GATA5	Congenital heart defects, multiple types, 5	OMIM:617912	MENDELIAN
GATA5	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
GATA5	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
GATA5	Familial bicuspid aortic valve	ORPHA:402075	UNKNOWN
GATA6	Conotruncal heart malformations	OMIM:217095	MENDELIAN
GATA6	Pancreatic agenesis and congenital heart defects	OMIM:600001	MENDELIAN
GATA6	Tetralogy of Fallot	OMIM:187500	MENDELIAN
GATA6	Atrioventricular septal defect 5	OMIM:614474	MENDELIAN
GATA6	Atrial septal defect 9	OMIM:614475	MENDELIAN
GATA6	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
GATA6	Atrial septal defect, ostium secundum type	ORPHA:99103	UNKNOWN
GATA6	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
GATA6	Congenital diaphragmatic hernia	ORPHA:2140	UNKNOWN
GATA6	Pancreatic hypoplasia-diabetes-congenital heart disease syndrome	ORPHA:2255	UNKNOWN
GATAD1	Cardiomyopathy, dilated, 2B	OMIM:614672	MENDELIAN
GATAD1	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
GATAD2B	GAND syndrome	OMIM:615074	MENDELIAN
GATAD2B	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome	ORPHA:363686	UNKNOWN
GATM	Cerebral creatine deficiency syndrome 3	OMIM:612718	MENDELIAN
GATM	Fanconi renotubular syndrome 1	OMIM:134600	MENDELIAN
GATM	Primary Fanconi renotubular syndrome	ORPHA:3337	UNKNOWN
GATM	L-Arginine:glycine amidinotransferase deficiency	ORPHA:35704	UNKNOWN
GBA	—		UNKNOWN
GBA2	Spastic paraplegia 46, autosomal recessive	OMIM:614409	MENDELIAN
GBA2	Autosomal recessive cerebellar ataxia with late-onset spasticity	ORPHA:352641	UNKNOWN
GBA2	Autosomal recessive spastic paraplegia type 46	ORPHA:320391	UNKNOWN
GBA3	—		UNKNOWN
GBE1	Polyglucosan body neuropathy, adult form	OMIM:263570	MENDELIAN
GBE1	Glycogen storage disease IV	OMIM:232500	MENDELIAN
GBE1	Adult polyglucosan body disease	ORPHA:206583	UNKNOWN
GBGT1	—		UNKNOWN
GC	—		UNKNOWN
GCDH	Glutaricaciduria, type I	OMIM:231670	MENDELIAN
GCDH	Glutaryl-CoA dehydrogenase deficiency	ORPHA:25	UNKNOWN
GCG	—		UNKNOWN
GCGR	Mahvash disease	OMIM:619290	MENDELIAN
GCGR	GCGR-related hyperglucagonemia	ORPHA:438274	UNKNOWN
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia	OMIM:128230	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
GCH1	Hyperphenylalaninemia, BH4-deficient, B	OMIM:233910	MENDELIAN
GCH1	Autosomal dominant dopa-responsive dystonia	ORPHA:98808	UNKNOWN
GCK	Diabetes mellitus, noninsulin-dependent	OMIM:125853	MENDELIAN
GCK	Maturity-onset diabetes of the young, type II	OMIM:125851	MENDELIAN
GCK	Diabetes mellitus, permanent neonatal 1	OMIM:606176	MENDELIAN
GCK	Hyperinsulinemic hypoglycemia, familial, 3	OMIM:602485	MENDELIAN
GCK	Isolated permanent neonatal diabetes mellitus	ORPHA:99885	UNKNOWN
GCK	MODY	ORPHA:552	UNKNOWN
GCK	Congenital glucokinase-related hyperinsulinism	ORPHA:79299	UNKNOWN
GCLC	Anemia, congenital, nonspherocytic hemolytic, 7	OMIM:230450	MENDELIAN
GCLC	Glutamate-cysteine ligase deficiency	ORPHA:33574	UNKNOWN
GCLC	Cystic fibrosis	ORPHA:586	UNKNOWN
GCM2	Hypoparathyroidism, familial isolated 2	OMIM:618883	MENDELIAN
GCM2	Hyperparathyroidism 4	OMIM:617343	MENDELIAN
GCM2	Familial isolated hyperparathyroidism	ORPHA:99879	UNKNOWN
GCM2	Familial isolated hypoparathyroidism due to agenesis of parathyroid gland	ORPHA:2239	UNKNOWN
GCNA	Spermatogenic failure, X-linked, 4	OMIM:301077	MENDELIAN
GCNA	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
GCNT1	—		UNKNOWN
GCNT2	Cataract 13 with adult i phenotype	OMIM:116700	MENDELIAN
GCSH	Multiple mitochondrial dysfunctions syndrome 7	OMIM:620423	MENDELIAN
GDAP1	Charcot-Marie-Tooth disease, recessive intermediate A	OMIM:608340	MENDELIAN
GDAP1	Charcot-Marie-Tooth disease, axonal, type 2K	OMIM:607831	MENDELIAN
GDAP1	Charcot-Marie-Tooth disease, type 4A	OMIM:214400	MENDELIAN
GDAP1	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, autosomalrecessive	OMIM:607706	MENDELIAN
GDAP1	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness	ORPHA:101097	UNKNOWN
GDAP1	Charcot-Marie-Tooth disease type 4A	ORPHA:99948	UNKNOWN
GDAP1	Autosomal dominant Charcot-Marie-Tooth disease type 2K	ORPHA:99944	UNKNOWN
GDF1	Congenital heart defects, multiple types, 6	OMIM:613854	MENDELIAN
GDF1	Right atrial isomerism (Ivemark)	OMIM:208530	MENDELIAN
GDF1	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
GDF15	Hyperemesis gravidarum, susceptibility to	OMIM:620730	POLYGENIC
GDF2	Telangiectasia, hereditary hemorrhagic, type 5	OMIM:615506	MENDELIAN
GDF2	Hereditary hemorrhagic telangiectasia	ORPHA:774	UNKNOWN
GDF3	Microphthalmia, isolated, with coloboma 6	OMIM:613703	MENDELIAN
GDF3	Klippel-Feil syndrome 3, autosomal dominant	OMIM:613702	MENDELIAN
GDF3	Microphthalmia, isolated 7	OMIM:613704	MENDELIAN
GDF3	Isolated Klippel-Feil syndrome	ORPHA:2345	UNKNOWN
GDF5	Symphalangism, proximal, 1B	OMIM:615298	MENDELIAN
GDF5	Brachydactyly, type A2	OMIM:112600	MENDELIAN
GDF5	Acromesomelic dysplasia, Hunter-Thompson type	OMIM:201250	MENDELIAN
GDF5	Brachydactyly, type A1, C	OMIM:615072	MENDELIAN
GDF5	Multiple synostoses syndrome 2	OMIM:610017	MENDELIAN
GDF5	Brachydactyly, type C	OMIM:113100	MENDELIAN
GDF5	Acromesomelic dysplasia 2A	OMIM:200700	MENDELIAN
GDF5	Fibular hypoplasia and complex brachydactyly	OMIM:228900	MENDELIAN
GDF5	Proximal symphalangism	ORPHA:3250	UNKNOWN
GDF5	Multiple synostoses syndrome	ORPHA:3237	UNKNOWN
GDF5	Acromesomelic dysplasia, Hunter-Thompson type	ORPHA:968	UNKNOWN
GDF5	Fibular aplasia-complex brachydactyly syndrome	ORPHA:2639	UNKNOWN
GDF5	Angel-shaped phalango-epiphyseal dysplasia	ORPHA:63442	UNKNOWN
GDF5	Brachydactyly type A2	ORPHA:93396	UNKNOWN
GDF5	Brachydactyly type C	ORPHA:93384	UNKNOWN
GDF5	Brachydactyly type A1	ORPHA:93388	UNKNOWN
GDF5	Acromesomelic dysplasia, Grebe type	ORPHA:2098	UNKNOWN
GDF6	Multiple synostoses syndrome 4	OMIM:617898	MENDELIAN
GDF6	Microphthalmia, isolated 4	OMIM:613094	MENDELIAN
GDF6	Leber congenital amaurosis 17	OMIM:615360	MENDELIAN
GDF6	Klippel-Feil syndrome 1, autosomal dominant	OMIM:118100	MENDELIAN
GDF6	Microphthalmia, isolated, with coloboma 6	OMIM:613703	MENDELIAN
GDF6	Multiple synostoses syndrome	ORPHA:3237	UNKNOWN
GDF6	Leber congenital amaurosis	ORPHA:65	UNKNOWN
GDF6	Isolated Klippel-Feil syndrome	ORPHA:2345	UNKNOWN
GDF9	Premature ovarian failure 14	OMIM:618014	MENDELIAN
GDI1	Intellectual developmental disorder, X-linked 41	OMIM:300849	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
GDNF	Hirschsprung disease, susceptibility to, 3	OMIM:613711	POLYGENIC
GDNF	Hirschsprung disease	ORPHA:388	UNKNOWN
GDNF	Congenital central hypoventilation syndrome	ORPHA:661	UNKNOWN
GEMIN2	—		UNKNOWN
GEMIN4	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities	OMIM:617913	MENDELIAN
GEN1	—		UNKNOWN
GFAP	Alexander disease	OMIM:203450	MENDELIAN
GFAP	Alexander disease type II	ORPHA:363722	UNKNOWN
GFAP	Alexander disease type I	ORPHA:363717	UNKNOWN
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	OMIM:613076	MENDELIAN
GFER	Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome	ORPHA:330054	UNKNOWN
GFI1	Neutropenia, severe congenital, 2, autosomal dominant	OMIM:613107	MENDELIAN
GFI1	Neutropenia, nonimmune chronic idiopathic, of adults	OMIM:607847	MENDELIAN
GFI1	Autosomal dominant severe congenital neutropenia	ORPHA:486	UNKNOWN
GFI1B	Bleeding disorder, platelet-type, 17	OMIM:187900	MENDELIAN
GFI1B	Autosomal dominant macrothrombocytopenia	ORPHA:140957	UNKNOWN
GFM1	Combined oxidative phosphorylation deficiency 1	OMIM:609060	MENDELIAN
GFM1	Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1	ORPHA:137681	UNKNOWN
GFM2	Combined oxidative phosphorylation deficiency 39	OMIM:618397	MENDELIAN
GFM2	Combined oxidative phosphorylation defect type 39	ORPHA:565624	UNKNOWN
GFPT1	Myasthenic syndrome, congenital, with tubular aggregates 1	OMIM:610542	MENDELIAN
GFPT1	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	OMIM:608931	MENDELIAN
GFPT1	Congenital myasthenic syndrome with glycosylation defect	ORPHA:353327	UNKNOWN
GFPT2	—		UNKNOWN
GFRA1	Renal hypodysplasia/aplasia 4	OMIM:619887	MENDELIAN
GFRA1	Renal agenesis, bilateral	ORPHA:1848	UNKNOWN
GFRA2	—		UNKNOWN
GGCX	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	OMIM:610842	MENDELIAN
GGCX	Vitamin K-dependent clotting factors, combined deficiency of, 1	OMIM:277450	MENDELIAN
GGCX	Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency	ORPHA:91135	UNKNOWN
GGCX	Pseudoxanthoma elasticum-like skin manifestations with retinitis pigmentosa	ORPHA:436274	UNKNOWN
GGCX	Hereditary combined deficiency of vitamin K-dependent clotting factors	ORPHA:98434	UNKNOWN
GGH	—		UNKNOWN
GGT5	—		UNKNOWN
GH1	Growth hormone deficiency, isolated, type IB	OMIM:612781	MENDELIAN
GH1	Growth hormone deficiency, isolated, type IA	OMIM:262400	MENDELIAN
GH1	Pituitary dwarfism IV	OMIM:262650	MENDELIAN
GH1	Isolated growth hormone deficiency, type II	OMIM:173100	MENDELIAN
GH2	—		UNKNOWN
GHR	Hypercholesterolemia, familial, 1	OMIM:143890	POLYGENIC
GHR	Laron syndrome	OMIM:262500	MENDELIAN
GHR	Growth hormone insensitivity, partial	OMIM:604271	MENDELIAN
GHR	Short stature due to partial GHR deficiency	ORPHA:314802	UNKNOWN
GHR	Laron syndrome	ORPHA:633	UNKNOWN
GHRH	—		UNKNOWN
GHRHR	Growth hormone deficiency, isolated, type IV	OMIM:618157	MENDELIAN
GHRL	Obesity	OMIM:601665	POLYGENIC
GHSR	Growth hormone deficiency, isolated partial	OMIM:615925	MENDELIAN
GHSR	Short stature due to GHSR deficiency	ORPHA:314811	UNKNOWN
GID4	—		UNKNOWN
GIF	—		UNKNOWN
GIGYF1	—		UNKNOWN
GIGYF2	Parkinson disease 11	OMIM:607688	POLYGENIC
GIGYF2	Hereditary late-onset Parkinson disease	ORPHA:411602	UNKNOWN
GIMAP8	—		UNKNOWN
GIP	—		UNKNOWN
GIPC3	Deafness, autosomal recessive 15	OMIM:601869	MENDELIAN
GIPR	—		UNKNOWN
GIT1	—		UNKNOWN
GJA1	Palmoplantar keratoderma with congenital alopecia	OMIM:104100	MENDELIAN
GJA1	Craniometaphyseal dysplasia, autosomal recessive	OMIM:218400	MENDELIAN
GJA1	Oculodentodigital dysplasia, autosomal recessive	OMIM:257850	MENDELIAN
GJA1	Erythrokeratoderma variabilis et progressiva 3	OMIM:617525	MENDELIAN
GJA1	Oculodentodigital dysplasia	OMIM:164200	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
GJA1	Syndactyly, type III	OMIM:186100	MENDELIAN
GJA1	Oculodentodigital dysplasia	ORPHA:2710	UNKNOWN
GJA1	Erythrokeratoderma variabilis	ORPHA:317	UNKNOWN
GJA1	Craniometaphyseal dysplasia	ORPHA:1522	UNKNOWN
GJA1	Autosomal dominant palmoplantar keratoderma and congenital alopecia	ORPHA:1010	UNKNOWN
GJA1	Syndactyly type 3	ORPHA:93404	UNKNOWN
GJA1	Hypoplastic left heart syndrome	ORPHA:2248	UNKNOWN
GJA3	Cataract, zonular pulverulent 3	OMIM:601885	MENDELIAN
GJA4	—		UNKNOWN
GJA5	Atrial fibrillation, familial, 11	OMIM:614049	MENDELIAN
GJA5	Atrial standstill 1	OMIM:108770	MENDELIAN
GJA5	Chromosome 1q21.1 deletion syndrome, 1.35-mb	OMIM:612474	MENDELIAN
GJA5	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
GJA5	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
GJA8	Cataract 1, multiple types	OMIM:116200	MENDELIAN
GJA8	Chromosome 1q21.1 deletion syndrome, 1.35-mb	OMIM:612474	MENDELIAN
GJA8	Cataract-microcornea syndrome	ORPHA:1377	UNKNOWN
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	OMIM:302800	MENDELIAN
GJB1	X-linked Charcot-Marie-Tooth disease type 1	ORPHA:101075	UNKNOWN
GJB1	X-linked progressive cerebellar ataxia	ORPHA:1175	UNKNOWN
GJB2	Vohwinkel syndrome	OMIM:124500	MENDELIAN
GJB2	Ichthyosis, hystrix-like, with deafness	OMIM:602540	MENDELIAN
GJB2	Bart-Pumphrey syndrome	OMIM:149200	MENDELIAN
GJB2	Keratitis-ichthyosis-deafness syndrome	OMIM:148210	MENDELIAN
GJB2	Deafness, autosomal recessive 1A	OMIM:220290	MENDELIAN
GJB2	Deafness, autosomal dominant nonsyndromic sensorineural 3	OMIM:601544	MENDELIAN
GJB2	Keratoderma, palmoplantar, with deafness	OMIM:148350	MENDELIAN
GJB2	Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome	ORPHA:2698	UNKNOWN
GJB2	KID syndrome	ORPHA:477	UNKNOWN
GJB2	Keratoderma hereditarium mutilans	ORPHA:494	UNKNOWN
GJB2	Palmoplantar keratoderma-deafness syndrome	ORPHA:2202	UNKNOWN
GJB3	Deafness, autosomal dominant 2B	OMIM:612644	MENDELIAN
GJB3	Erythrokeratoderma variabilis et progressiva 1	OMIM:133200	MENDELIAN
GJB3	Deafness, autosomal recessive 1A	OMIM:220290	MENDELIAN
GJB3	Erythrokeratoderma variabilis	ORPHA:317	UNKNOWN
GJB4	Erythrokeratoderma variabilis et progressiva 2	OMIM:617524	MENDELIAN
GJB4	Erythrokeratoderma variabilis	ORPHA:317	UNKNOWN
GJB6	Clouston syndrome	OMIM:129500	MENDELIAN
GJB6	Deafness, autosomal dominant 3B	OMIM:612643	MENDELIAN
GJB6	Deafness, autosomal recessive 1B	OMIM:612645	MENDELIAN
GJB6	Deafness, autosomal recessive 1A	OMIM:220290	MENDELIAN
GJB6	Hidrotic ectodermal dysplasia	ORPHA:189	UNKNOWN
GJB6	KID syndrome	ORPHA:477	UNKNOWN
GJC2	Lymphatic malformation 3	OMIM:613480	MENDELIAN
GJC2	Leukodystrophy, hypomyelinating, 2	OMIM:608804	MENDELIAN
GJC2	Spastic paraplegia 44, autosomal recessive	OMIM:613206	MENDELIAN
GJC2	Autosomal recessive spastic paraplegia type 44	ORPHA:320401	UNKNOWN
GJC2	GJC2-related late-onset primary lymphedema	ORPHA:568051	UNKNOWN
GJC3	—		UNKNOWN
GJD2	—		UNKNOWN
GK	Glycerol kinase deficiency	OMIM:307030	MENDELIAN
GLA	Fabry disease	OMIM:301500	MENDELIAN
GLA	Fabry disease	ORPHA:324	UNKNOWN
GLB1	GM1-gangliosidosis, type I	OMIM:230500	MENDELIAN
GLB1	Mucopolysaccharidosis type IVB (Morquio)	OMIM:253010	MENDELIAN
GLB1	GM1-gangliosidosis, type II	OMIM:230600	MENDELIAN
GLB1	GM1-gangliosidosis, type III	OMIM:230650	MENDELIAN
GLB1	GM1 gangliosidosis type 1	ORPHA:79255	UNKNOWN
GLDC	Glycine encephalopathy	OMIM:605899	MENDELIAN
GLE1	Congenital arthrogryposis with anterior horn cell disease	OMIM:611890	MENDELIAN
GLE1	Lethal congenital contracture syndrome 1	OMIM:253310	MENDELIAN
GLE1	Lethal congenital contracture syndrome type 1	ORPHA:1486	UNKNOWN
GLE1	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
GLI1	Polydactyly, postaxial, type A8	OMIM:618123	MENDELIAN
GLI1	Polydactyly, preaxial I	OMIM:174400	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
GLI1	Ellis-Van Creveld syndrome	ORPHA:289	UNKNOWN
GLI2	Holoprosencephaly 9	OMIM:610829	MENDELIAN
GLI2	Culler-Jones syndrome	OMIM:615849	MENDELIAN
GLI2	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
GLI2	Combined pituitary hormone deficiencies, genetic forms	ORPHA:95494	UNKNOWN
GLI2	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
GLI2	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
GLI2	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
GLI2	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
GLI2	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
GLI3	Pallister-Hall syndrome	OMIM:146510	MENDELIAN
GLI3	Polydactyly, postaxial, types A1 and B	OMIM:174200	MENDELIAN
GLI3	Greig cephalopolysyndactyly syndrome	OMIM:175700	MENDELIAN
GLI3	Polydactyly, preaxial IV	OMIM:174700	MENDELIAN
GLI3	Greig cephalopolysyndactyly syndrome	ORPHA:380	UNKNOWN
GLI3	Acrocallosal syndrome	ORPHA:36	UNKNOWN
GLI3	Pallister-Hall syndrome	ORPHA:672	UNKNOWN
GLI3	Isolated tibial hemimelia	ORPHA:93322	UNKNOWN
GLIS2	Nephronophtsis 7	OMIM:611498	MENDELIAN
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism	OMIM:610199	MENDELIAN
GLMN	Glomuvenous malformations	OMIM:138000	MENDELIAN
GLMN	Glomuvenous malformation	ORPHA:83454	UNKNOWN
GLO1	—		UNKNOWN
GLP1R	—		UNKNOWN
GLRA1	Hyperekplexia 1	OMIM:149400	MENDELIAN
GLRA1	Hereditary hyperekplexia	ORPHA:3197	UNKNOWN
GLRA4	—		UNKNOWN
GLRB	Hyperekplexia 2	OMIM:614619	MENDELIAN
GLRB	Hereditary hyperekplexia	ORPHA:3197	UNKNOWN
GLRX5	Spasticity, childhood-onset, with hyperglycinemia	OMIM:616859	MENDELIAN
GLRX5	Anemia, sideroblastic, 3, pyridoxine-refractory	OMIM:616860	MENDELIAN
GLRX5	Childhood-onset spasticity with hyperglycinemia	ORPHA:401866	UNKNOWN
GLS	Epileptic encephalopathy, early infantile, 71	OMIM:618328	MENDELIAN
GLS	Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development	OMIM:618339	MENDELIAN
GLS	Global developmental delay, progressive ataxia, and elevated glutamine	OMIM:618412	MENDELIAN
GLTP	—		UNKNOWN
GLTSCR1	—		UNKNOWN
GLUD1	Hyperinsulinemic hypoglycemia, familial, 6	OMIM:606762	MENDELIAN
GLUD1	Hyperinsulinism-hyperammonemia syndrome	ORPHA:35878	UNKNOWN
GLUD2	—		UNKNOWN
GLUL	Glutamine deficiency, congenital	OMIM:610015	MENDELIAN
GLUL	Developmental and epileptic encephalopathy 116	OMIM:620806	MENDELIAN
GLYCTK	D-glyceric aciduria	OMIM:220120	MENDELIAN
GLYCTK	D-glyceric aciduria	ORPHA:941	UNKNOWN
GM2A	GM2-gangliosidosis, AB variant	OMIM:272750	MENDELIAN
GM2A	GM2 gangliosidosis, AB variant	ORPHA:309246	UNKNOWN
GMDS	—		UNKNOWN
GMIP	—		UNKNOWN
GMPPA	Alacrima, achalasia, and impaired intellectual development syndrome	OMIM:615510	MENDELIAN
GMPPA	Triple A syndrome	ORPHA:869	UNKNOWN
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14	OMIM:615350	MENDELIAN
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14	OMIM:615351	MENDELIAN
GMPPB	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14	OMIM:615352	MENDELIAN
GMPPB	Congenital muscular dystrophy with intellectual disability	ORPHA:370968	UNKNOWN
GMPPB	Congenital muscular dystrophy with cerebellar involvement	ORPHA:370959	UNKNOWN
GMPPB	Congenital myasthenic syndrome with glycosylation defect	ORPHA:353327	UNKNOWN
GMPPB	Muscle-eye-brain disease	ORPHA:588	UNKNOWN
GMPPB	GMPPB-related limb-girdle muscular dystrophy R19	ORPHA:363623	UNKNOWN
GMPS	—		UNKNOWN
GNA11	Hypocalcemia, autosomal dominant 2	OMIM:615361	MENDELIAN
GNA11	Hypocalciuric hypercalcemia, familial, type II	OMIM:145981	MENDELIAN
GNA11	Cutis marmorata telangiectatica congenita	ORPHA:1556	UNKNOWN
GNA11	Autosomal dominant hypocalcemia	ORPHA:428	UNKNOWN
GNA13	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
GNA14	Tufted angioma	ORPHA:1063	UNKNOWN
GNA14	Kaposiform hemangioendothelioma	ORPHA:2122	UNKNOWN
GNAI1	Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities	OMIM:619854	MENDELIAN
GNAI2	Ventricular tachycardia, familial	OMIM:192605	MENDELIAN
GNAI3	Auriculocondylar syndrome 1	OMIM:602483	MENDELIAN
GNAI3	Auriculocondylar syndrome	ORPHA:137888	UNKNOWN
GNAL	Dystonia 25	OMIM:615073	MENDELIAN
GNAL	Autosomal dominant focal dystonia, DYT25 type	ORPHA:329466	UNKNOWN
GNAO1	Developmental and epileptic encephalopathy 17	OMIM:615473	MENDELIAN
GNAO1	Neurodevelopmental disorder with involuntary movements	OMIM:617493	MENDELIAN
GNAO1	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
GNAQ	Nevi flammei, familial multiple	OMIM:163000	MENDELIAN
GNAQ	Sturge-Weber syndrome	OMIM:185300	MENDELIAN
GNAQ	Sturge-Weber syndrome	ORPHA:3205	UNKNOWN
GNAQ	Familial multiple nevi flammei	ORPHA:624	UNKNOWN
GNAS	Pseudohypoparathyroidism, type IB	OMIM:603233	MENDELIAN
GNAS	McCune-Albright syndrome, somatic, mosaic	OMIM:174800	MENDELIAN
GNAS	Osseous heteroplasia, progressive	OMIM:166350	MENDELIAN
GNAS	Pituitary adenoma 3, multiple types, somatic	OMIM:617686	MENDELIAN
GNAS	Pseudohypoparathyroidism, type IA	OMIM:103580	MENDELIAN
GNAS	ACTH-independent macronodular adrenal hyperplasia	OMIM:219080	MENDELIAN
GNAS	Pseudopseudohypoparathyroidism	OMIM:612463	MENDELIAN
GNAS	Pseudohypoparathyroidism, type IC	OMIM:612462	MENDELIAN
GNAS	Pseudohypoparathyroidism type 1A	ORPHA:79443	UNKNOWN
GNAS	Pseudohypoparathyroidism type 1C	ORPHA:79444	UNKNOWN
GNAS	Pseudopseudohypoparathyroidism	ORPHA:79445	UNKNOWN
GNAS	Progressive osseous heteroplasia	ORPHA:2762	UNKNOWN
GNAS	Cushing syndrome due to bilateral macronodular adrenocortical disease	ORPHA:189427	UNKNOWN
GNAS	Mazabraud syndrome	ORPHA:57782	UNKNOWN
GNAS	McCune-Albright syndrome	ORPHA:562	UNKNOWN
GNAS	Pseudohypoparathyroidism type 1B	ORPHA:94089	UNKNOWN
GNAS-AS1	Pseudohypoparathyroidism, type IB	OMIM:603233	MENDELIAN
GNAT1	Night blindness, congenital stationary, autosomal dominant 3	OMIM:610444	MENDELIAN
GNAT1	Night blindness, congenital stationary, type 1G	OMIM:616389	MENDELIAN
GNAT2	Achromatopsia 4	OMIM:613856	MENDELIAN
GNAT2	Progressive cone dystrophy	ORPHA:1871	UNKNOWN
GNAT2	Achromatopsia	ORPHA:49382	UNKNOWN
GNB1L	—		UNKNOWN
GNB2	Neurodevelopmental disorder with hypotonia and dysmorphic facies	OMIM:619503	MENDELIAN
GNB2	Sick sinus syndrome 4	OMIM:619464	MENDELIAN
GNB3	Night blindness, congenital stationary, type 1H	OMIM:617024	MENDELIAN
GNB3	Hypertension, essential	OMIM:145500	POLYGENIC
GNB4	Charcot-Marie-Tooth disease, dominant intermediate F	OMIM:615185	MENDELIAN
GNB5	Intellectual developmental disorder with cardiac arrhythmia	OMIM:617173	MENDELIAN
GNB5	Language delay and attention deficit-hyperactivity disorder/cognitive impairment with or without cardiac arrhythmia	OMIM:617182	MENDELIAN
GNB5	GNB5-related intellectual disability-cardiac arrhythmia syndrome	ORPHA:542306	UNKNOWN
GNE	Nonaka myopathy	OMIM:605820	MENDELIAN
GNE	Sialuria	OMIM:269921	MENDELIAN
GNE	Thrombocytopenia 12 with or without myopathy	OMIM:620757	MENDELIAN
GNE	Sialuria	ORPHA:3166	UNKNOWN
GNE	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
GNE	GNE myopathy	ORPHA:602	UNKNOWN
GNG2	—		UNKNOWN
GNG7	—		UNKNOWN
GNMT	Glycine N-methyltransferase deficiency	OMIM:606664	MENDELIAN
GNPAT	Rhizomelic chondrodysplasia punctata, type 2	OMIM:222765	MENDELIAN
GNPTAB	Mucopolipidosis II alpha/beta	OMIM:252500	MENDELIAN
GNPTAB	Mucopolipidosis III alpha/beta	OMIM:252600	MENDELIAN
GNPTAB	Mucopolipidosis type III alpha/beta	ORPHA:423461	UNKNOWN
GNPTAB	Mucopolipidosis type II	ORPHA:576	UNKNOWN
GNPTG	Mucopolipidosis III gamma	OMIM:252605	MENDELIAN
GNRH1	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
GNRH1	Hypogonadotropic hypogonadism 12 with or without anosmia	OMIM:614841	MENDELIAN
GNRH1	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
GNRHR	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
GNRHR	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
GNS	Mucopolysaccharidosis, type IIID	OMIM:252940	MENDELIAN
GOLGA3	—		UNKNOWN
GON4L	Li-Takada-Miyake syndrome	OMIM:621212	MENDELIAN
GORAB	Geroderma osteodysplasticum	OMIM:231070	MENDELIAN
GORAB	Geroderma osteodysplastica	ORPHA:2078	UNKNOWN
GOSR2	Muscular dystrophy, congenital, with or without seizures	OMIM:620166	MENDELIAN
GOSR2	Epilepsy, progressive myoclonic, 6	OMIM:614018	MENDELIAN
GP1BA	Bernard-Soulier syndrome	OMIM:231200	MENDELIAN
GP1BA	Bernard-Soulier syndrome, type A2, autosomal dominant	OMIM:153670	MENDELIAN
GP1BA	Pseudo-Von willebrand disease	OMIM:177820	MENDELIAN
GP1BA	Nonarteritic anterior ischemic optic neuropathy, susceptibility to	OMIM:258660	POLYGENIC
GP1BA	Bernard-Soulier syndrome	ORPHA:274	UNKNOWN
GP1BA	Fetal and neonatal alloimmune thrombocytopenia	ORPHA:853	UNKNOWN
GP1BA	Autosomal dominant macrothrombocytopenia	ORPHA:140957	UNKNOWN
GP1BB	Bernard-Soulier syndrome	OMIM:231200	MENDELIAN
GP1BB	Bernard-Soulier syndrome	ORPHA:274	UNKNOWN
GP1BB	Fetal and neonatal alloimmune thrombocytopenia	ORPHA:853	UNKNOWN
GP1BB	Autosomal dominant macrothrombocytopenia	ORPHA:140957	UNKNOWN
GP1BB	22q11.2 deletion syndrome	ORPHA:567	UNKNOWN
GP2	—		UNKNOWN
GP6	Bleeding disorder, platelet-type, 11	OMIM:614201	MENDELIAN
GP9	Bernard-Soulier syndrome	OMIM:231200	MENDELIAN
GP9	Bernard-Soulier syndrome	ORPHA:274	UNKNOWN
GPAM	—		UNKNOWN
GPANK1	—		UNKNOWN
GPATCH8	—		UNKNOWN
GPBAR1	—		UNKNOWN
GPC3	Wilms tumor 1	OMIM:194070	MENDELIAN
GPC3	Simpson-Golabi-Behmel syndrome, type 1	OMIM:312870	MENDELIAN
GPC3	Simpson-Golabi-Behmel syndrome	ORPHA:373	UNKNOWN
GPC3	Nephroblastoma	ORPHA:654	UNKNOWN
GPC4	Wilms tumor 1	OMIM:194070	MENDELIAN
GPC4	Keipert syndrome	OMIM:301026	MENDELIAN
GPC4	Simpson-Golabi-Behmel syndrome	ORPHA:373	UNKNOWN
GPC4	Keipert syndrome	ORPHA:2662	UNKNOWN
GPC5	—		UNKNOWN
GPC6	Omodysplasia 1	OMIM:258315	MENDELIAN
GPC6	Autosomal recessive omodysplasia	ORPHA:93329	UNKNOWN
GPD1	Hypertriglyceridemia, transient infantile	OMIM:614480	MENDELIAN
GPD1L	Brugada syndrome 2	OMIM:611777	MENDELIAN
GPD1L	Brugada syndrome	ORPHA:130	UNKNOWN
GPD2	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
GPHN	Molybdenum cofactor deficiency, complementation group C	OMIM:615501	MENDELIAN
GPHN	Hyperekplexia 1	OMIM:149400	MENDELIAN
GPHN	Hereditary hyperekplexia	ORPHA:3197	UNKNOWN
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerasedeficiency	OMIM:613470	MENDELIAN
GPI	Hemolytic anemia due to glucophosphate isomerase deficiency	ORPHA:712	UNKNOWN
GPIHBP1	Hyperlipoproteinemia, type ID	OMIM:615947	MENDELIAN
GNMB	Amyloidosis, primary localized cutaneous, 3	OMIM:617920	MENDELIAN
GPR1	—		UNKNOWN
GPR101	Pituitary adenoma 2, GH-secreting	OMIM:300943	MENDELIAN
GPR101	Chromosome Xq26.3 duplication syndrome	OMIM:300942	MENDELIAN
GPR101	Acromegaly	ORPHA:963	UNKNOWN
GPR112	—		UNKNOWN
GPR12	—		UNKNOWN
GPR124	—		UNKNOWN
GPR125	—		UNKNOWN
GPR132	—		UNKNOWN
GPR137B	—		UNKNOWN
GPR139	—		UNKNOWN
GPR143	Nystagmus 6, congenital, X-linked	OMIM:300814	MENDELIAN
GPR143	Albinism, ocular, type I	OMIM:300500	MENDELIAN
GPR143	X-linked recessive ocular albinism	ORPHA:54	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
GPR152	—		UNKNOWN
GPR179	Night blindness, congenital stationary, type 1E	OMIM:614565	MENDELIAN
GPR33	—		UNKNOWN
GPR37	—		UNKNOWN
GPR55	—		UNKNOWN
GPR56	—		UNKNOWN
GPR68	Amelogenesis imperfecta, hypomaturation type, IIA6	OMIM:617217	MENDELIAN
GPR98	—		UNKNOWN
GPRC5A	—		UNKNOWN
GPRIN3	—		UNKNOWN
GPS1	—		UNKNOWN
GPS2	—		UNKNOWN
GPSM2	Chudley-Mccullough syndrome	OMIM:604213	MENDELIAN
GPSM2	Chudley-McCullough syndrome	ORPHA:314597	UNKNOWN
GPT	—		UNKNOWN
GPT2	Neurodevelopmental disorder with microcephaly and spastic paraplegia	OMIM:616281	MENDELIAN
GPT2	Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome	ORPHA:477673	UNKNOWN
GPX1	Glutathione peroxidase deficiency	OMIM:614164	MENDELIAN
GPX4	Spondylometaphyseal dysplasia, Sedaghatian type	OMIM:250220	MENDELIAN
GPX4	Spondylometaphyseal dysplasia, Sedaghatian type	ORPHA:93317	UNKNOWN
GRB10	Silver-Russell syndrome due to maternal uniparental disomy of chromosome 7	ORPHA:96182	UNKNOWN
GREM1	Hereditary mixed polyposis syndrome	ORPHA:157794	UNKNOWN
GRHL1	—		UNKNOWN
GRHL2	Corneal dystrophy, posterior polymorphous, 4	OMIM:618031	MENDELIAN
GRHL2	Deafness, autosomal dominant nonsyndromic sensorineural 28	OMIM:608641	MENDELIAN
GRHL2	Ectodermal dysplasia/short stature syndrome	OMIM:616029	MENDELIAN
GRHL2	Posterior polymorphous corneal dystrophy	ORPHA:98973	UNKNOWN
GRHL3	Van der woude syndrome 2	OMIM:606713	MENDELIAN
GRHL3	Cleft hard palate	ORPHA:101023	UNKNOWN
GRHL3	Van der Woude syndrome	ORPHA:888	UNKNOWN
GRHL3	Submucosal cleft palate	ORPHA:155878	UNKNOWN
GRHL3	Bifid uvula	ORPHA:99771	UNKNOWN
GRHL3	Cleft velum	ORPHA:99772	UNKNOWN
GRHPR	Hyperoxaluria, primary, type II	OMIM:260000	MENDELIAN
GRHPR	Primary hyperoxaluria type 2	ORPHA:93599	UNKNOWN
GRIA1	Intellectual developmental disorder, autosomal dominant 67	OMIM:619927	MENDELIAN
GRIA1	Intellectual developmental disorder, autosomal recessive 76	OMIM:619931	MENDELIAN
GRIA2	Neurodevelopmental disorder with language impairment and behavioral abnormalities	OMIM:618917	MENDELIAN
GRIA3	Intellectual developmental disorder, X-linked syndromic, Wu type	OMIM:300699	MENDELIAN
GRIA3	X-linked intellectual disability due to GRIA3 mutations	ORPHA:364028	UNKNOWN
GRID1	—		UNKNOWN
GRID2	Spinocerebellar ataxia, autosomal recessive 18	OMIM:616204	MENDELIAN
GRID2	Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency	ORPHA:363432	UNKNOWN
GRIK1	—		UNKNOWN
GRIK2	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures	OMIM:619580	MENDELIAN
GRIK2	Intellectual developmental disorder, autosomal recessive 6	OMIM:611092	MENDELIAN
GRIK3	—		UNKNOWN
GRIK4	—		UNKNOWN
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive	OMIM:617820	MENDELIAN
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant	OMIM:614254	MENDELIAN
GRIN1	Developmental and epileptic encephalopathy 101	OMIM:619814	MENDELIAN
GRIN1	Bilateral generalized polymicrogyria	ORPHA:208447	UNKNOWN
GRIN1	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
GRIN2A	Epilepsy, focal, with speech disorder and with or without impaired intellectual development	OMIM:245570	MENDELIAN
GRIN2A	Landau-Kleffner syndrome	ORPHA:98818	UNKNOWN
GRIN2A	Rolandic epilepsy-speech dyspraxia syndrome	ORPHA:163721	UNKNOWN
GRIN2A	Self-limited epilepsy with centrotemporal spikes	ORPHA:1945	UNKNOWN
GRIN2A	Developmental and epileptic encephalopathy with spike-wave activation in sleep	ORPHA:725	UNKNOWN
GRIN2A	Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation	ORPHA:289266	UNKNOWN
GRIN2B	Intellectual developmental disorder, autosomal dominant 6, with or without seizures	OMIM:613970	MENDELIAN
GRIN2B	Developmental and epileptic encephalopathy 27	OMIM:616139	MENDELIAN
GRIN2B	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
GRIN3A	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
GRIP1	Fraser syndrome 3	OMIM:617667	MENDELIAN
GRIP1	Fraser syndrome	ORPHA:2052	UNKNOWN
GRK1	Oguchi disease-2	OMIM:613411	MENDELIAN
GRK1	Oguchi disease	ORPHA:75382	UNKNOWN
GRK4	—		UNKNOWN
GRK5	—		UNKNOWN
GRM1	Spinocerebellar ataxia 44	OMIM:617691	MENDELIAN
GRM1	Spinocerebellar ataxia, autosomal recessive 13	OMIM:614831	MENDELIAN
GRM1	Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency	ORPHA:324262	UNKNOWN
GRM3	—		UNKNOWN
GRM5	—		UNKNOWN
GRM6	Night blindness, congenital stationary, type 1B	OMIM:257270	MENDELIAN
GRM7	Neurodevelopmental disorder with seizures, hypotonia, and brain imaging abnormalities	OMIM:618922	MENDELIAN
GRM7	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
GRM8	—		UNKNOWN
GRN	Ceroid lipofuscinosis, neuronal, 11	OMIM:614706	MENDELIAN
GRN	Frontotemporal lobar degeneration with TDP43 inclusions	OMIM:607485	MENDELIAN
GRN	Behavioral variant of frontotemporal dementia	ORPHA:275864	UNKNOWN
GRN	Progressive non-fluent aphasia	ORPHA:100070	UNKNOWN
GRN	Semantic dementia	ORPHA:100069	UNKNOWN
GRPR	—		UNKNOWN
GRXCR1	Deafness, autosomal recessive 25	OMIM:613285	MENDELIAN
GRXCR2	Deafness, autosomal recessive 101	OMIM:615837	MENDELIAN
GSC	Short stature, auditory canal atresia, mandibular hypoplasia, and skeletal abnormalities	OMIM:602471	MENDELIAN
GSDMA	—		UNKNOWN
GSDMB	—		UNKNOWN
GSE1	—		UNKNOWN
GSK3B	—		UNKNOWN
GSN	Amyloidosis, Finnish type	OMIM:105120	MENDELIAN
GSN	AGel amyloidosis	ORPHA:85448	UNKNOWN
GSPT1	—		UNKNOWN
GSPT2	—		UNKNOWN
GSR	Hemolytic anemia due to glutathione reductase deficiency	OMIM:618660	MENDELIAN
GSS	Glutathione synthetase deficiency	OMIM:266130	MENDELIAN
GSS	Hemolytic anemia due to glutathione synthetase deficiency	OMIM:231900	MENDELIAN
GSTA1	—		UNKNOWN
GSTA2	—		UNKNOWN
GSTA3	—		UNKNOWN
GSTK1	—		UNKNOWN
GSTM1	—		UNKNOWN
GSTM3	Cystic fibrosis	ORPHA:586	UNKNOWN
GSTM4	—		UNKNOWN
GSTO1	—		UNKNOWN
GSTO2	—		UNKNOWN
GSTP1	—		UNKNOWN
GSTT1	—		UNKNOWN
GSTT2	—		UNKNOWN
GSTT2B	—		UNKNOWN
GSTZ1	Maleylacetoacetate isomerase deficiency	OMIM:617596	MENDELIAN
GTDC2	—		UNKNOWN
GTF2E1	—		UNKNOWN
GTF2H1	—		UNKNOWN
GTF2H2	—		UNKNOWN
GTF2H2C	—		UNKNOWN
GTF2H3	—		UNKNOWN
GTF2H4	Xeroderma pigmentosum, complementation group J	OMIM:621435	MENDELIAN
GTF2H5	Trichothiodystrophy 3, photosensitive	OMIM:616395	MENDELIAN
GTF2H5	Trichothiodystrophy	ORPHA:33364	UNKNOWN
GTF2I	Williams syndrome	ORPHA:904	UNKNOWN
GTF2IRD1	Williams syndrome	ORPHA:904	UNKNOWN
GTF2IRD2	Williams syndrome	ORPHA:904	UNKNOWN
GTPBP3	Combined oxidative phosphorylation deficiency 23	OMIM:616198	MENDELIAN
GTPBP3	Combined oxidative phosphorylation defect type 23	ORPHA:444013	UNKNOWN
GUCA1A	Cone dystrophy 3	OMIM:602093	MENDELIAN
GUCA1A	Central areolar choroidal dystrophy	ORPHA:75377	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
GUCA1A	Cone rod dystrophy	ORPHA:1872	UNKNOWN
GUCA1A	Progressive cone dystrophy	ORPHA:1871	UNKNOWN
GUCA1B	Retinitis pigmentosa 48	OMIM:613827	MENDELIAN
GUCA1B	Retinitis pigmentosa	ORPHA:791	UNKNOWN
GUCY1A2	—		UNKNOWN
GUCY1A3	—		UNKNOWN
GUCY2C	Diarrhea 6	OMIM:614616	MENDELIAN
GUCY2C	Meconium ileus	OMIM:614665	MENDELIAN
GUCY2D	Leber congenital amaurosis, type I	OMIM:204000	MENDELIAN
GUCY2D	Choroidal dystrophy, central areolar 1	OMIM:215500	MENDELIAN
GUCY2D	Cone-Rod dystrophy 6	OMIM:601777	MENDELIAN
GUCY2D	Night blindness, congenital stationary, type 11	OMIM:618555	MENDELIAN
GUCY2D	Leber congenital amaurosis	ORPHA:65	UNKNOWN
GUCY2D	Central areolar choroidal dystrophy	ORPHA:75377	UNKNOWN
GUCY2D	Cone rod dystrophy	ORPHA:1872	UNKNOWN
GUCY2F	—		UNKNOWN
GULOP	—		UNKNOWN
GUSB	Mucopolysaccharidosis VII	OMIM:253220	MENDELIAN
GUSB	Mucopolysaccharidosis type 7	ORPHA:584	UNKNOWN
GYG1	Polyglucosan body myopathy 2	OMIM:616199	MENDELIAN
GYG1	Glycogen storage disease XV	OMIM:613507	MENDELIAN
GYG1	Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency	ORPHA:263297	UNKNOWN
GYG2	—		UNKNOWN
GYLTL1B	—		UNKNOWN
GYPC	Hereditary elliptocytosis	ORPHA:288	UNKNOWN
GYPE	—		UNKNOWN
GYS1	Glycogen storage disease 0, muscle	OMIM:611556	MENDELIAN
GYS1	Glycogen storage disease due to muscle and heart glycogen synthase deficiency	ORPHA:137625	UNKNOWN
GYS2	Glycogen storage disease 0, liver	OMIM:240600	MENDELIAN
GYS2	Glycogen storage disease due to hepatic glycogen synthase deficiency	ORPHA:2089	UNKNOWN
GZMB	—		UNKNOWN
H1-4	Rahman syndrome	OMIM:617537	MENDELIAN
H19	Wilms tumor 1	OMIM:194070	MENDELIAN
H19	Silver-Russell syndrome due to an imprinting defect of 11p15	ORPHA:231140	UNKNOWN
H19	Silver-Russell syndrome due to 11p15 microduplication	ORPHA:231144	UNKNOWN
H19	Nephroblastoma	ORPHA:654	UNKNOWN
H19	Isolated hemihyperplasia	ORPHA:2128	UNKNOWN
H2AFX	—		UNKNOWN
H2BFWT	—		UNKNOWN
H2BW1	—		UNKNOWN
H3-3B	Bryant-Li-Bhoj neurodevelopmental syndrome 2	OMIM:619721	MENDELIAN
H3F3A	—		UNKNOWN
H3F3AP4	—		UNKNOWN
H3F3B	—		UNKNOWN
H3F3C	—		UNKNOWN
H4C11	Tessadori-van Haaften neurodevelopmental syndrome 2	OMIM:619759	MENDELIAN
H4C3	Tessadori-van Haaften neurodevelopmental syndrome 1	OMIM:619758	MENDELIAN
H4C5	Tessadori-van Haaften neurodevelopmental syndrome 3	OMIM:619950	MENDELIAN
H6PD	Cortisone reductase deficiency 1	OMIM:604931	MENDELIAN
H6PD	Hyperandrogenism due to cortisone reductase deficiency	ORPHA:168588	UNKNOWN
HABP2	Thyroid cancer, nonmedullary, 5	OMIM:616535	POLYGENIC
HABP2	Thrombophiliavenous thromboembolism, included	OMIM:188050	POLYGENIC
HABP2	Familial papillary or follicular thyroid carcinoma	ORPHA:319487	UNKNOWN
HACD1	Myopathy, congenital, nonprogressive	OMIM:619967	MENDELIAN
HACD1	Congenital fiber-type disproportion myopathy	ORPHA:2020	UNKNOWN
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures	OMIM:616756	MENDELIAN
HACE1	Neuroblastoma	ORPHA:635	UNKNOWN
HACE1	Spastic paraplegia-severe developmental delay-epilepsy syndrome	ORPHA:464282	UNKNOWN
HADH	3-Hydroxyacyl-CoA dehydrogenase deficiency	OMIM:231530	MENDELIAN
HADH	Hyperinsulinemic hypoglycemia, familial, 4	OMIM:609975	MENDELIAN
HADH	Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency	ORPHA:71212	UNKNOWN
HADHA	Mitochondrial trifunctional protein deficiency	OMIM:609015	MENDELIAN
HADHA	Long-Chain 3-hydroxyacyl-coa dehydrogenase deficiency	OMIM:609016	MENDELIAN
HADHA	Mitochondrial trifunctional protein deficiency	ORPHA:746	UNKNOWN
HADHA	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	ORPHA:5	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
HADHB	Mitochondrial trifunctional protein deficiency 2	OMIM:620300	MENDELIAN
HADHB	Mitochondrial trifunctional protein deficiency	ORPHA:746	UNKNOWN
HAGH	Glyoxalase II deficiency	OMIM:614033	MENDELIAN
HAL	Histidinemia	OMIM:235800	MENDELIAN
HAL	Histidinemia	ORPHA:2157	UNKNOWN
HAMP	Hemochromatosis, type 2B	OMIM:613313	MENDELIAN
HAMP	HJV or HAMP-related hemochromatosis	ORPHA:79230	UNKNOWN
HAND1	—		UNKNOWN
HAND2	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
HAP1	—		UNKNOWN
HAPLN1	—		UNKNOWN
HARS	—		UNKNOWN
HARS2	Perrault syndrome 2	OMIM:614926	MENDELIAN
HAS1	—		UNKNOWN
HAS2	—		UNKNOWN
HAUS7	—		UNKNOWN
HAVCR1	—		UNKNOWN
HAX1	Neutropenia, severe congenital 3, autosomal recessive	OMIM:610738	MENDELIAN
HBA1	Hemoglobin H disease	OMIM:613978	MENDELIAN
HBA1	Methemoglobinemia, Alpha type	OMIM:617973	MENDELIAN
HBA1	Erythrocytosis 7	OMIM:617981	MENDELIAN
HBA1	Heinz body anemias	OMIM:140700	MENDELIAN
HBA1	Alpha-thalassemia	OMIM:604131	MENDELIAN
HBA1	Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16	ORPHA:98791	UNKNOWN
HBA1	Hemoglobin Bart's fetalis syndrome	ORPHA:163596	UNKNOWN
HBA2	Hemoglobin H disease	OMIM:613978	MENDELIAN
HBA2	Erythrocytosis 7	OMIM:617981	MENDELIAN
HBA2	Heinz body anemias	OMIM:140700	MENDELIAN
HBA2	Alpha-thalassemia	OMIM:604131	MENDELIAN
HBA2	Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16	ORPHA:98791	UNKNOWN
HBA2	Hemoglobin Bart's fetalis syndrome	ORPHA:163596	UNKNOWN
HBB	Beta-thalassemia	OMIM:613985	MENDELIAN
HBB	Fetal hemoglobin quantitative trait locus 1	OMIM:141749	MENDELIAN
HBB	Methemoglobinemia, Beta type	OMIM:617971	MENDELIAN
HBB	Erythrocytosis 6	OMIM:617980	MENDELIAN
HBB	Heinz body anemias	OMIM:140700	MENDELIAN
HBB	Sickle cell anemia	OMIM:603903	MENDELIAN
HBB	Thalassemia-beta, dominant inclusion-body	OMIM:603902	MENDELIAN
HBB	Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome	ORPHA:46532	UNKNOWN
HBB	Sickle cell anemia	ORPHA:232	UNKNOWN
HBB	Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome	ORPHA:251380	UNKNOWN
HBB	Hemoglobin D disease	ORPHA:90039	UNKNOWN
HBB	Hemoglobin E disease	ORPHA:2133	UNKNOWN
HBB	Delta-beta-thalassemia	ORPHA:231237	UNKNOWN
HBB	Hemoglobin C-beta-thalassemia syndrome	ORPHA:231242	UNKNOWN
HBB	Beta-thalassemia major	ORPHA:231214	UNKNOWN
HBB	Beta-thalassemia intermedia	ORPHA:231222	UNKNOWN
HBB	Unstable beta globin chain variant disease	ORPHA:231226	UNKNOWN
HBD	Delta-beta-thalassemia	ORPHA:231237	UNKNOWN
HBE1	—		UNKNOWN
HBEGF	—		UNKNOWN
HBG1	Fetal hemoglobin quantitative trait locus 1	OMIM:141749	MENDELIAN
HBG1	Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome	ORPHA:46532	UNKNOWN
HBG1	Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome	ORPHA:251380	UNKNOWN
HBG1	Delta-beta-thalassemia	ORPHA:231237	UNKNOWN
HBG2	Cyanosis, transient neonatal	OMIM:613977	MENDELIAN
HBG2	Fetal hemoglobin quantitative trait locus 1	OMIM:141749	MENDELIAN
HBG2	Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome	ORPHA:46532	UNKNOWN
HBG2	Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome	ORPHA:251380	UNKNOWN
HBM	—		UNKNOWN
HBQ1	—		UNKNOWN
HBS1L	—		UNKNOWN
HBZ	—		UNKNOWN
HCAR1	—		UNKNOWN
HCCS	Linear skin defects with multiple congenital anomalies 1	OMIM:309801	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
HCCS	Microphthalmia with linear skin defects syndrome	ORPHA:2556	UNKNOWN
HCFC1	Methylmalonic acidemia and homocysteinemia, Cblx type	OMIM:309541	MENDELIAN
HCK	Autoinflammation with pulmonary and cutaneous vasculitis	OMIM:620296	MENDELIAN
HCLS1	—		UNKNOWN
HCN1	Epileptic encephalopathy, early infantile, 24	OMIM:615871	MENDELIAN
HCN1	Generalized epilepsy with febrile seizures plus, type 10	OMIM:618482	MENDELIAN
HCN1	Genetic epilepsy with febrile seizure plus	ORPHA:36387	UNKNOWN
HCN1	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
HCN2	Febrile seizures, familial, 2	OMIM:602477	MENDELIAN
HCN3	—		UNKNOWN
HCN4	Epilepsy, idiopathic generalized, susceptibility to, 18	OMIM:619521	POLYGENIC
HCN4	Brugada syndrome 8	OMIM:613123	MENDELIAN
HCN4	Sick sinus syndrome 2	OMIM:163800	MENDELIAN
HCN4	Brugada syndrome	ORPHA:130	UNKNOWN
HCP5	—		UNKNOWN
HCRT	Narcolepsy 1	OMIM:161400	MENDELIAN
HCRT	Narcolepsy type 2	ORPHA:83465	UNKNOWN
HCRT	Narcolepsy type 1	ORPHA:2073	UNKNOWN
HCRTR1	—		UNKNOWN
HCRTR2	—		UNKNOWN
HDAC1	—		UNKNOWN
HDAC4	Chromosome 2q37 deletion syndrome	OMIM:600430	MENDELIAN
HDAC4	Neurodevelopmental disorder with central hypotonia and dysmorphic facies	OMIM:619797	MENDELIAN
HDAC4	2q37 microdeletion syndrome	ORPHA:1001	UNKNOWN
HDAC6	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia	OMIM:300863	MENDELIAN
HDAC6	X-linked dominant chondrodysplasia, Chassaing-Lacombe type	ORPHA:163966	UNKNOWN
HDAC8	Cornelia de Lange syndrome 5	OMIM:300882	MENDELIAN
HDAC8	Cornelia de Lange syndrome	ORPHA:199	UNKNOWN
HDAC8	Wilson-Turner syndrome	ORPHA:3459	UNKNOWN
HDAC9	Auriculocondylar syndrome 4	OMIM:620457	MENDELIAN
HDC	Gilles de la tourette syndrome	OMIM:137580	POLYGENIC
HDGFRP2	—		UNKNOWN
HDLBP	—		UNKNOWN
HDX	—		UNKNOWN
HEATR2	—		UNKNOWN
HECTD4	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum	OMIM:620250	MENDELIAN
HELQ	—		UNKNOWN
HENMT1	—		UNKNOWN
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A	OMIM:613925	MENDELIAN
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development	OMIM:613926	MENDELIAN
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 1	OMIM:604004	MENDELIAN
HEPACAM	Macrocephaly-intellectual disability-autism syndrome	ORPHA:210548	UNKNOWN
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts	ORPHA:2478	UNKNOWN
HEPACAM2	—		UNKNOWN
HEPH	—		UNKNOWN
HERC1	Macrocephaly, dysmorphic facies, and psychomotor retardation	OMIM:617011	MENDELIAN
HERC1	Megalencephaly-severe kyphoscoliosis-overgrowth syndrome	ORPHA:457359	UNKNOWN
HERC2	Prader-Willi syndrome	OMIM:176270	MENDELIAN
HERC2	Intellectual developmental disorder, autosomal recessive 38	OMIM:615516	MENDELIAN
HES1	—		UNKNOWN
HES6	—		UNKNOWN
HES7	Spondylocostal dysostosis 4, autosomal recessive	OMIM:613686	MENDELIAN
HES7	Autosomal recessive spondylocostal dysostosis	ORPHA:2311	UNKNOWN
HESX1	Septooptic dysplasia	OMIM:182230	MENDELIAN
HESX1	Septo-optic dysplasia spectrum	ORPHA:3157	UNKNOWN
HESX1	Pituitary stalk interruption syndrome	ORPHA:95496	UNKNOWN
HESX1	Combined pituitary hormone deficiencies, genetic forms	ORPHA:95494	UNKNOWN
HESX1	Hypothyroidism due to deficient transcription factors involved in pituitary development or function	ORPHA:226307	UNKNOWN
HESX1	Kallmann syndrome	ORPHA:478	UNKNOWN
HEXA	Tay-Sachs disease	OMIM:272800	MENDELIAN
HEXB	Sandhoff disease	OMIM:268800	MENDELIAN
HEXB	Sandhoff disease, juvenile form	ORPHA:309162	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
HEXB	Sandhoff disease, adult form	ORPHA:309169	UNKNOWN
HEXB	Sandhoff disease, infantile form	ORPHA:309155	UNKNOWN
HEY1	—		UNKNOWN
HEY2	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
HFE	Hemochromatosis, type 1	OMIM:235200	MENDELIAN
HFE	Symptomatic form of HFE-related hemochromatosis	ORPHA:465508	UNKNOWN
HFE	Cystic fibrosis	ORPHA:586	UNKNOWN
HFE2	—		UNKNOWN
HFM1	Premature ovarian failure 9	OMIM:615724	MENDELIAN
HGD	Alkaptonuria	OMIM:203500	MENDELIAN
HGD	Alkaptonuria	ORPHA:56	UNKNOWN
HGF	Deafness, congenital neurosensory, autosomal recessive 39	OMIM:608265	MENDELIAN
HGSNAT	Mucopolysaccharidosis type IIIC	OMIM:252930	MENDELIAN
HGSNAT	Retinitis pigmentosa 73	OMIM:616544	MENDELIAN
HGSNAT	Retinitis pigmentosa	ORPHA:791	UNKNOWN
HHEX	—		UNKNOWN
HHIP	—		UNKNOWN
HIBADH	—		UNKNOWN
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency	OMIM:250620	MENDELIAN
HIBCH	Neurodegeneration due to 3-hydroxyisobutryl-CoA hydrolase deficiency	ORPHA:88639	UNKNOWN
HID1	Developmental and epileptic encephalopathy 105 with hypopituitarism	OMIM:619983	MENDELIAN
HIF1A	—		UNKNOWN
HIF1AN	—		UNKNOWN
HIGD2A	—		UNKNOWN
HINT1	Neuromyotonia and axonal neuropathy, autosomal recessive	OMIM:137200	MENDELIAN
HINT1	Autosomal recessive axonal neuropathy with neuromyotonia	ORPHA:324442	UNKNOWN
HIP1	—		UNKNOWN
HIST1H1C	—		UNKNOWN
HIST1H2AE	—		UNKNOWN
HIST1H2BD	—		UNKNOWN
HIST1H3A	—		UNKNOWN
HIST1H3B	—		UNKNOWN
HIST1H3C	—		UNKNOWN
HIST1H3D	—		UNKNOWN
HIST1H3E	—		UNKNOWN
HIST1H3F	—		UNKNOWN
HIST1H3G	—		UNKNOWN
HIST1H3H	—		UNKNOWN
HIST1H3I	—		UNKNOWN
HIST1H3J	—		UNKNOWN
HIST1H4B	—		UNKNOWN
HIST2H3A	—		UNKNOWN
HIST2H3C	—		UNKNOWN
HIST2H3D	—		UNKNOWN
HIST2H4A	—		UNKNOWN
HIST3H3	—		UNKNOWN
HIVEP2	Intellectual developmental disorder, autosomal dominant 43	OMIM:616977	MENDELIAN
HK1	Hemolytic anemia, nonspherocytic, due to hexokinase deficiency	OMIM:235700	MENDELIAN
HK1	Retinitis pigmentosa 79	OMIM:617460	MENDELIAN
HK1	Neurodevelopmental disorder with visual defects and brain anomalies	OMIM:618547	MENDELIAN
HK1	Neuropathy, hereditary motor and sensory, Russe type	OMIM:605285	MENDELIAN
HK1	Charcot-Marie-Tooth disease type 4G	ORPHA:99953	UNKNOWN
HK2	—		UNKNOWN
HLA-A	Birdshot chorioretinopathy	ORPHA:179	UNKNOWN
HLA-B	Spondyloarthropathy, susceptibility to, 1	OMIM:106300	POLYGENIC
HLA-B	Giant cell arteritis	ORPHA:397	UNKNOWN
HLA-B	Reactive arthritis	ORPHA:29207	UNKNOWN
HLA-B	Takayasu arteritis	ORPHA:3287	UNKNOWN
HLA-B	Behçet disease	ORPHA:117	UNKNOWN
HLA-B	Stevens-Johnson syndrome	ORPHA:36426	UNKNOWN
HLA-C	Psoriasis 1, susceptibility to	OMIM:177900	POLYGENIC
HLA-DMB	—		UNKNOWN
HLA-DOA	—		UNKNOWN
HLA-DPB1	Chronic beryllium disease	ORPHA:133	UNKNOWN
HLA-DPB1	Granulomatosis with polyangiitis	ORPHA:900	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
HLA-DPB2	—		UNKNOWN
HLA-DQA1	Celiac disease, susceptibility to, 1	OMIM:212750	POLYGENIC
HLA-DQA1	Idiopathic achalasia	ORPHA:930	UNKNOWN
HLA-DQB1	Creutzfeldt-Jakob disease	OMIM:123400	POLYGENIC
HLA-DQB1	Multiple sclerosis, susceptibility to	OMIM:126200	POLYGENIC
HLA-DQB1	Celiac disease, susceptibility to, 1	OMIM:212750	POLYGENIC
HLA-DQB1	Narcolepsy type 2	ORPHA:83465	UNKNOWN
HLA-DQB1	Idiopathic achalasia	ORPHA:930	UNKNOWN
HLA-DQB1	Bullous pemphigoid	ORPHA:703	UNKNOWN
HLA-DQB1	Narcolepsy type 1	ORPHA:2073	UNKNOWN
HLA-DRA	Graham Little-Piccardi-Lassueur syndrome	ORPHA:505	UNKNOWN
HLA-DRB1	Sarcoidosis, susceptibility to, 1	OMIM:181000	POLYGENIC
HLA-DRB1	Multiple sclerosis, susceptibility to	OMIM:126200	POLYGENIC
HLA-DRB1	Giant cell arteritis	ORPHA:397	UNKNOWN
HLA-DRB1	Limited cutaneous systemic sclerosis	ORPHA:220402	UNKNOWN
HLA-DRB1	Narcolepsy type 2	ORPHA:83465	UNKNOWN
HLA-DRB1	Diffuse cutaneous systemic sclerosis	ORPHA:220393	UNKNOWN
HLA-DRB1	Systemic-onset juvenile idiopathic arthritis	ORPHA:85414	UNKNOWN
HLA-DRB1	Sarcoidosis	ORPHA:797	UNKNOWN
HLA-DRB1	Autoimmune pulmonary alveolar proteinosis	ORPHA:747	UNKNOWN
HLA-DRB1	Bullous pemphigoid	ORPHA:703	UNKNOWN
HLA-DRB1	Follicular lymphoma	ORPHA:545	UNKNOWN
HLA-DRB1	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
HLA-DRB1	Narcolepsy type 1	ORPHA:2073	UNKNOWN
HLA-DRB1	Vogt-Koyanagi-Harada disease	ORPHA:3437	UNKNOWN
HLA-DRB5	—		UNKNOWN
HLA-E	—		UNKNOWN
HLA-G	Asthma, susceptibility to	OMIM:600807	POLYGENIC
HLCS	Holocarboxylase synthetase deficiency	OMIM:253270	MENDELIAN
HLCS	Holocarboxylase synthetase deficiency	ORPHA:79242	UNKNOWN
HLTF	—		UNKNOWN
HMBS	Porphyria, acute intermittent	OMIM:176000	MENDELIAN
HMBS	Encephalopathy, porphyria-related	OMIM:620704	MENDELIAN
HMBS	Leukoencephalopathy, porphyria-related	OMIM:620711	MENDELIAN
HMBS	Acute intermittent porphyria	ORPHA:79276	UNKNOWN
HMCN1	Macular degeneration, age-related, 1	OMIM:603075	POLYGENIC
HMG20B	—		UNKNOWN
HMGA1	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
HMGA2	Silver-Russell syndrome 5	OMIM:618908	MENDELIAN
HMGA2	Well-differentiated liposarcoma	ORPHA:99971	UNKNOWN
HMGA2	Silver-Russell syndrome due to a point mutation	ORPHA:397590	UNKNOWN
HMGA2	12q14 microdeletion syndrome	ORPHA:94063	UNKNOWN
HMGB1	—		UNKNOWN
HMGB3	Microphthalmia, syndromic 13	OMIM:300915	MENDELIAN
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	OMIM:246450	MENDELIAN
HMGCL	3-hydroxy-3-methylglutaric aciduria	ORPHA:20	UNKNOWN
HMGCR	Muscular dystrophy, limb-girdle, autosomal recessive 28	OMIM:620375	MENDELIAN
HMGCS2	HMG-CoA synthase-2 deficiency	OMIM:605911	MENDELIAN
HMGCS2	3-hydroxy-3-methylglutaryl-CoA synthase deficiency	ORPHA:35701	UNKNOWN
HMHA1	—		UNKNOWN
HMMR	Breast cancer	OMIM:114480	POLYGENIC
HMOX1	Pulmonary disease, chronic obstructive	OMIM:606963	POLYGENIC
HMOX1	Heme oxygenase-1 deficiency	OMIM:614034	MENDELIAN
HMOX1	Heme oxygenase-1 deficiency	ORPHA:562509	UNKNOWN
HMOX1	Cystic fibrosis	ORPHA:586	UNKNOWN
HMOX2	—		UNKNOWN
HMSD	—		UNKNOWN
HMX1	Oculoauricular syndrome	OMIM:612109	MENDELIAN
HMX2	—		UNKNOWN
HNF1A	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
HNF1A	Hepatic adenomas, familial	OMIM:142330	MENDELIAN
HNF1A	Diabetes mellitus, insulin-dependent-1	OMIM:222100	POLYGENIC
HNF1A	Diabetes mellitus, insulin-dependent, 20	OMIM:612520	MENDELIAN
HNF1A	Renal cell carcinoma, nonpapillary	OMIM:144700	MENDELIAN
HNF1A	Maturity-onset diabetes of the young, type III	OMIM:600496	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
HNF1A	Hyperinsulinism due to HNF1A deficiency	ORPHA:324575	UNKNOWN
HNF1A	MODY	ORPHA:552	UNKNOWN
HNF1B	Renal cysts and diabetes syndrome	OMIM:137920	MENDELIAN
HNF1B	Diabetes mellitus, noninsulin-dependent	OMIM:125853	MENDELIAN
HNF1B	Renal cell carcinoma, nonpapillary	OMIM:144700	POLYGENIC
HNF1B	Mayer-Rokitansky-K ¹ / ₄ ster-Hauser syndrome type 2	ORPHA:2578	UNKNOWN
HNF1B	HNF1B-related autosomal dominant tubulointerstitial kidney disease	ORPHA:93111	UNKNOWN
HNF1B	Medullary sponge kidney	ORPHA:1309	UNKNOWN
HNF1B	17q12 microdeletion syndrome	ORPHA:261265	UNKNOWN
HNF4A	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
HNF4A	Maturity-onset diabetes of the young, type 1	OMIM:125850	MENDELIAN
HNF4A	Fanconi renotubular syndrome 4 with maturity-onset diabetes of the young	OMIM:616026	MENDELIAN
HNF4A	Congenital hyperinsulinism due to HNF4A deficiency	ORPHA:263455	UNKNOWN
HNF4A	MODY	ORPHA:552	UNKNOWN
HNMT	Asthma, susceptibility to	OMIM:600807	POLYGENIC
HNMT	Intellectual developmental disorder, autosomal recessive 51	OMIM:616739	MENDELIAN
HNRNPA1	Myopathy, distal 3	OMIM:610099	MENDELIAN
HNRNPA1	Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3	OMIM:615424	MENDELIAN
HNRNPA1	Amyotrophic lateral sclerosis 20	OMIM:615426	MENDELIAN
HNRNPA1	HNRNPA1-related adult-onset distal myopathy	ORPHA:399086	UNKNOWN
HNRNPA1	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	ORPHA:52430	UNKNOWN
HNRNPA1	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
HNRNPA2B1	Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2	OMIM:615422	MENDELIAN
HNRNPA2B1	Oculopharyngeal muscular dystrophy 2	OMIM:620460	MENDELIAN
HNRNPA2B1	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	ORPHA:52430	UNKNOWN
HNRNPD	—		UNKNOWN
HNRNPDL	Limb-girdle muscular dystrophy, type 1G	OMIM:609115	MENDELIAN
HNRNPH2	Intellectual developmental disorder, X-linked syndromic, Bain type	OMIM:300986	MENDELIAN
HNRNPH3	—		UNKNOWN
HNRNPK	Au-Kline syndrome	OMIM:616580	MENDELIAN
HNRNPK	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome due to 9q21.3 microdeletion	ORPHA:352665	UNKNOWN
HNRNPK	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome due to a point mutation	ORPHA:453504	UNKNOWN
HNRNPR	Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities	OMIM:620073	MENDELIAN
HNRNPU	Epileptic encephalopathy, early infantile, 54	OMIM:617391	MENDELIAN
HNRNPU	1q44 microdeletion syndrome	ORPHA:238769	UNKNOWN
HNRNPUL2	—		UNKNOWN
HOGA1	Hyperoxaluria, primary, type III	OMIM:613616	MENDELIAN
HOGA1	Primary hyperoxaluria type 3	ORPHA:93600	UNKNOWN
HOMER2	Deafness, autosomal dominant 68	OMIM:616707	MENDELIAN
HOMEZ	—		UNKNOWN
HORMAD1	—		UNKNOWN
HORMAD2	—		UNKNOWN
HOXA1	Athabaskan brainstem dysgenesis syndrome	OMIM:601536	MENDELIAN
HOXA10	—		UNKNOWN
HOXA11	Radioulnar synostosis with amegakaryocytic thrombocytopenia	OMIM:605432	MENDELIAN
HOXA11	Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome	ORPHA:71289	UNKNOWN
HOXA13	Hand-foot-genital syndrome	OMIM:140000	MENDELIAN
HOXA13	Preaxial deficiency, postaxial polydactyly, and hypospadias	OMIM:176305	MENDELIAN
HOXA13	Hand-foot-genital syndrome	ORPHA:2438	UNKNOWN
HOXA2	Microtia, hearing impairment, and cleft palate	OMIM:612290	MENDELIAN
HOXA2	Microtia	ORPHA:83463	UNKNOWN
HOXA3	—		UNKNOWN
HOXA4	—		UNKNOWN
HOXB1	Facial paresis, hereditary congenital, 3	OMIM:614744	MENDELIAN
HOXB6	—		UNKNOWN
HOXB8	—		UNKNOWN
HOXC13	Ectodermal dysplasia 9, hair/nail type	OMIM:614931	MENDELIAN
HOXD10	Vertical talus, congenital	OMIM:192950	MENDELIAN
HOXD13	Brachydactyly-Syndactyly syndrome	OMIM:610713	MENDELIAN
HOXD13	Synpolydactyly 1	OMIM:186000	MENDELIAN
HOXD13	Brachydactyly, type E	OMIM:113300	MENDELIAN
HOXD13	Syndactyly, type V	OMIM:186300	MENDELIAN
HOXD13	Brachydactyly, type D	OMIM:113200	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
HOXD13	VACTERL/VATER association	ORPHA:887	UNKNOWN
HOXD13	Brachydactyly type E	ORPHA:93387	UNKNOWN
HOXD13	Syndactyly type 5	ORPHA:93406	UNKNOWN
HOXD13	Brachydactyly-syndactyly, Zhao type	ORPHA:93409	UNKNOWN
HOXD4	—		UNKNOWN
HPCA	Dystonia 2, torsion, autosomal recessive	OMIM:224500	MENDELIAN
HPCA	Primary dystonia, DYT2 type	ORPHA:99657	UNKNOWN
HPCAL4	—		UNKNOWN
HPD	Hawkinsinuria	OMIM:140350	MENDELIAN
HPD	Tyrosinemia, type III	OMIM:276710	MENDELIAN
HPD	Tyrosinemia type 3	ORPHA:69723	UNKNOWN
HPD	Hawkinsinuria	ORPHA:2118	UNKNOWN
HPGD	Hypertrophic osteoarthropathy, primary, autosomal recessive 1	OMIM:259100	MENDELIAN
HPGD	Digital clubbing, isolated congenital	OMIM:119900	MENDELIAN
HPGD	Pachydermoperiostosis	ORPHA:2796	UNKNOWN
HPGD	Cranio-osteoarthropathy	ORPHA:1525	UNKNOWN
HPGD	Isolated nail clubbing	ORPHA:217059	UNKNOWN
HPRT1	Lesch-Nyhan syndrome	OMIM:300322	MENDELIAN
HPRT1	Gout, hprt-related	OMIM:300323	MENDELIAN
HPRT1	Lesch-Nyhan syndrome	ORPHA:510	UNKNOWN
HPRT1	Hypoxanthine guanine phosphoribosyltransferase partial deficiency	ORPHA:79233	UNKNOWN
HPS1	Hermansky-Pudlak syndrome 1	OMIM:203300	MENDELIAN
HPS3	Hermansky-Pudlak syndrome 3	OMIM:614072	MENDELIAN
HPS4	Hermansky-Pudlak syndrome 4	OMIM:614073	MENDELIAN
HPS5	Hermansky-Pudlak syndrome 5	OMIM:614074	MENDELIAN
HPS6	Hermansky-Pudlak syndrome 6	OMIM:614075	MENDELIAN
HPSE2	Urofacial syndrome	OMIM:236730	MENDELIAN
HPSE2	Urofacial syndrome	ORPHA:2704	UNKNOWN
HR	Atrichia with papular lesions	OMIM:209500	MENDELIAN
HR	Alopecia universalis congenita	OMIM:203655	MENDELIAN
HR	Alopecia universalis	ORPHA:701	UNKNOWN
HR	Marie Unna hereditary hypotrichosis	ORPHA:444	UNKNOWN
HRAS	Epidermal nevus, somatic	OMIM:162900	MENDELIAN
HRAS	Costello syndrome	OMIM:218040	MENDELIAN
HRAS	Thyroid cancer, nonmedullary, 2	OMIM:188470	MENDELIAN
HRAS	Schimmelpenning-Feuerstein-Mims syndrome	OMIM:163200	MENDELIAN
HRAS	Bladder cancer	OMIM:109800	MENDELIAN
HRAS	Melanocytic nevus syndrome, congenital	OMIM:137550	MENDELIAN
HRAS	Woolly hair nevus	ORPHA:79414	UNKNOWN
HRAS	Phakomatosis pigmentokeratotica	ORPHA:2874	UNKNOWN
HRAS	Linear nevus sebaceus syndrome	ORPHA:2612	UNKNOWN
HRAS	Costello syndrome	ORPHA:3071	UNKNOWN
HRC	—		UNKNOWN
HRG	Thrombophilia due to histidine-rich glycoprotein deficiency	OMIM:613116	MENDELIAN
HRH2	—		UNKNOWN
HRH3	—		UNKNOWN
HS1BP3	—		UNKNOWN
HS6ST1	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
HS6ST1	Hypogonadotropic hypogonadism 15 with or without anosmia	OMIM:614880	POLYGENIC
HS6ST1	Kallmann syndrome	ORPHA:478	UNKNOWN
HS6ST1	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
HSD11B1	Cortisone reductase deficiency 2	OMIM:614662	MENDELIAN
HSD11B1	Hyperandrogenism due to cortisone reductase deficiency	ORPHA:168588	UNKNOWN
HSD11B2	Apparent mineralocorticoid excess	OMIM:218030	MENDELIAN
HSD11B2	Apparent mineralocorticoid excess	ORPHA:320	UNKNOWN
HSD17B1	—		UNKNOWN
HSD17B10	HSD10 mitochondrial disease	OMIM:300438	MENDELIAN
HSD17B10	HSD10 disease, atypical type	ORPHA:85295	UNKNOWN
HSD17B10	HSD10 disease, neonatal type	ORPHA:391457	UNKNOWN
HSD17B10	HSD10 disease, infantile type	ORPHA:391428	UNKNOWN
HSD17B2	—		UNKNOWN
HSD17B3	Pseudohermaphroditism, male, with gynecomastia	OMIM:264300	MENDELIAN
HSD17B3	46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	ORPHA:752	UNKNOWN
HSD17B4	D-bifunctional protein deficiency	OMIM:261515	MENDELIAN
HSD17B4	Perrault syndrome 1	OMIM:233400	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
HSD3B1	—		UNKNOWN
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	OMIM:201810	MENDELIAN
HSD3B2	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	ORPHA:90791	UNKNOWN
HSD3B7	Bile acid synthesis defect, congenital, 1	OMIM:607765	MENDELIAN
HSD3B7	Congenital bile acid synthesis defect type 1	ORPHA:79301	UNKNOWN
HSF1	—		UNKNOWN
HSF2	—		UNKNOWN
HSF4	Cataract, lamellar	OMIM:116800	MENDELIAN
HSFY1	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
HSN2	—		UNKNOWN
HSP90AA1	—		UNKNOWN
HSP90B1	—		UNKNOWN
HSPA1A	—		UNKNOWN
HSPA1B	—		UNKNOWN
HSPA1L	—		UNKNOWN
HSPA5	—		UNKNOWN
HSPA8	—		UNKNOWN
HSPA9	Anemia, sideroblastic, autosomal dominant	OMIM:182170	MENDELIAN
HSPA9	Even-Plus syndrome	OMIM:616854	MENDELIAN
HSPB1	Neuronopathy, distal hereditary motor, type IIB	OMIM:608634	MENDELIAN
HSPB1	Charcot-Marie-Tooth disease, axonal, type 2F	OMIM:606595	MENDELIAN
HSPB1	Autosomal dominant Charcot-Marie-Tooth disease type 2F	ORPHA:99940	UNKNOWN
HSPB3	Neuronopathy, distal hereditary motor, type IIC	OMIM:613376	MENDELIAN
HSPB6	—		UNKNOWN
HSPB7	—		UNKNOWN
HSPB8	Myopathy, myofibrillar, 13, with rimmed vacuoles	OMIM:621078	MENDELIAN
HSPB8	Neuronopathy, distal hereditary motor, type IIA	OMIM:158590	MENDELIAN
HSPB8	Charcot-Marie-Tooth disease, axonal, type 2L	OMIM:608673	MENDELIAN
HSPD1	Spastic paraplegia 13, autosomal dominant	OMIM:605280	MENDELIAN
HSPD1	Leukodystrophy, hypomyelinating, 4	OMIM:612233	MENDELIAN
HSPD1	Autosomal dominant spastic paraplegia type 13	ORPHA:100994	UNKNOWN
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type	OMIM:224410	MENDELIAN
HSPG2	Schwartz-jampel syndrome, type 1	OMIM:255800	MENDELIAN
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type	ORPHA:1865	UNKNOWN
HSPG2	Schwartz-Jampel syndrome	ORPHA:800	UNKNOWN
HSPG2	1p36 deletion syndrome	ORPHA:1606	UNKNOWN
HTN3	—		UNKNOWN
HTR1A	Periodic fever, menstrual cycle-dependent	OMIM:614674	MENDELIAN
HTR1B	—		UNKNOWN
HTR2A	Obsessive-Compulsive disorder 1	OMIM:164230	POLYGENIC
HTR2A	Major depressive disorder	OMIM:608516	POLYGENIC
HTR2A	Schizophrenia	OMIM:181500	POLYGENIC
HTR2B	—		UNKNOWN
HTR2C	—		UNKNOWN
HTR3A	—		UNKNOWN
HTR3B	—		UNKNOWN
HTR3C	—		UNKNOWN
HTR3E	—		UNKNOWN
HTR5A	—		UNKNOWN
HTR6	—		UNKNOWN
HTR7	—		UNKNOWN
HTRA1	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2	OMIM:616779	MENDELIAN
HTRA1	Autosomal recessive cerebral arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL)	OMIM:600142	MENDELIAN
HTRA1	Cerebral autosomal recessive arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:199354	UNKNOWN
HTRA2	Parkinson disease 13	OMIM:610297	POLYGENIC
HTRA2	3-methylglutaconic aciduria, type VIII	OMIM:617248	MENDELIAN
HTRA2	Young-onset Parkinson disease	ORPHA:2828	UNKNOWN
HTT	Huntington disease	OMIM:143100	MENDELIAN
HTT	Lopes-Maciel-Rodan syndrome	OMIM:617435	MENDELIAN
HTT	Huntington disease	ORPHA:399	UNKNOWN
HTT	Juvenile Huntington disease	ORPHA:248111	UNKNOWN
HUS1	—		UNKNOWN
HUS1B	—		UNKNOWN
HUWE1	Intellectual developmental disorder, X-linked syndromic, Turner type	OMIM:309590	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
HVCN1	—		UNKNOWN
HYAL1	Mucopolysaccharidosis type IX	OMIM:601492	MENDELIAN
HYAL1	Hyaluronidase deficiency	ORPHA:67041	UNKNOWN
HYDIN	Ciliary dyskinesia, primary, 5	OMIM:608647	MENDELIAN
HYDIN	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
HYLS1	Hydrolethalus syndrome 1	OMIM:236680	MENDELIAN
HYLS1	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
HYLS1	Hydrolethalus	ORPHA:2189	UNKNOWN
HYMAI	Diabetes mellitus, transient neonatal, 1	OMIM:601410	MENDELIAN
HYMAI	Transient neonatal diabetes mellitus	ORPHA:99886	UNKNOWN
HYMAI	Paternal uniparental disomy of chromosome 6 syndrome	ORPHA:96191	UNKNOWN
IAPP	—		UNKNOWN
IARS2	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	OMIM:616007	MENDELIAN
IARS2	Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome	ORPHA:436174	UNKNOWN
IARS2	Leigh syndrome	ORPHA:506	UNKNOWN
IBA57	Spastic paraplegia 74, autosomal recessive	OMIM:616451	MENDELIAN
IBA57	Multiple mitochondrial dysfunctions syndrome 3	OMIM:615330	MENDELIAN
IBA57	Multiple mitochondrial dysfunctions syndrome type 3	ORPHA:363424	UNKNOWN
IBA57	Autosomal recessive spastic paraplegia type 74	ORPHA:468661	UNKNOWN
IBSP	—		UNKNOWN
ICAM5	—		UNKNOWN
ICK	—		UNKNOWN
ICOS	Immunodeficiency, common variable, 1	OMIM:607594	MENDELIAN
ICOS	Immunodeficiency, common variable, 2	OMIM:240500	MENDELIAN
ICOSLG	Immunodeficiency 119	OMIM:620825	MENDELIAN
ID3	—		UNKNOWN
ID4	—		UNKNOWN
IDE	—		UNKNOWN
IDH1	Glioma susceptibility 1	OMIM:137800	POLYGENIC
IDH1	Ollier disease	ORPHA:296	UNKNOWN
IDH1	Maffucci syndrome	ORPHA:163634	UNKNOWN
IDH1	Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria	ORPHA:99646	UNKNOWN
IDH2	D-2-hydroxyglutaric aciduria 2	OMIM:613657	MENDELIAN
IDH2	Ollier disease	ORPHA:296	UNKNOWN
IDH2	Maffucci syndrome	ORPHA:163634	UNKNOWN
IDH2	D-2-hydroxyglutaric aciduria	ORPHA:79315	UNKNOWN
IDH3B	Retinitis pigmentosa 46	OMIM:612572	MENDELIAN
IDH3B	Retinitis pigmentosa	ORPHA:791	UNKNOWN
IDO1	—		UNKNOWN
IDS	Mucopolysaccharidosis, type II	OMIM:309900	MENDELIAN
IDUA	Hurler syndrome	OMIM:607014	MENDELIAN
IDUA	Scheie syndrome	OMIM:607016	MENDELIAN
IDUA	Hurler-Scheie syndrome	OMIM:607015	MENDELIAN
IDUA	Scheie syndrome	ORPHA:93474	UNKNOWN
IDUA	Hurler syndrome	ORPHA:93473	UNKNOWN
IDUA	Hurler-Scheie syndrome	ORPHA:93476	UNKNOWN
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome	OMIM:614231	MENDELIAN
IFI30	—		UNKNOWN
IFI44L	—		UNKNOWN
IFIH1	Singleton-Merten syndrome 1	OMIM:182250	MENDELIAN
IFIH1	Aicardi-Goutieres syndrome 7	OMIM:615846	MENDELIAN
IFIH1	Immunodeficiency 95	OMIM:619773	MENDELIAN
IFIH1	Aicardi-Gouti�res syndrome	ORPHA:51	UNKNOWN
IFITM5	Osteogenesis imperfecta, type V	OMIM:610967	MENDELIAN
IFNA1	—		UNKNOWN
IFNA10	—		UNKNOWN
IFNA17	—		UNKNOWN
IFNA2	—		UNKNOWN
IFNAR1	Immunodeficiency 106, susceptibility to viral infections	OMIM:619935	MENDELIAN
IFNAR2	Immunodeficiency 45	OMIM:616669	MENDELIAN
IFNB1	—		UNKNOWN
IFNG	Aplastic anemia	OMIM:609135	POLYGENIC
IFNG	Immunodeficiency 69, mycobacteriosis	OMIM:618963	MENDELIAN
IFNG	Tuberous sclerosis-2	OMIM:613254	POLYGENIC

Gene_symbol	Disease_name	Disease_ID	Association_type
IFNG	Idiopathic aplastic anemia	ORPHA:88	UNKNOWN
IFNG	Tuberous sclerosis complex	ORPHA:805	UNKNOWN
IFNGR1	Immunodeficiency 27A, mycobacteriosis, AR	OMIM:209950	MENDELIAN
IFNGR1	Immunodeficiency 27B	OMIM:615978	MENDELIAN
IFNGR1	Helicobacter pylori infection, susceptibility to	OMIM:600263	POLYGENIC
IFNGR1	Behçet disease	ORPHA:117	UNKNOWN
IFNGR2	Immunodeficiency 28	OMIM:614889	MENDELIAN
IFNL4	—		UNKNOWN
IFRD1	Spinocerebellar ataxia type 18	ORPHA:98771	UNKNOWN
IFT122	Cranioectodermal dysplasia	OMIM:218330	MENDELIAN
IFT122	Cranioectodermal dysplasia	ORPHA:1515	UNKNOWN
IFT140	Retinitis pigmentosa 80	OMIM:617781	MENDELIAN
IFT140	Polycystic kidney disease 9, susceptibility to	OMIM:621164	POLYGENIC
IFT140	Cranioectodermal dysplasia 5	OMIM:621180	MENDELIAN
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly	OMIM:266920	MENDELIAN
IFT140	Leber congenital amaurosis	ORPHA:65	UNKNOWN
IFT140	Retinitis pigmentosa	ORPHA:791	UNKNOWN
IFT140	Autosomal dominant polycystic kidney disease	ORPHA:730	UNKNOWN
IFT140	Jeune syndrome	ORPHA:474	UNKNOWN
IFT172	Retinitis pigmentosa 71	OMIM:616394	MENDELIAN
IFT172	Short-Rib thoracic dysplasia 10 with or without polydactyly	OMIM:615630	MENDELIAN
IFT172	Bardet-Biedl syndrome 20	OMIM:619471	MENDELIAN
IFT172	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
IFT172	Retinitis pigmentosa	ORPHA:791	UNKNOWN
IFT172	Jeune syndrome	ORPHA:474	UNKNOWN
IFT27	Bardet-Biedl syndrome 19	OMIM:615996	MENDELIAN
IFT27	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
IFT43	Short-Rib thoracic dysplasia 18 with polydactyly	OMIM:617866	MENDELIAN
IFT43	Retinitis pigmentosa 81	OMIM:617871	MENDELIAN
IFT43	Cranioectodermal dysplasia 3	OMIM:614099	MENDELIAN
IFT43	Cranioectodermal dysplasia	ORPHA:1515	UNKNOWN
IFT43	Retinitis pigmentosa	ORPHA:791	UNKNOWN
IFT80	Asphyxiating thoracic dystrophy 2	OMIM:611263	MENDELIAN
IFT80	Short rib-polydactyly syndrome, Verma-Naumoff type	ORPHA:93271	UNKNOWN
IFT80	Jeune syndrome	ORPHA:474	UNKNOWN
IFT88	Retinitis pigmentosa	ORPHA:791	UNKNOWN
IGBP1	Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia	OMIM:300472	MENDELIAN
IGBP1	Corpus callosum agenesis-intellectual disability-coloboma-micrognathia syndrome	ORPHA:52055	UNKNOWN
IGF1	Insulin-Like growth factor I deficiency	OMIM:608747	MENDELIAN
IGF1	Growth delay due to insulin-like growth factor type 1 deficiency	ORPHA:73272	UNKNOWN
IGF1R	Insulin-Like growth factor I, resistance to	OMIM:270450	MENDELIAN
IGF1R	Growth delay due to insulin-like growth factor I resistance	ORPHA:73273	UNKNOWN
IGF2	Silver-Russell syndrome 3	OMIM:616489	MENDELIAN
IGF2	Silver-Russell syndrome 1	OMIM:180860	MENDELIAN
IGF2	Beckwith-Wiedemann syndrome	OMIM:130650	MENDELIAN
IGF2	Wilms tumor 1	OMIM:194070	MENDELIAN
IGF2	Silver-Russell syndrome due to an imprinting defect of 11p15	ORPHA:231140	UNKNOWN
IGF2	Silver-Russell syndrome due to 11p15 microduplication	ORPHA:231144	UNKNOWN
IGF2	Silver-Russell syndrome due to a point mutation	ORPHA:397590	UNKNOWN
IGF2	Isolated hemihyperplasia	ORPHA:2128	UNKNOWN
IGF2BP2	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
IGF2R	Hepatocellular carcinoma	OMIM:114550	MENDELIAN
IGFALS	Acid-labile subunit, deficiency of	OMIM:615961	MENDELIAN
IGFALS	Short stature due to primary acid-labile subunit deficiency	ORPHA:140941	UNKNOWN
IGFBP1	—		UNKNOWN
IGFBP3	—		UNKNOWN
IGFBP5	—		UNKNOWN
IGFBP7	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis	OMIM:614224	MENDELIAN
IGHA1	—		UNKNOWN
IGHG2	Recurrent infections associated with rare immunoglobulin isotypes deficiency	ORPHA:183675	UNKNOWN
IGHM	Agammaglobulinemia 1, autosomal recessive	OMIM:601495	MENDELIAN
IGHM	Autosomal non-syndromic agammaglobulinemia	ORPHA:33110	UNKNOWN
IGHMBP2	Spinal muscular atrophy, distal, autosomal recessive, 1	OMIM:604320	MENDELIAN
IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S	OMIM:616155	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
IGKC	Immunoglobulin kappa light chain deficiency	OMIM:614102	MENDELIAN
IGKC	Recurrent infections associated with rare immunoglobulin isotypes deficiency	ORPHA:183675	UNKNOWN
IGKV	—		UNKNOWN
IGLL1	Agammaglobulinemia 2, autosomal recessive	OMIM:613500	MENDELIAN
IGLL1	Autosomal non-syndromic agammaglobulinemia	ORPHA:33110	UNKNOWN
IGSF1	Hypothyroidism, central, and testicular enlargement	OMIM:300888	MENDELIAN
IGSF3	Lacrimal duct defect	OMIM:149700	MENDELIAN
IGSF8	—		UNKNOWN
IHH	Brachydactyly, type A1	OMIM:112500	MENDELIAN
IHH	Acrocapitofemoral dysplasia	OMIM:607778	MENDELIAN
IHH	Acrocapitofemoral dysplasia	ORPHA:63446	UNKNOWN
IHH	Brachydactyly type A1	ORPHA:93388	UNKNOWN
IKBIP	—		UNKNOWN
IKBKAP	—		UNKNOWN
IKBKB	Immunodeficiency 15	OMIM:615592	MENDELIAN
IKBKB	Immunodeficiency 15A	OMIM:618204	MENDELIAN
IKBKE	—		UNKNOWN
IKBKG	Incontinentia pigmenti	OMIM:308300	MENDELIAN
IKBKG	Autoinflammatory disease, systemic, X-linked	OMIM:301081	MENDELIAN
IKBKG	Ectodermal dysplasia and immunodeficiency 1	OMIM:300291	MENDELIAN
IKBKG	Immunodeficiency 33	OMIM:300636	MENDELIAN
IKBKG	Hypohidrotic ectodermal dysplasia with immunodeficiency	ORPHA:98813	UNKNOWN
IKBKG	Incontinentia pigmenti	ORPHA:464	UNKNOWN
IKZF1	Immunodeficiency, common variable, 13	OMIM:616873	MENDELIAN
IKZF1	Stevens-Johnson syndrome	ORPHA:36426	UNKNOWN
IKZF3	Immunodeficiency 84	OMIM:619437	MENDELIAN
IL10	Rheumatoid arthritis	OMIM:180300	POLYGENIC
IL10	Behçet disease	ORPHA:117	UNKNOWN
IL10	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
IL10RA	Inflammatory bowel disease 28, early onset, autosomal recessive	OMIM:613148	MENDELIAN
IL10RB	Inflammatory bowel disease 25, early onset, autosomal recessive	OMIM:612567	MENDELIAN
IL11	—		UNKNOWN
IL11RA	Craniosynostosis and dental anomalies	OMIM:614188	MENDELIAN
IL12A	Behçet disease	ORPHA:117	UNKNOWN
IL12A	Primary biliary cholangitis	ORPHA:186	UNKNOWN
IL12B	Immunodeficiency 29	OMIM:614890	MENDELIAN
IL12B	Takayasu arteritis	ORPHA:3287	UNKNOWN
IL12RB1	Immunodeficiency 30	OMIM:614891	MENDELIAN
IL12RB1	Primary biliary cholangitis	ORPHA:186	UNKNOWN
IL12RB1	Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency	ORPHA:319552	UNKNOWN
IL12RB2	—		UNKNOWN
IL13	Asthma, susceptibility to	OMIM:600807	POLYGENIC
IL15	—		UNKNOWN
IL16	—		UNKNOWN
IL17A	—		UNKNOWN
IL17F	Candidiasis, familial, 6, autosomal dominant	OMIM:613956	MENDELIAN
IL17F	Chronic mucocutaneous candidiasis	ORPHA:1334	UNKNOWN
IL17RA	Immunodeficiency 51	OMIM:613953	MENDELIAN
IL17RA	Chronic mucocutaneous candidiasis	ORPHA:1334	UNKNOWN
IL17RB	—		UNKNOWN
IL17RD	Hypogonadotropic hypogonadism 18 with or without anosmia	OMIM:615267	MENDELIAN
IL17RD	Kallmann syndrome	ORPHA:478	UNKNOWN
IL17REL	—		UNKNOWN
IL18	—		UNKNOWN
IL18R1	—		UNKNOWN
IL18RAP	—		UNKNOWN
IL19	—		UNKNOWN
IL1A	—		UNKNOWN
IL1B	Gastric cancer, somatic	OMIM:613659	POLYGENIC
IL1R1	Chronic recurrent multifocal osteomyelitis	OMIM:259680	MENDELIAN
IL1RAPL1	Intellectual developmental disorder, X-linked 21	OMIM:300143	MENDELIAN
IL1RL1	—		UNKNOWN
IL1RN	Interleukin 1 receptor antagonist deficiency	OMIM:612852	MENDELIAN
IL1RN	Gastric cancer, somatic	OMIM:613659	POLYGENIC
IL2	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
IL20RA	—		UNKNOWN
IL20RB	—		UNKNOWN
IL21	Immunodeficiency, common variable, 11	OMIM:615767	MENDELIAN
IL21R	Immunodeficiency 56	OMIM:615207	MENDELIAN
IL23R	Behçet disease	ORPHA:117	UNKNOWN
IL2RA	Immunodeficiency 41 with lymphoproliferation and autoimmunity	OMIM:606367	MENDELIAN
IL2RA	Diabetes mellitus, insulin-dependent, 10	OMIM:601942	POLYGENIC
IL2RA	Oligoarticular juvenile idiopathic arthritis	ORPHA:85410	UNKNOWN
IL2RA	Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	ORPHA:85408	UNKNOWN
IL2RG	Severe combined immunodeficiency, X-linked	OMIM:300400	MENDELIAN
IL2RG	Combined immunodeficiency, X-linked	OMIM:312863	MENDELIAN
IL2RG	T-B+NK- severe combined immunodeficiency due to gamma chain deficiency	ORPHA:276	UNKNOWN
IL2RG	Omenn syndrome	ORPHA:39041	UNKNOWN
IL3	—		UNKNOWN
IL31RA	Amyloidosis, primary localized cutaneous, 2	OMIM:613955	MENDELIAN
IL34	—		UNKNOWN
IL36RN	Psoriasis 14, pustular	OMIM:614204	MENDELIAN
IL36RN	Generalized pustular psoriasis	ORPHA:247353	UNKNOWN
IL4	—		UNKNOWN
IL4R	—		UNKNOWN
IL5	—		UNKNOWN
IL6	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
IL6	Arteriovenous malformation of the brain, somatic	OMIM:108010	POLYGENIC
IL6	Inflammatory bowel disease 1, Crohn disease	OMIM:266600	POLYGENIC
IL6	Diabetes mellitus, insulin-dependent-1	OMIM:222100	POLYGENIC
IL6	Kaposi sarcoma, susceptibility to	OMIM:148000	POLYGENIC
IL6	Systemic-onset juvenile idiopathic arthritis	ORPHA:85414	UNKNOWN
IL6R	Hyper-IgE recurrent infection syndrome 5, autosomal recessive	OMIM:618944	MENDELIAN
IL6ST	Hyper-IgE recurrent infection syndrome 4, autosomal recessive	OMIM:618523	MENDELIAN
IL6ST	Stuve-Wiedemann syndrome 2	OMIM:619751	MENDELIAN
IL6ST	Hyper-IgE recurrent infection syndrome 4A, autosomal dominant	OMIM:619752	MENDELIAN
IL6ST	Immunodeficiency 94 with autoinflammation and dysmorphic facies	OMIM:619750	MENDELIAN
IL7	Epidermodysplasia verruciformis, susceptibility to, 5	OMIM:618309	MENDELIAN
IL7	Inherited epidermodysplasia verruciformis	ORPHA:302	UNKNOWN
IL7R	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-positive	OMIM:608971	MENDELIAN
IL7R	T-B+NK+ severe combined immunodeficiency due to IL-7Ralpha deficiency	ORPHA:169154	UNKNOWN
IL7R	Omenn syndrome	ORPHA:39041	UNKNOWN
IL8	—		UNKNOWN
IL9	—		UNKNOWN
IL9R	—		UNKNOWN
ILDNR1	Deafness, neurosensory, autosomal recessive 42	OMIM:609646	MENDELIAN
ILK	—		UNKNOWN
IMMP2L	—		UNKNOWN
IMMT	—		UNKNOWN
IMPA2	—		UNKNOWN
IMPAD1	—		UNKNOWN
IMPDH1	Retinitis pigmentosa 10	OMIM:180105	MENDELIAN
IMPDH1	Leber congenital amaurosis 11	OMIM:613837	MENDELIAN
IMPDH1	Leber congenital amaurosis	ORPHA:65	UNKNOWN
IMPDH1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
IMPDH2	Autosomal dominant dopa-responsive dystonia	ORPHA:98808	UNKNOWN
IMPG1	Macular dystrophy, concentric annular	OMIM:153870	MENDELIAN
IMPG1	Macular dystrophy, vitelliform, 4	OMIM:616151	MENDELIAN
IMPG1	Adult-onset foveomacular vitelliform dystrophy	ORPHA:99000	UNKNOWN
IMPG1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
IMPG2	Macular dystrophy, vitelliform, 5	OMIM:616152	MENDELIAN
IMPG2	Retinitis pigmentosa 56	OMIM:613581	MENDELIAN
IMPG2	Adult-onset foveomacular vitelliform dystrophy	ORPHA:99000	UNKNOWN
IMPG2	Retinitis pigmentosa	ORPHA:791	UNKNOWN
INF2	Glomerulosclerosis, focal segmental, 5	OMIM:613237	MENDELIAN
INF2	Charcot-Marie-Tooth disease, dominant intermediate E	OMIM:614455	MENDELIAN
INF2	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
ING1	Squamous cell carcinoma, head and neck	OMIM:275355	MENDELIAN
INHA	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
INHBB	—		UNKNOWN
INMT	—		UNKNOWN
INPP4A	Neurodevelopmental disorder with growth impairment, quadriparesis, and poor or absent speech	OMIM:621354	MENDELIAN
INPP4B	—		UNKNOWN
INPP5B	—		UNKNOWN
INPP5D	—		UNKNOWN
INPP5K	Muscular dystrophy, congenital, with cataracts and intellectual disability	OMIM:617404	MENDELIAN
INPPL1	Opsismodysplasia	OMIM:258480	MENDELIAN
INPPL1	Opsismodysplasia	ORPHA:2746	UNKNOWN
INPPL1	Schneckenbecken dysplasia	ORPHA:3144	UNKNOWN
INS	Diabetes mellitus, insulin-dependent, 2	OMIM:125852	MENDELIAN
INS	Hyperproinsulinemia	OMIM:616214	MENDELIAN
INS	Diabetes mellitus, permanent neonatal 4	OMIM:618858	MENDELIAN
INS	Maturity-onset diabetes of the young, type 10	OMIM:613370	MENDELIAN
INS	Isolated permanent neonatal diabetes mellitus	ORPHA:99885	UNKNOWN
INS	MODY	ORPHA:552	UNKNOWN
INSIG1	—		UNKNOWN
INSIG2	—		UNKNOWN
INS-IGF2	—		UNKNOWN
INSL3	Cryptorchidism, unilateral or bilateral	OMIM:219050	MENDELIAN
INSL6	—		UNKNOWN
INSR	Pineal hyperplasia, insulin-resistant diabetes mellitus, and somatic abnormalities	OMIM:262190	MENDELIAN
INSR	Diabetes mellitus, insulin-resistant, with acanthosis nigricans	OMIM:610549	MENDELIAN
INSR	Donohue syndrome	OMIM:246200	MENDELIAN
INSR	Hyperinsulinemic hypoglycemia, familial, 5	OMIM:609968	MENDELIAN
INSR	Hyperinsulinism due to INSR deficiency	ORPHA:263458	UNKNOWN
INSR	Rabson-Mendenhall syndrome	ORPHA:769	UNKNOWN
INSR	Donohue syndrome	ORPHA:508	UNKNOWN
INSR	Insulin-resistance syndrome type A	ORPHA:2297	UNKNOWN
INTS1	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies	OMIM:618571	MENDELIAN
INTU	Short-Rib thoracic dysplasia 20 with polydactyly	OMIM:617925	MENDELIAN
INTU	Orofaciodigital syndrome XVII	OMIM:617926	MENDELIAN
INVS	Nephronophthisis 2	OMIM:602088	MENDELIAN
INVS	Senior-Loken syndrome	ORPHA:3156	UNKNOWN
IPP	—		UNKNOWN
IQCB1	Senior-Loken syndrome 5	OMIM:609254	MENDELIAN
IQCB1	Senior-Loken syndrome	ORPHA:3156	UNKNOWN
IQCB1	Leber congenital amaurosis	ORPHA:65	UNKNOWN
IQCE	Polydactyly, postaxial, type A7	OMIM:617642	MENDELIAN
IQGAP1	—		UNKNOWN
IQGAP2	—		UNKNOWN
IQGAP3	—		UNKNOWN
IQSEC2	Intellectual developmental disorder, X-linked 1	OMIM:309530	MENDELIAN
IQSEC2	Microduplication Xp11.22p11.23 syndrome	ORPHA:217377	UNKNOWN
IQSEC2	Severe intellectual disability-progressive postnatal microcephaly-midline stereotypic hand movements syndrome	ORPHA:397933	UNKNOWN
IRAK1	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
IRAK1	Pediatric systemic lupus erythematosus	ORPHA:93552	UNKNOWN
IRAK4	Immunodeficiency 67	OMIM:607676	MENDELIAN
IRAK4	Transient predisposition to invasive pyogenic bacterial infection	ORPHA:70592	UNKNOWN
IRF1	Immunodeficiency 117, mycobacteriosis, autosomal recessive	OMIM:620668	MENDELIAN
IRF1	Lung cancer, susceptibility to	OMIM:211980	MENDELIAN
IRF1	Gastric cancer, somatic	OMIM:613659	MENDELIAN
IRF2	—		UNKNOWN
IRF2BPL	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures	OMIM:618088	MENDELIAN
IRF4	Immunodeficiency 131	OMIM:621097	MENDELIAN
IRF4	Whipple disease	ORPHA:3452	UNKNOWN
IRF5	Limited cutaneous systemic sclerosis	ORPHA:220402	UNKNOWN
IRF5	Diffuse cutaneous systemic sclerosis	ORPHA:220393	UNKNOWN
IRF5	Primary biliary cholangitis	ORPHA:186	UNKNOWN
IRF5	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
IRF6	van der Woude syndrome 1	OMIM:119300	MENDELIAN
IRF6	Orofacial cleft 6, susceptibility to	OMIM:608864	POLYGENIC
IRF6	Popliteal pterygium syndrome	OMIM:119500	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
IRF6	Isolated cleft lip	ORPHA:199302	UNKNOWN
IRF6	Cleft lip/palate	ORPHA:199306	UNKNOWN
IRF6	Van der Woude syndrome	ORPHA:888	UNKNOWN
IRF6	Autosomal dominant popliteal pterygium syndrome	ORPHA:1300	UNKNOWN
IRF6	Cleft lip and alveolus	ORPHA:141291	UNKNOWN
IRF6	Oligodontia	ORPHA:99798	UNKNOWN
IRF7	Immunodeficiency 39	OMIM:616345	MENDELIAN
IRF8	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive	OMIM:226990	MENDELIAN
IRF8	Immunodeficiency 32A	OMIM:614893	MENDELIAN
IRF8	Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency	ORPHA:319600	UNKNOWN
IRF9	Immunodeficiency 65, susceptibility to viral infections	OMIM:618648	MENDELIAN
IRS1	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
IRS2	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
IRS4	Hypothyroidism, congenital, nongoitrous, 9	OMIM:301035	MENDELIAN
IRX4	—		UNKNOWN
IRX5	Hamamy syndrome	OMIM:611174	MENDELIAN
ISCA2	Multiple mitochondrial dysfunctions syndrome 4	OMIM:616370	MENDELIAN
ISCA2	Multiple mitochondrial dysfunctions syndrome type 4	ORPHA:457406	UNKNOWN
ISCU	Myopathy with exercise intolerance, Swedish type	OMIM:255125	MENDELIAN
ISG15	Immunodeficiency 38 with basal ganglia calcification	OMIM:616126	MENDELIAN
ISL1	Classic bladder exstrophy	ORPHA:93930	UNKNOWN
ISPD	—		UNKNOWN
ISYNA1	—		UNKNOWN
ITCH	Autoimmune disease, multisystem, with facial dysmorphism	OMIM:613385	MENDELIAN
ITCH	Syndromic multisystem autoimmune disease due to Itch deficiency	ORPHA:228426	UNKNOWN
ITGA1	—		UNKNOWN
ITGA11	—		UNKNOWN
ITGA2	Fetal and neonatal alloimmune thrombocytopenia	ORPHA:853	UNKNOWN
ITGA2B	Glanzmann thrombasthenia	OMIM:273800	MENDELIAN
ITGA2B	Bleeding disorder, platelet-type, 16	OMIM:187800	MENDELIAN
ITGA2B	Fetal and neonatal alloimmune thrombocytopenia	ORPHA:853	UNKNOWN
ITGA2B	Glanzmann thrombasthenia	ORPHA:849	UNKNOWN
ITGA2B	Autosomal dominant macrothrombocytopenia	ORPHA:140957	UNKNOWN
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	OMIM:614748	MENDELIAN
ITGA4	—		UNKNOWN
ITGA5	—		UNKNOWN
ITGA6	Epidermolysis bullosa, junctional, with pyloric atresia	OMIM:226730	MENDELIAN
ITGA6	Epidermolysis bullosa, junctional 6, with pyloric atresia	OMIM:619817	MENDELIAN
ITGA6	Junctional epidermolysis bullosa with pyloric atresia	ORPHA:79403	UNKNOWN
ITGA7	Muscular dystrophy, congenital, due to integrin alpha-7 deficiency	OMIM:613204	MENDELIAN
ITGA7	Congenital fiber-type disproportion myopathy	ORPHA:2020	UNKNOWN
ITGA8	Renal hypodysplasia/aplasia 1	OMIM:191830	MENDELIAN
ITGA8	Renal agenesis, bilateral	ORPHA:1848	UNKNOWN
ITGA9	—		UNKNOWN
ITGAE	—		UNKNOWN
ITGAM	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
ITGB1	—		UNKNOWN
ITGB2	Leukocyte adhesion deficiency, type I	OMIM:116920	MENDELIAN
ITGB3	Glanzmann thrombasthenia 2	OMIM:619267	MENDELIAN
ITGB3	Bleeding disorder, platelet-type, 24, autosomal dominant	OMIM:619271	MENDELIAN
ITGB3	Fetal and neonatal alloimmune thrombocytopenia	ORPHA:853	UNKNOWN
ITGB3	Glanzmann thrombasthenia	ORPHA:849	UNKNOWN
ITGB3	Autosomal dominant macrothrombocytopenia	ORPHA:140957	UNKNOWN
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia	OMIM:226730	MENDELIAN
ITGB4	Epidermolysis bullosa, junctional 5A, intermediate	OMIM:619816	MENDELIAN
ITGB4	Junctional epidermolysis bullosa with pyloric atresia	ORPHA:79403	UNKNOWN
ITGB4	Intermediate generalized junctional epidermolysis bullosa	ORPHA:79402	UNKNOWN
ITGB4	Epidermolysis bullosa simplex with pyloric atresia	ORPHA:158684	UNKNOWN
ITGB4	Localized junctional epidermolysis bullosa	ORPHA:251393	UNKNOWN
ITGB4	Aplasia cutis congenita	ORPHA:1114	UNKNOWN
ITGB6	Amelogenesis imperfecta, type IH	OMIM:616221	MENDELIAN
ITGB6	Alopecia-intellectual disability syndrome	ORPHA:2850	UNKNOWN
ITIH1	—		UNKNOWN
ITIH3	—		UNKNOWN
ITIH4	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
ITIH6	—		UNKNOWN
ITK	Lymphoproliferative syndrome 1	OMIM:613011	MENDELIAN
ITM2B	Cerebral amyloid angiopathy, itm2b-related, 1	OMIM:176500	MENDELIAN
ITM2B	Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities	OMIM:616079	MENDELIAN
ITM2B	Dementia, familial danish	OMIM:117300	MENDELIAN
ITPA	Epileptic encephalopathy, early infantile, 35	OMIM:616647	MENDELIAN
ITPA	Inosine triphosphatase deficiency	OMIM:613850	MENDELIAN
ITPK1	—		UNKNOWN
ITPKC	—		UNKNOWN
ITPR1	Gillespie syndrome	OMIM:206700	MENDELIAN
ITPR1	Spinocerebellar ataxia 15	OMIM:606658	MENDELIAN
ITPR1	Spinocerebellar ataxia 29, congenital nonprogressive	OMIM:117360	MENDELIAN
ITPR1	Spinocerebellar ataxia type 29	ORPHA:208513	UNKNOWN
ITPR1	Spinocerebellar ataxia type 15/16	ORPHA:98769	UNKNOWN
ITPR1	Aniridia-cerebellar ataxia-intellectual disability syndrome	ORPHA:1065	UNKNOWN
ITPR2	Anhidrosis, isolated, with normal sweat glands	OMIM:106190	MENDELIAN
ITPR3	Charcot-Marie-Tooth disease, demyelinating, type 1J	OMIM:620111	MENDELIAN
ITPR3	Immunodeficiency 133 with ectodermal dysplasia with or without peripheral neuropathy	OMIM:621254	MENDELIAN
ITSN2	—		UNKNOWN
IVD	Isovaleric acidemia	OMIM:243500	MENDELIAN
IVD	Isovaleric acidemia	ORPHA:33	UNKNOWN
IYD	Thyroid hormonogenesis, genetic defect in, 4	OMIM:274800	MENDELIAN
IYD	Familial thyroid dyshormonogenesis	ORPHA:95716	UNKNOWN
JAG1	Deafness, congenital heart defects, and posterior embryotoxon	OMIM:617992	MENDELIAN
JAG1	Charcot-Marie-Tooth disease, axonal, type 2HH	OMIM:619574	MENDELIAN
JAG1	Tetralogy of Fallot	OMIM:187500	MENDELIAN
JAG1	Alagille syndrome 1	OMIM:118450	MENDELIAN
JAG1	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
JAG2	Muscular dystrophy, limb-girdle, autosomal recessive 27	OMIM:619566	MENDELIAN
JAGN1	Neutropenia, severe congenital, 6, autosomal recessive	OMIM:616022	MENDELIAN
JAK1	Autoinflammation, immune dysregulation, and eosinophilia	OMIM:618999	MENDELIAN
JAK2	Budd-Chiari syndrome	OMIM:600880	POLYGENIC
JAK2	Erythrocytosis, familial, 1	OMIM:133100	MENDELIAN
JAK2	Myelofibrosis with myeloid metaplasia, somatic	OMIM:254450	MENDELIAN
JAK2	Thrombocythemia 3	OMIM:614521	MENDELIAN
JAK2	Polycythemia vera	OMIM:263300	MENDELIAN
JAK2	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
JAK2	Familial thrombocytosis	ORPHA:71493	UNKNOWN
JAK2	Essential thrombocythemia	ORPHA:3318	UNKNOWN
JAK2	Budd-Chiari syndrome	ORPHA:131	UNKNOWN
JAK2	Primary myelofibrosis	ORPHA:824	UNKNOWN
JAK2	Polycythemia vera	ORPHA:729	UNKNOWN
JAK3	Scid, autosomal recessive, T-Negative/b-Positive type	OMIM:600802	MENDELIAN
JAK3	T-B+NK- severe combined immunodeficiency due to JAK3 deficiency	ORPHA:35078	UNKNOWN
JAM3	Hemorrhagic destruction of the brain, subependymal calcification,and cataracts	OMIM:613730	MENDELIAN
JARID2	Developmental delay with variable intellectual disability and dysmorphic facies	OMIM:620098	MENDELIAN
JMJD1C	22q11.2 deletion syndrome	ORPHA:567	UNKNOWN
JMJD7	—		UNKNOWN
JMJD7-PLA2G4B	—		UNKNOWN
JPH2	Cardiomyopathy, dilated, 2E	OMIM:619492	MENDELIAN
JPH2	Cardiomyopathy, familial hypertrophic, 17	OMIM:613873	MENDELIAN
JPH2	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
JPH3	Huntington disease-like 2	OMIM:606438	MENDELIAN
JPH3	Huntington disease-like 2	ORPHA:98934	UNKNOWN
JRK	Juvenile myoclonic epilepsy	ORPHA:307	UNKNOWN
JRK	Childhood absence epilepsy	ORPHA:64280	UNKNOWN
JUN	—		UNKNOWN
JUNB	—		UNKNOWN
JUP	Arrhythmogenic right ventricular dysplasia 12	OMIM:611528	MENDELIAN
JUP	Naxos disease	OMIM:601214	MENDELIAN
JUP	Lethal acantholytic erosive disorder	ORPHA:158687	UNKNOWN
JUP	Naxos disease	ORPHA:34217	UNKNOWN
KAL1	—		UNKNOWN
KALRN	—		UNKNOWN
KANK1	Cerebral palsy, spastic quadriplegic, 2	OMIM:612900	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
KANK2	Nephrotic syndrome, type 16	OMIM:617783	MENDELIAN
KANK2	Palmoplantar keratoderma and woolly hair	OMIM:616099	MENDELIAN
KANK2	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
KANK4	—		UNKNOWN
KANSL1	Koolen-De Vries syndrome	OMIM:610443	MENDELIAN
KANSL2	—		UNKNOWN
KARS	—		UNKNOWN
KASH5	Premature ovarian failure 22	OMIM:620548	MENDELIAN
KASH5	Spermatogenic failure 88	OMIM:620547	MENDELIAN
KAT2A	—		UNKNOWN
KAT6A	Arboleda-Tham syndrome	OMIM:616268	MENDELIAN
KAT6A	KAT6-related intellectual disability-craniofacial anomalies-cardiac defects syndrome	ORPHA:457193	UNKNOWN
KAT6B	Genitopatellar syndrome	OMIM:606170	MENDELIAN
KAT6B	Ohdo syndrome, Say-Barber-Biesecker-Young-Simpson variant	OMIM:603736	MENDELIAN
KAT6B	Genitopatellar syndrome	ORPHA:85201	UNKNOWN
KAT6B	Blepharophimosis-intellectual disability syndrome, SBBYS type	ORPHA:3047	UNKNOWN
KATNAL1	—		UNKNOWN
KATNAL2	—		UNKNOWN
KATNB1	Lissencephaly 6, with microcephaly	OMIM:616212	MENDELIAN
KATNB1	Lissencephaly syndrome, Norman-Roberts type	ORPHA:89844	UNKNOWN
KBTBD13	Nemaline myopathy 6	OMIM:609273	MENDELIAN
KBTBD13	Childhood-onset nemaline myopathy	ORPHA:171439	UNKNOWN
KCNA1	Episodic ataxia, type 1	OMIM:160120	MENDELIAN
KCNA1	Episodic ataxia type 1	ORPHA:37612	UNKNOWN
KCNA1	Hereditary continuous muscle fiber activity	ORPHA:972	UNKNOWN
KCNA1	Paroxysmal kinesigenic dyskinesia	ORPHA:98809	UNKNOWN
KCNA1	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
KCNA2	Epileptic encephalopathy, early infantile, 32	OMIM:616366	MENDELIAN
KCNA2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
KCNA3	—		UNKNOWN
KCNA4	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum	OMIM:618284	MENDELIAN
KCNA5	Atrial fibrillation, familial, 7	OMIM:612240	MENDELIAN
KCNA5	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
KCNA6	—		UNKNOWN
KCNAB1	—		UNKNOWN
KCNAB2	1p36 deletion syndrome	ORPHA:1606	UNKNOWN
KCNB1	Developmental and epileptic encephalopathy 26	OMIM:616056	MENDELIAN
KCNB1	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
KCNB2	—		UNKNOWN
KCNC1	Epilepsy, progressive myoclonic 7	OMIM:616187	MENDELIAN
KCNC2	Developmental and epileptic encephalopathy 103	OMIM:619913	MENDELIAN
KCNC2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
KCNC3	Spinocerebellar ataxia 13	OMIM:605259	MENDELIAN
KCNC3	Spinocerebellar ataxia type 13	ORPHA:98768	UNKNOWN
KCND2	—		UNKNOWN
KCND3	Brugada syndrome 9	OMIM:616399	MENDELIAN
KCND3	Spinocerebellar ataxia 19	OMIM:607346	MENDELIAN
KCND3	Brugada syndrome	ORPHA:130	UNKNOWN
KCND3	Spinocerebellar ataxia type 19/22	ORPHA:98772	UNKNOWN
KCNE1	Jervell and Lange-Nielsen syndrome 2	OMIM:612347	MENDELIAN
KCNE1	Long QT syndrome 5	OMIM:613695	MENDELIAN
KCNE1	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
KCNE1	Jervell and Lange-Nielsen syndrome	ORPHA:90647	UNKNOWN
KCNE1L	—		UNKNOWN
KCNE2	Atrial fibrillation, familial, 4	OMIM:611493	MENDELIAN
KCNE2	Long QT syndrome 6	OMIM:613693	MENDELIAN
KCNE2	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
KCNE3	Brugada syndrome 6	OMIM:613119	MENDELIAN
KCNE3	Brugada syndrome	ORPHA:130	UNKNOWN
KCNE3	Hypokalemic periodic paralysis	ORPHA:681	UNKNOWN
KCNE4	—		UNKNOWN
KCNH1	Zimmermann-Laband syndrome 1	OMIM:135500	MENDELIAN
KCNH1	Temple-Baraitser syndrome	OMIM:611816	MENDELIAN
KCNH1	Temple-Baraitser syndrome	ORPHA:420561	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
KCNH1	Zimmermann-Laband syndrome	ORPHA:3473	UNKNOWN
KCNH2	Short QT syndrome 1	OMIM:609620	MENDELIAN
KCNH2	Long QT syndrome 2	OMIM:613688	MENDELIAN
KCNH2	Congenital short QT syndrome	ORPHA:51083	UNKNOWN
KCNH3	—		UNKNOWN
KCNH5	Developmental and epileptic encephalopathy 112	OMIM:620537	MENDELIAN
KCNH5	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
KCNIP4	—		UNKNOWN
KCNJ1	Bartter syndrome, type 2	OMIM:241200	MENDELIAN
KCNJ10	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	OMIM:600791	MENDELIAN
KCNJ10	SESAME syndrome	OMIM:612780	MENDELIAN
KCNJ10	Pendred syndrome	OMIM:274600	MENDELIAN
KCNJ10	EAST syndrome	ORPHA:199343	UNKNOWN
KCNJ10	Paroxysmal kinesigenic dyskinesia	ORPHA:98809	UNKNOWN
KCNJ10	Pendred syndrome	ORPHA:705	UNKNOWN
KCNJ11	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
KCNJ11	Maturity-onset diabetes of the young, type 13	OMIM:616329	MENDELIAN
KCNJ11	Diabetes, permanent neonatal 2, with or without neurologic features	OMIM:618856	MENDELIAN
KCNJ11	Diabetes mellitus, transient neonatal, 3	OMIM:610582	MENDELIAN
KCNJ11	Hyperinsulinemic hypoglycemia, familial, 2	OMIM:601820	MENDELIAN
KCNJ11	Isolated permanent neonatal diabetes mellitus	ORPHA:99885	UNKNOWN
KCNJ11	Transient neonatal diabetes mellitus	ORPHA:99886	UNKNOWN
KCNJ11	Autosomal recessive hyperinsulinism due to Kir6.2 deficiency	ORPHA:79644	UNKNOWN
KCNJ11	Autosomal dominant hyperinsulinism due to Kir6.2 deficiency	ORPHA:276580	UNKNOWN
KCNJ11	MODY	ORPHA:552	UNKNOWN
KCNJ11	DEND syndrome	ORPHA:79134	UNKNOWN
KCNJ12	—		UNKNOWN
KCNJ13	Leber congenital amaurosis 16	OMIM:614186	MENDELIAN
KCNJ13	Snowflake vitreoretinal degeneration	OMIM:193230	MENDELIAN
KCNJ13	Leber congenital amaurosis	ORPHA:65	UNKNOWN
KCNJ13	Severe early-childhood-onset retinal dystrophy	ORPHA:364055	UNKNOWN
KCNJ15	—		UNKNOWN
KCNJ18	Thyrotoxic periodic paralysis, susceptibility to, 2	OMIM:613239	POLYGENIC
KCNJ18	Thyrotoxic periodic paralysis	ORPHA:79102	UNKNOWN
KCNJ2	Atrial fibrillation, familial, 9	OMIM:613980	MENDELIAN
KCNJ2	Short QT syndrome 3	OMIM:609622	MENDELIAN
KCNJ2	Andersen cardiomyopathic periodic paralysis	OMIM:170390	MENDELIAN
KCNJ2	Congenital short QT syndrome	ORPHA:51083	UNKNOWN
KCNJ2	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
KCNJ2	Andersen-Tawil syndrome	ORPHA:37553	UNKNOWN
KCNJ3	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
KCNJ5	Long QT syndrome 13	OMIM:613485	MENDELIAN
KCNJ5	Hyperaldosteronism, familial, type III	OMIM:613677	MENDELIAN
KCNJ5	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
KCNJ5	Andersen-Tawil syndrome	ORPHA:37553	UNKNOWN
KCNJ5	Familial hyperaldosteronism type III	ORPHA:251274	UNKNOWN
KCNJ6	Keppen-Lubinsky syndrome	OMIM:614098	MENDELIAN
KCNJ6	Keppen-Lubinsky syndrome	ORPHA:435628	UNKNOWN
KCNJ8	CantÃ³ syndrome	ORPHA:1517	UNKNOWN
KCNJ8	Brugada syndrome	ORPHA:130	UNKNOWN
KCNJ9	—		UNKNOWN
KCNK17	—		UNKNOWN
KCNK18	Migraine, with or without aura, susceptibility to, 13	OMIM:613656	POLYGENIC
KCNK3	Developmental delay with sleep apnea	OMIM:621402	MENDELIAN
KCNK3	Pulmonary hypertension, primary, 4	OMIM:615344	MENDELIAN
KCNK6	—		UNKNOWN
KCNK9	Birk-Barel syndrome	OMIM:612292	MENDELIAN
KCNK9	Birk-Barel syndrome	ORPHA:166108	UNKNOWN
KCNMA1	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy	OMIM:609446	MENDELIAN
KCNMA1	Cerebellar atrophy, developmental delay, and seizures	OMIM:617643	MENDELIAN
KCNMA1	Liang-Wang syndrome	OMIM:618729	MENDELIAN
KCNMA1	Epilepsy, idiopathic generalized, susceptibility to, 16	OMIM:618596	POLYGENIC
KCNMA1	Generalized epilepsy-paroxysmal dyskinesia syndrome	ORPHA:79137	UNKNOWN
KCNMB1	HYPERTENSION, DIASTOLIC, RESISTANCE TO	OMIM:608622	POLYGENIC
KCNMB3	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
KCNMB4	—		UNKNOWN
KCNN2	Dystonia 34, myoclonic	OMIM:619724	MENDELIAN
KCNN2	Neurodevelopmental disorder with or without variable movement or behavioral abnormalities	OMIM:619725	MENDELIAN
KCNN3	Zimmermann-Laband syndrome 3	OMIM:618658	MENDELIAN
KCNN3	Zimmermann-Laband syndrome	ORPHA:3473	UNKNOWN
KCNQ1	Jervell and Lange-Nielsen syndrome 1	OMIM:220400	MENDELIAN
KCNQ1	Beckwith-Wiedemann syndrome	OMIM:130650	MENDELIAN
KCNQ1	Short QT syndrome 2	OMIM:609621	MENDELIAN
KCNQ1	Atrial fibrillation, familial, 3	OMIM:607554	MENDELIAN
KCNQ1	Long QT syndrome 1	OMIM:192500	MENDELIAN
KCNQ1	Congenital short QT syndrome	ORPHA:51083	UNKNOWN
KCNQ1	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
KCNQ1	Jervell and Lange-Nielsen syndrome	ORPHA:90647	UNKNOWN
KCNQ10T1	Beckwith-Wiedemann syndrome	OMIM:130650	MENDELIAN
KCNQ10T1	Isolated hemihyperplasia	ORPHA:2128	UNKNOWN
KCNQ2	Seizures, benign familial neonatal, 1	OMIM:121200	MENDELIAN
KCNQ2	Epileptic encephalopathy, early infantile, 7	OMIM:613720	MENDELIAN
KCNQ2	Self-limited infantile epilepsy	ORPHA:306	UNKNOWN
KCNQ2	Self-limited neonatal epilepsy	ORPHA:1949	UNKNOWN
KCNQ2	KCNQ2-related developmental and epileptic encephalopathy	ORPHA:439218	UNKNOWN
KCNQ2	Epilepsy of infancy with migrating focal seizures	ORPHA:293181	UNKNOWN
KCNQ2	Self-limited neonatal/infantile epilepsy	ORPHA:140927	UNKNOWN
KCNQ3	Epilepsy, benign neonatal, 2	OMIM:121201	MENDELIAN
KCNQ3	Juvenile myoclonic epilepsy	ORPHA:307	UNKNOWN
KCNQ3	Self-limited infantile epilepsy	ORPHA:306	UNKNOWN
KCNQ3	Self-limited neonatal epilepsy	ORPHA:1949	UNKNOWN
KCNQ4	Deafness, autosomal dominant nonsyndromic sensorineural 2	OMIM:600101	MENDELIAN
KCNQ5	Intellectual developmental disorder, autosomal dominant 46	OMIM:617601	MENDELIAN
KCNS1	—		UNKNOWN
KCNS3	—		UNKNOWN
KCNT1	Epilepsy, nocturnal frontal lobe, 5	OMIM:615005	MENDELIAN
KCNT1	Developmental and epileptic encephalopathy 14	OMIM:614959	MENDELIAN
KCNT1	Sleep-related hypermotor epilepsy	ORPHA:98784	UNKNOWN
KCNT1	Epilepsy of infancy with migrating focal seizures	ORPHA:293181	UNKNOWN
KCNV1	—		UNKNOWN
KCNV2	Retinal cone dystrophy 3B	OMIM:610356	MENDELIAN
KCTD1	Dental radicular dysplasia	OMIM:621434	MENDELIAN
KCTD1	Scalp-ear-nipple syndrome	OMIM:181270	MENDELIAN
KCTD1	Scalp-ear-nipple syndrome	ORPHA:2036	UNKNOWN
KCTD13	—		UNKNOWN
KCTD19	—		UNKNOWN
KCTD3	—		UNKNOWN
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions	OMIM:611726	MENDELIAN
KCTD7	Progressive myoclonic epilepsy type 3	ORPHA:263516	UNKNOWN
KDM1A	Cleft palate, psychomotor retardation, and distinctive facial features	OMIM:616728	MENDELIAN
KDM1A	ACTH-independent macronodular adrenal hyperplasia 3	OMIM:620990	POLYGENIC
KDM1A	Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome	ORPHA:477993	UNKNOWN
KDM1A	Cushing syndrome due to bilateral macronodular adrenocortical disease	ORPHA:189427	UNKNOWN
KDM2B	Neurodevelopmental disorder with congenital cardiac defects and variable renal and ocular abnormalities	OMIM:621474	MENDELIAN
KDM3A	—		UNKNOWN
KDM3B	Diets-Jongmans syndrome	OMIM:618846	MENDELIAN
KDM4C	—		UNKNOWN
KDM5A	El Hayek-Chahrour neurodevelopmental syndrome	OMIM:620820	MENDELIAN
KDM5B	Intellectual developmental disorder, autosomal recessive 65	OMIM:618109	MENDELIAN
KDM5C	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type	OMIM:300534	MENDELIAN
KDM5C	KDM5C-related syndromic X-linked intellectual disability	ORPHA:85279	UNKNOWN
KDM5D	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
KDM6A	Kabuki syndrome 1	OMIM:147920	MENDELIAN
KDM6A	Kabuki syndrome 2	OMIM:300867	MENDELIAN
KDM6A	Kabuki syndrome	ORPHA:2322	UNKNOWN
KDM6B	Stolerman neurodevelopmental syndrome	OMIM:618505	MENDELIAN
KDR	Hemangioma, capillary infantile	OMIM:602089	MENDELIAN
KDR	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
KDSR	Erythrokeratoderma variabilis et progressiva 4	OMIM:617526	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
KDSR	Erythrokeratoderma variabilis	ORPHA:317	UNKNOWN
KDSR	Progressive symmetric erythrokeratodermia	ORPHA:316	UNKNOWN
KEAP1	Familial multinodular goiter	ORPHA:276399	UNKNOWN
KERA	Cornea plana 2	OMIM:217300	MENDELIAN
KHDC3L	Hydatidiform mole, recurrent, 2	OMIM:614293	MENDELIAN
KHK	Fructosuria, essential	OMIM:229800	MENDELIAN
KHK	Essential fructosuria	ORPHA:2056	UNKNOWN
KIAA0100	—		UNKNOWN
KIAA0196	—		UNKNOWN
KIAA0226	—		UNKNOWN
KIAA0232	—		UNKNOWN
KIAA0319	—		UNKNOWN
KIAA0513	—		UNKNOWN
KIAA1033	—		UNKNOWN
KIAA1109	—		UNKNOWN
KIAA1199	—		UNKNOWN
KIAA1210	—		UNKNOWN
KIAA1217	—		UNKNOWN
KIAA1279	—		UNKNOWN
KIAA1377	—		UNKNOWN
KIAA1432	—		UNKNOWN
KIAA1462	—		UNKNOWN
KIAA1549	Retinitis pigmentosa 86	OMIM:618613	MENDELIAN
KIAA1549	Retinitis pigmentosa	ORPHA:791	UNKNOWN
KIAA1586	—		UNKNOWN
KIAA1919	—		UNKNOWN
KIAA2022	—		UNKNOWN
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development	OMIM:152950	MENDELIAN
KIF11	Microcephaly-chorioretinopathy-lymphedema syndrome	ORPHA:2526	UNKNOWN
KIF14	Meckel syndrome 12	OMIM:616258	MENDELIAN
KIF14	Microcephaly 20, primary, autosomal recessive	OMIM:617914	MENDELIAN
KIF14	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
KIF17	—		UNKNOWN
KIF18A	—		UNKNOWN
KIF1A	Spastic paraplegia 30, autosomal recessive	OMIM:610357	MENDELIAN
KIF1A	Neuropathy, hereditary sensory and autonomic, type II	OMIM:201300	MENDELIAN
KIF1A	Neuropathy, hereditary sensory, type IIC	OMIM:614213	MENDELIAN
KIF1A	NESCAV syndrome	OMIM:614255	MENDELIAN
KIF1A	Spastic paraplegia 30, autosomal recessive	OMIM:620607	MENDELIAN
KIF1A	Autosomal spastic paraplegia type 30	ORPHA:101010	UNKNOWN
KIF1A	Hereditary sensory and autonomic neuropathy type 2	ORPHA:970	UNKNOWN
KIF1B	Pheochromocytoma	OMIM:171300	MENDELIAN
KIF1B	Neuroblastoma, susceptibility to	OMIM:256700	POLYGENIC
KIF1B	Charcot-Marie-Tooth disease, axonal, type 2A1	OMIM:118210	MENDELIAN
KIF1B	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
KIF1C	Spastic ataxia 2, autosomal recessive	OMIM:611302	MENDELIAN
KIF1C	Autosomal spastic paraplegia type 58	ORPHA:397946	UNKNOWN
KIF21A	Fibrosis of extraocular muscles, congenital, 1	OMIM:135700	MENDELIAN
KIF21A	Fetal akinesia deformation sequence	ORPHA:994	UNKNOWN
KIF21A	Congenital fibrosis of extraocular muscles	ORPHA:45358	UNKNOWN
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2	OMIM:603546	MENDELIAN
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, leptodactylic type	ORPHA:93360	UNKNOWN
KIF23	Anemia, dyserythropoietic congenital, type III	OMIM:105600	MENDELIAN
KIF23	Congenital dyserythropoietic anemia type III	ORPHA:98870	UNKNOWN
KIF27	—		UNKNOWN
KIF2A	Cortical dysplasia, complex, with other brain malformations 3	OMIM:615411	MENDELIAN
KIF3C	—		UNKNOWN
KIF4A	Intellectual developmental disorder, X-linked 100	OMIM:300923	MENDELIAN
KIF5A	Amyotrophic lateral sclerosis, susceptibility to, 25	OMIM:617921	POLYGENIC
KIF5A	Spastic paraplegia 10, autosomal dominant	OMIM:604187	MENDELIAN
KIF5A	Myoclonus, intractable, neonatal	OMIM:617235	MENDELIAN
KIF5A	Autosomal dominant spastic paraplegia type 10	ORPHA:100991	UNKNOWN
KIF5C	Cortical dysplasia, complex, with other brain malformations 2	OMIM:615282	MENDELIAN
KIF6	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
KIF7	Al-Gazali-Bakalinova syndrome	OMIM:607131	MENDELIAN
KIF7	Hydrolethalus syndrome 2	OMIM:614120	MENDELIAN
KIF7	Acrocallosal syndrome	OMIM:200990	MENDELIAN
KIF7	Acrocallosal syndrome	ORPHA:36	UNKNOWN
KIF7	Orofaciodigital syndrome type 6	ORPHA:2754	UNKNOWN
KIF7	Multiple epiphyseal dysplasia-macrocephaly-facial dysmorphism syndrome	ORPHA:166024	UNKNOWN
KIF7	Hydrolethalus	ORPHA:2189	UNKNOWN
KIFAP3	—		UNKNOWN
KIR2DL1	—		UNKNOWN
KIR2DL3	—		UNKNOWN
KIR2DL4	—		UNKNOWN
KIR3DL1	—		UNKNOWN
KIR3DL2	—		UNKNOWN
KISS1	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
KISS1	Hypogonadotropic hypogonadism 13 with or without anosmia	OMIM:614842	MENDELIAN
KISS1	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
KISS1R	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
KISS1R	Precocious puberty, central	OMIM:176400	MENDELIAN
KISS1R	Hypogonadotropic hypogonadism 8 with or without anosmia	OMIM:614837	MENDELIAN
KISS1R	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
KIT	Testicular tumor, somatic	OMIM:273300	MENDELIAN
KIT	Mastocytosis, cutaneous	OMIM:154800	MENDELIAN
KIT	Piebald trait	OMIM:172800	MENDELIAN
KIT	Gastrointestinal stromal tumor	OMIM:606764	MENDELIAN
KIT	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
KIT	Cutaneous mastocytoma	ORPHA:79455	UNKNOWN
KIT	Gastrointestinal stromal tumor	ORPHA:44890	UNKNOWN
KIT	Piebaldism	ORPHA:2884	UNKNOWN
KIT	Systemic mastocytosis with associated hematologic neoplasm	ORPHA:98849	UNKNOWN
KIT	Bullous diffuse cutaneous mastocytosis	ORPHA:280785	UNKNOWN
KIT	Pseudoxanthomatous diffuse cutaneous mastocytosis	ORPHA:280794	UNKNOWN
KITLG	Hyperpigmentation, familial progressive	OMIM:145250	MENDELIAN
KITLG	Deafness, autosomal dominant 69	OMIM:616697	MENDELIAN
KITLG	Waardenburg syndrome, type 2F	OMIM:619947	MENDELIAN
KITLG	Waardenburg syndrome type 2	ORPHA:895	UNKNOWN
KIZ	Retinitis pigmentosa 69	OMIM:615780	MENDELIAN
KIZ	Retinitis pigmentosa	ORPHA:791	UNKNOWN
KL	Tumoral calcinosis, hyperphosphatemic, familial, 3	OMIM:617994	MENDELIAN
KLB	—		UNKNOWN
KLC1	—		UNKNOWN
KLF1	Anemia, congenital dyserythropoietic, type IVb	OMIM:620969	MENDELIAN
KLF1	Anemia, dyserythropoietic congenital, type IV	OMIM:613673	MENDELIAN
KLF1	Hereditary persistence of fetal hemoglobin	OMIM:613566	MENDELIAN
KLF1	Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome	ORPHA:46532	UNKNOWN
KLF1	Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome	ORPHA:251380	UNKNOWN
KLF10	—		UNKNOWN
KLF11	Maturity-onset diabetes of the young, type VII	OMIM:610508	MENDELIAN
KLF11	MODY	ORPHA:552	UNKNOWN
KLF4	—		UNKNOWN
KLF5	—		UNKNOWN
KLF6	Prostate cancer	OMIM:176807	MENDELIAN
KLF6	Gastric cancer, somatic	OMIM:613659	MENDELIAN
KLF7	—		UNKNOWN
KLF8	—		UNKNOWN
KLHDC8B	Lymphoma, hodgkin	OMIM:236000	POLYGENIC
KLHL10	Spermatogenic failure 11	OMIM:615081	MENDELIAN
KLHL10	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
KLHL15	Intellectual developmental disorder, X-linked 103	OMIM:300982	MENDELIAN
KLHL20	Neurodevelopmental disorder with early-onset seizures, facial dysmorphism, and behavioral abnormalities	OMIM:621390	MENDELIAN
KLHL3	Pseudohypoadosteronism, type IID	OMIM:614495	MENDELIAN
KLHL40	Nemaline myopathy 8	OMIM:615348	MENDELIAN
KLHL40	Severe congenital nemaline myopathy	ORPHA:171430	UNKNOWN
KLHL41	Nemaline myopathy 9	OMIM:615731	MENDELIAN
KLHL41	Typical nemaline myopathy	ORPHA:171436	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
KLHL41	Intermediate nemaline myopathy	ORPHA:171433	UNKNOWN
KLHL41	Severe congenital nemaline myopathy	ORPHA:171430	UNKNOWN
KLHL41	Childhood-onset nemaline myopathy	ORPHA:171439	UNKNOWN
KLHL6	—		UNKNOWN
KLHL7	Retinitis pigmentosa 42	OMIM:612943	MENDELIAN
KLHL7	PERCHING syndrome	OMIM:617055	MENDELIAN
KLHL7	Retinitis pigmentosa	ORPHA:791	UNKNOWN
KLHL9	KLHL9-related early-onset distal myopathy	ORPHA:399081	UNKNOWN
CLK12	—		UNKNOWN
CLK15	—		UNKNOWN
CLK3	—		UNKNOWN
CLK4	Amelogenesis imperfecta, hypomaturation type, iia1	OMIM:204700	MENDELIAN
CLK7	—		UNKNOWN
CLKB1	Prekallikrein deficiency	OMIM:612423	MENDELIAN
KLIN	Cowden syndrome 4	OMIM:615107	MENDELIAN
KLK1	—		UNKNOWN
KMT2A	Wiedemann-Steiner syndrome	OMIM:605130	MENDELIAN
KMT2A	Wiedemann-Steiner syndrome	ORPHA:319182	UNKNOWN
KMT2B	Intellectual developmental disorder, autosomal dominant 68	OMIM:619934	MENDELIAN
KMT2B	Dystonia 28, childhood-onset	OMIM:617284	MENDELIAN
KMT2B	Dystonia 28	ORPHA:589618	UNKNOWN
KMT2C	Kleefstra syndrome 2	OMIM:617768	MENDELIAN
KMT2C	Kleefstra syndrome due to a point mutation	ORPHA:261652	UNKNOWN
KMT2D	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome	OMIM:620186	MENDELIAN
KMT2D	Kabuki syndrome 1	OMIM:147920	MENDELIAN
KMT2D	Kabuki syndrome	ORPHA:2322	UNKNOWN
KMT2E	O'donnell-Luria-Rodan syndrome	OMIM:618512	MENDELIAN
KMT5B	Intellectual developmental disorder, autosomal dominant 51	OMIM:617788	MENDELIAN
KNG1	Angioedema, hereditary, 6	OMIM:619363	MENDELIAN
KNG1	Flaujeac factor deficiency	OMIM:228960	MENDELIAN
KPNA1	—		UNKNOWN
KPNA7	Oocyte/zygote/embryo maturation arrest 17	OMIM:620319	MENDELIAN
KPNA7	Female infertility due to oocyte meiotic arrest	ORPHA:488191	UNKNOWN
KPTN	Intellectual developmental disorder, autosomal recessive 41	OMIM:615637	MENDELIAN
KPTN	Macrocephaly-developmental delay syndrome	ORPHA:397612	UNKNOWN
KRAS	Cardiofaciocutaneous syndrome 2	OMIM:615278	MENDELIAN
KRAS	Cardiofaciocutaneous syndrome 1	OMIM:115150	MENDELIAN
KRAS	Arteriovenous malformation of the brain, somatic	OMIM:108010	MENDELIAN
KRAS	Pancreatic cancer	OMIM:260350	MENDELIAN
KRAS	Schimmelpenning-Feuerstein-Mims syndrome	OMIM:163200	MENDELIAN
KRAS	Breast cancer	OMIM:114480	MENDELIAN
KRAS	Bladder cancer	OMIM:109800	MENDELIAN
KRAS	Noonan syndrome 3	OMIM:609942	MENDELIAN
KRAS	Lung cancer, susceptibility to	OMIM:211980	MENDELIAN
KRAS	Gastric cancer, somatic	OMIM:613659	MENDELIAN
KRAS	Oculoectodermal syndrome	OMIM:600268	MENDELIAN
KRAS	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
KRAS	Oculoectodermal syndrome	ORPHA:3339	UNKNOWN
KRAS	Lynch syndrome	ORPHA:144	UNKNOWN
KRAS	Linear nevus sebaceus syndrome	ORPHA:2612	UNKNOWN
KRAS	Cardiofaciocutaneous syndrome	ORPHA:1340	UNKNOWN
KRAS	Familial pancreatic carcinoma	ORPHA:1333	UNKNOWN
KRAS	Encephalocraniocutaneous lipomatosis	ORPHA:2396	UNKNOWN
KRAS	Noonan syndrome	ORPHA:648	UNKNOWN
KRIT1	Cerebral cavernous malformations 1	OMIM:116860	MENDELIAN
KRIT1	Familial cerebral cavernous malformation	ORPHA:221061	UNKNOWN
KRT1	Epidermolytic hyperkeratosis	OMIM:113800	MENDELIAN
KRT1	Palmoplantar keratoderma, nonepidermolytic	OMIM:600962	MENDELIAN
KRT1	Ichthyosis, annular epidermolytic 2	OMIM:620148	MENDELIAN
KRT1	Ichthyosis hystrix, Curth-Macklin type	OMIM:146590	MENDELIAN
KRT1	Keratosis palmoplantaris striata III	OMIM:607654	MENDELIAN
KRT1	Palmoplantar keratoderma, epidermolytic, 2	OMIM:620411	MENDELIAN
KRT1	Autosomal dominant epidermolytic ichthyosis	ORPHA:312	UNKNOWN
KRT1	Ichthyosis hystrix of Curth-Macklin	ORPHA:79503	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
KRT1	KRT1-related diffuse nonepidermolytic keratoderma	ORPHA:530838	UNKNOWN
KRT1	Striate palmoplantar keratoderma	ORPHA:50942	UNKNOWN
KRT1	Epidermolytic palmoplantar keratoderma	ORPHA:2199	UNKNOWN
KRT10	Erythroderma, ichthyosiform, congenital reticular	OMIM:609165	MENDELIAN
KRT10	Epidermolytic hyperkeratosis 2	OMIM:620150	MENDELIAN
KRT10	Ichthyosis hystrix gravior	OMIM:146600	MENDELIAN
KRT10	Ichthyosis, cyclic, with epidermolytic hyperkeratosis	OMIM:607602	MENDELIAN
KRT10	Epidermolytic hyperkeratosis 2B, autosomal recessive	OMIM:620707	MENDELIAN
KRT10	Autosomal dominant epidermolytic ichthyosis	ORPHA:312	UNKNOWN
KRT12	Meesmann corneal dystrophy 1	OMIM:122100	MENDELIAN
KRT13	White sponge nevus 2	OMIM:615785	MENDELIAN
KRT14	Epidermolysis bullosa simplex, Weber-Cockayne type	OMIM:131800	MENDELIAN
KRT14	Epidermolysis bullosa simplex, Dowling-Meara type	OMIM:131760	MENDELIAN
KRT14	Epidermolysis bullosa simplex 1B, generalized intermediate	OMIM:131900	MENDELIAN
KRT14	Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive	OMIM:601001	MENDELIAN
KRT14	Naegeli-Franceschetti-Jadassohn syndrome	OMIM:161000	MENDELIAN
KRT14	Dermatopathia pigmentosa reticularis	OMIM:125595	MENDELIAN
KRT14	Localized epidermolysis bullosa simplex	ORPHA:79400	UNKNOWN
KRT14	Autosomal dominant generalized epidermolysis bullosa simplex, severe form	ORPHA:79396	UNKNOWN
KRT14	Epidermolysis bullosa simplex with mottled pigmentation	ORPHA:79397	UNKNOWN
KRT14	Autosomal dominant generalized epidermolysis bullosa simplex, intermediate form	ORPHA:79399	UNKNOWN
KRT14	Autosomal recessive generalized epidermolysis bullosa simplex	ORPHA:89838	UNKNOWN
KRT14	Naegeli-Franceschetti-Jadassohn syndrome	ORPHA:69087	UNKNOWN
KRT16	Palmoplantar keratoderma, nonepidermolytic, focal 1	OMIM:613000	MENDELIAN
KRT16	Pachyonychia congenita, type 1	OMIM:167200	MENDELIAN
KRT16	Pachyonychia congenita	ORPHA:2309	UNKNOWN
KRT16	Epidermolytic palmoplantar keratoderma	ORPHA:2199	UNKNOWN
KRT17	Steatocystoma multiplex	OMIM:184500	MENDELIAN
KRT17	Pachyonychia congenita 2	OMIM:167210	MENDELIAN
KRT17	Sebocystomatosis	ORPHA:841	UNKNOWN
KRT17	Pachyonychia congenita	ORPHA:2309	UNKNOWN
KRT18	Cirrhosis, familial	OMIM:215600	MENDELIAN
KRT2	Ichthyosis, Bullous type	OMIM:146800	MENDELIAN
KRT2	Superficial epidermolytic ichthyosis	ORPHA:455	UNKNOWN
KRT3	Meesmann corneal dystrophy 2	OMIM:618767	MENDELIAN
KRT31	—		UNKNOWN
KRT37	—		UNKNOWN
KRT38	—		UNKNOWN
KRT4	White sponge nevus 1	OMIM:193900	MENDELIAN
KRT5	Epidermolysis bullosa simplex 2E, with migratory circinate erythema	OMIM:609352	MENDELIAN
KRT5	Epidermolysis bullosa simplex, Dowling-Meara type	OMIM:131760	MENDELIAN
KRT5	Dowling-Degos disease	OMIM:179850	MENDELIAN
KRT5	Epidermolysis bullosa simplex with mottled pigmentation	OMIM:131960	MENDELIAN
KRT5	Epidermolysis bullosa simplex 2A, generalized severe	OMIM:619555	MENDELIAN
KRT5	Epidermolysis bullosa simplex 2B, generalized intermediate	OMIM:619588	MENDELIAN
KRT5	Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive	OMIM:619599	MENDELIAN
KRT5	Epidermolysis bullosa simplex 2C, localized	OMIM:619594	MENDELIAN
KRT5	Localized epidermolysis bullosa simplex	ORPHA:79400	UNKNOWN
KRT5	Autosomal dominant generalized epidermolysis bullosa simplex, severe form	ORPHA:79396	UNKNOWN
KRT5	Epidermolysis bullosa simplex with mottled pigmentation	ORPHA:79397	UNKNOWN
KRT5	Autosomal dominant generalized epidermolysis bullosa simplex, intermediate form	ORPHA:79399	UNKNOWN
KRT5	Epidermolysis bullosa simplex with circinate migratory erythema	ORPHA:158681	UNKNOWN
KRT5	Autosomal recessive generalized epidermolysis bullosa simplex	ORPHA:89838	UNKNOWN
KRT5	Dowling-Degos disease	ORPHA:79145	UNKNOWN
KRT6A	Pachyonychia congenita 3	OMIM:615726	MENDELIAN
KRT6A	Pachyonychia congenita, type 1	OMIM:167200	MENDELIAN
KRT6A	Pachyonychia congenita	ORPHA:2309	UNKNOWN
KRT6B	Pachyonychia congenita 4	OMIM:615728	MENDELIAN
KRT6B	Pachyonychia congenita 2	OMIM:167210	MENDELIAN
KRT6B	Pachyonychia congenita	ORPHA:2309	UNKNOWN
KRT6C	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse	OMIM:615735	MENDELIAN
KRT71	Hypotrichosis 13	OMIM:615896	MENDELIAN
KRT71	Woolly hair	ORPHA:170	UNKNOWN
KRT74	Hypotrichosis 3	OMIM:613981	MENDELIAN
KRT74	Woolly hair, autosomal dominant	OMIM:194300	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
KRT74	Ectodermal dysplasia 7, Hair/nail type	OMIM:614929	MENDELIAN
KRT74	Woolly hair	ORPHA:170	UNKNOWN
KRT74	Hypotrichosis simplex of the scalp	ORPHA:90368	UNKNOWN
KRT8	—		UNKNOWN
KRT81	Monilethrix 2	OMIM:621169	MENDELIAN
KRT81	Monilethrix	ORPHA:573	UNKNOWN
KRT83	Erythrokeratoderma variabilis et progressiva 5	OMIM:617756	MENDELIAN
KRT83	Monilethrix 3	OMIM:621170	MENDELIAN
KRT83	Progressive symmetric erythrokeratoderma	ORPHA:316	UNKNOWN
KRT83	Monilethrix	ORPHA:573	UNKNOWN
KRT85	Ectodermal dysplasia 4, hair/nail type	OMIM:602032	MENDELIAN
KRT86	Monilethrix	OMIM:158000	MENDELIAN
KRT86	Monilethrix	ORPHA:573	UNKNOWN
KRT9	Palmoplantar keratoderma, epidermolytic	OMIM:144200	MENDELIAN
KRT9	Epidermolytic palmoplantar keratoderma	ORPHA:2199	UNKNOWN
KRTAP1-1	—		UNKNOWN
KRTAP4-8	—		UNKNOWN
KRTAP9-3	—		UNKNOWN
KYNU	Vertebral, cardiac, renal, and limb defects syndrome 2	OMIM:617661	MENDELIAN
KYNU	Hydroxykynureninuria	OMIM:236800	MENDELIAN
KYNU	Hydroxykynureninuria	ORPHA:79155	UNKNOWN
L1CAM	Hydrocephalus due to congenital stenosis of aqueduct of sylvius	OMIM:307000	MENDELIAN
L1CAM	MASA syndrome	OMIM:303350	MENDELIAN
L1CAM	Corpus callosum, partial agenesis of, X-linked	OMIM:304100	MENDELIAN
L1CAM	X-linked complicated corpus callosum dysgenesis	ORPHA:1497	UNKNOWN
L1CAM	X-linked complicated spastic paraplegia type 1	ORPHA:306617	UNKNOWN
L1CAM	MASA syndrome	ORPHA:2466	UNKNOWN
L1CAM	Hydrocephalus with stenosis of the aqueduct of Sylvius	ORPHA:2182	UNKNOWN
L2HGDH	L-2-hydroxyglutaric aciduria	OMIM:236792	MENDELIAN
L2HGDH	L-2-hydroxyglutaric aciduria	ORPHA:79314	UNKNOWN
L3MBTL1	—		UNKNOWN
LACTB	—		UNKNOWN
LAMA1	Poretti-Boltshauser syndrome	OMIM:615960	MENDELIAN
LAMA1	Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome	ORPHA:370022	UNKNOWN
LAMA2	Muscular dystrophy, limb-girdle, autosomal recessive 23	OMIM:618138	MENDELIAN
LAMA2	Muscular dystrophy, congenital, merosin deficient or partially deficient	OMIM:607855	MENDELIAN
LAMA2	Laminin subunit alpha 2-related congenital muscular dystrophy	ORPHA:258	UNKNOWN
LAMA3	Laryngoonychocutaneous syndrome	OMIM:245660	MENDELIAN
LAMA3	Epidermolysis bullosa, junctional, Herlitz type	OMIM:226700	MENDELIAN
LAMA3	Epidermolysis bullosa, junctional 2B, severe	OMIM:619784	MENDELIAN
LAMA3	Epidermolysis bullosa, junctional 2A, intermediate	OMIM:619783	MENDELIAN
LAMA3	Severe generalized junctional epidermolysis bullosa	ORPHA:79404	UNKNOWN
LAMA3	Intermediate generalized junctional epidermolysis bullosa	ORPHA:79402	UNKNOWN
LAMA3	Laryngo-onycho-cutaneous syndrome	ORPHA:2407	UNKNOWN
LAMA4	Cardiomyopathy, dilated, 1JJ	OMIM:615235	MENDELIAN
LAMA4	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
LAMA5	Nephrotic syndrome, type 26	OMIM:620049	MENDELIAN
LAMA5	Bent bone dysplasia syndrome 2	OMIM:620076	MENDELIAN
LAMA5	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
LAMB1	Lissencephaly 5	OMIM:615191	MENDELIAN
LAMB1	Leukoencephalopathy without lacunae, adult-onset	OMIM:621424	MENDELIAN
LAMB1	Cobblestone lissencephaly without muscular or ocular involvement	ORPHA:352682	UNKNOWN
LAMB2	Pierson syndrome	OMIM:609049	MENDELIAN
LAMB2	Nephrotic syndrome, type 5, with or without ocular abnormalities	OMIM:614199	MENDELIAN
LAMB2	Pierson syndrome	ORPHA:2670	UNKNOWN
LAMB2	Synaptic congenital myasthenic syndrome	ORPHA:98915	UNKNOWN
LAMB3	Epidermolysis bullosa, junctional, Non-Herlitz type	OMIM:226650	MENDELIAN
LAMB3	Epidermolysis bullosa, junctional, Herlitz type	OMIM:226700	MENDELIAN
LAMB3	Amelogenesis imperfecta, type IA	OMIM:104530	MENDELIAN
LAMB3	Severe generalized junctional epidermolysis bullosa	ORPHA:79404	UNKNOWN
LAMB3	Intermediate generalized junctional epidermolysis bullosa	ORPHA:79402	UNKNOWN
LAMB4	—		UNKNOWN
LAMC1	—		UNKNOWN
LAMC2	Epidermolysis bullosa, junctional, Herlitz type	OMIM:226700	MENDELIAN
LAMC2	Epidermolysis bullosa, junctional 3A, intermediate	OMIM:619785	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
LAMC2	Epidermolysis bullosa, junctional 3B, severe	OMIM:619786	MENDELIAN
LAMC2	Severe generalized junctional epidermolysis bullosa	ORPHA:79404	UNKNOWN
LAMC2	Intermediate generalized junctional epidermolysis bullosa	ORPHA:79402	UNKNOWN
LAMC3	Cortical malformations, occipital	OMIM:614115	MENDELIAN
LAMP1	—		UNKNOWN
LAMP2	Danon disease	OMIM:300257	MENDELIAN
LAMP2	Danon disease	ORPHA:34587	UNKNOWN
LAMTOR2	Immunodeficiency due to defect in mapbp-interacting protein	OMIM:610798	MENDELIAN
LAMTOR2	Primary immunodeficiency syndrome due to P14/LAMTOR2 deficiency	ORPHA:90023	UNKNOWN
LAPTM4B	—		UNKNOWN
LARGE	—		UNKNOWN
LARP7	Alazami syndrome	OMIM:615071	MENDELIAN
LARP7	Alazami syndrome	ORPHA:319671	UNKNOWN
LARS	—		UNKNOWN
LARS2	Perrault syndrome 4	OMIM:615300	MENDELIAN
LARS2	Hydrops, lactic acidosis, and sideroblastic anemia	OMIM:617021	MENDELIAN
LAS1L	Wilson-Turner syndrome	OMIM:309585	MENDELIAN
LAS1L	Wilson-Turner syndrome	ORPHA:3459	UNKNOWN
LAT2	—		UNKNOWN
LATS1	—		UNKNOWN
LATS2	—		UNKNOWN
LBH	—		UNKNOWN
LBP	—		UNKNOWN
LBR	Greenberg skeletal dysplasia	OMIM:215140	MENDELIAN
LBR	Reynolds syndrome	OMIM:613471	MENDELIAN
LBR	Pelger-Huet anomaly	OMIM:169400	MENDELIAN
LBR	Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly	OMIM:618019	MENDELIAN
LBR	Greenberg dysplasia	ORPHA:1426	UNKNOWN
LBR	Reynolds syndrome	ORPHA:779	UNKNOWN
LCA5	Leber congenital amaurosis 5	OMIM:604537	MENDELIAN
LCA5	Leber congenital amaurosis	ORPHA:65	UNKNOWN
LCA5	Severe early-childhood-onset retinal dystrophy	ORPHA:364055	UNKNOWN
LCAT	Lecithin:cholesterol acyltransferase deficiency	OMIM:245900	MENDELIAN
LCAT	Fish-Eye disease	OMIM:136120	MENDELIAN
LCAT	Fish-eye disease	ORPHA:79292	UNKNOWN
LCE3B	—		UNKNOWN
LCE3C	—		UNKNOWN
LCE5A	—		UNKNOWN
LCK	Immunodeficiency 22	OMIM:615758	MENDELIAN
LCN10	—		UNKNOWN
LCT	Lactase deficiency, congenital	OMIM:223000	MENDELIAN
LDB1	—		UNKNOWN
LDB3	Myopathy, myofibrillar, 4	OMIM:609452	MENDELIAN
LDB3	Cardiomyopathy, dilated, 2L	OMIM:621237	MENDELIAN
LDB3	Cardiomyopathy, dilated, 1C, with or without left ventricular noncompaction	OMIM:601493	MENDELIAN
LDB3	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
LDB3	Late-onset distal myopathy, Markesbery-Griggs type	ORPHA:98912	UNKNOWN
LDHA	Glycogen storage disease XI	OMIM:612933	MENDELIAN
LDHA	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency	ORPHA:284426	UNKNOWN
LDHB	Lactate dehydrogenase B deficiency	OMIM:614128	MENDELIAN
LDLR	Hypercholesterolemia, familial, 1	OMIM:143890	MENDELIAN
LDLR	Homozygous familial hypercholesterolemia	ORPHA:391665	UNKNOWN
LDLRAD4	—		UNKNOWN
LDLRAP1	Hypercholesterolemia, autosomal recessive	OMIM:603813	MENDELIAN
LDLRAP1	Homozygous familial hypercholesterolemia	ORPHA:391665	UNKNOWN
LECT2	—		UNKNOWN
LEF1	Ectodermal dysplasia 17 with or without limb malformations	OMIM:621224	MENDELIAN
LEFTY2	—		UNKNOWN
LEMD3	Buschke-Ollendorff syndrome	OMIM:166700	MENDELIAN
LEMD3	Isolated osteopoikilosis	ORPHA:166119	UNKNOWN
LEMD3	Melorheostosis with osteopoikilosis	ORPHA:1879	UNKNOWN
LEMD3	12q14 microdeletion syndrome	ORPHA:94063	UNKNOWN
LEP	Leptin deficiency or dysfunction	OMIM:614962	MENDELIAN
LEP	Obesity due to congenital leptin deficiency	ORPHA:66628	UNKNOWN
LEPR	Leptin receptor deficiency	OMIM:614963	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
LEPR	Obesity due to leptin receptor gene deficiency	ORPHA:179494	UNKNOWN
LEPRE1	—		UNKNOWN
LEPREL1	—		UNKNOWN
LETM1	Wolf-Hirschhorn syndrome	OMIM:194190	MENDELIAN
LETM1	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction	OMIM:620089	MENDELIAN
LETM1	Wolf-Hirschhorn syndrome	ORPHA:280	UNKNOWN
LFNG	Spondylocostal dysostosis 3, autosomal recessive	OMIM:609813	MENDELIAN
LFNG	Autosomal recessive spondylocostal dysostosis	ORPHA:2311	UNKNOWN
LGALS13	—		UNKNOWN
LGALS3	—		UNKNOWN
LGI1	Developmental and epileptic encephalopathy 121	OMIM:621475	MENDELIAN
LGI1	Epilepsy, familial temporal lobe, 1	OMIM:600512	MENDELIAN
LGI1	Epilepsy with auditory features	ORPHA:101046	UNKNOWN
LGI1	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
LGI4	Arthrogryposis multiplex congenita, neurogenic, with myelin defect	OMIM:617468	MENDELIAN
LGI4	Hypomyelination neuropathy-arthrogryposis syndrome	ORPHA:2680	UNKNOWN
LGR4	Delayed puberty, self-limited	OMIM:619613	MENDELIAN
LGR5	—		UNKNOWN
LHB	Hypogonadotropic hypogonadism 23 without anosmia	OMIM:228300	MENDELIAN
LHCGR	Precocious puberty, male	OMIM:176410	MENDELIAN
LHCGR	Hypergonadotropic hypogonadism	OMIM:238320	MENDELIAN
LHCGR	Familial peripheral male-limited precocious puberty	ORPHA:3000	UNKNOWN
LHFPL5	Deafness, autosomal recessive 67	OMIM:610265	MENDELIAN
LHR	—		UNKNOWN
LHX1	17q12 microdeletion syndrome	ORPHA:261265	UNKNOWN
LHX3	Deafness, sensorineural, with pituitary dwarfism	OMIM:221750	MENDELIAN
LHX3	Non-acquired combined pituitary hormone deficiency-sensorineural hearing loss-spine abnormalities syndrome	ORPHA:231720	UNKNOWN
LHX3	Hypothyroidism due to deficient transcription factors involved in pituitary development or function	ORPHA:226307	UNKNOWN
LHX4	Pituitary hormone deficiency, combined, 4	OMIM:262700	MENDELIAN
LHX4	Pituitary stalk interruption syndrome	ORPHA:95496	UNKNOWN
LHX4	Combined pituitary hormone deficiencies, genetic forms	ORPHA:95494	UNKNOWN
LHX4	Hypothyroidism due to deficient transcription factors involved in pituitary development or function	ORPHA:226307	UNKNOWN
LHX8	—		UNKNOWN
LIAS	Hyperglycinemia, lactic acidosis, and seizures	OMIM:614462	MENDELIAN
LIF	—		UNKNOWN
LIFR	Stuve-Wiedemann syndrome	OMIM:601559	MENDELIAN
LIFR	StÃ¼ve-Wiedemann syndrome	ORPHA:3206	UNKNOWN
LIG1	Immunodeficiency 96	OMIM:619774	MENDELIAN
LIG3	Mitochondrial DNA depletion syndrome 20 (MNGIE type)	OMIM:619780	MENDELIAN
LIG3	Mitochondrial neurogastrointestinal encephalomyopathy	ORPHA:298	UNKNOWN
LIG4	Multiple myeloma	OMIM:254500	POLYGENIC
LIG4	LIG4 syndrome	OMIM:606593	MENDELIAN
LIG4	LIG4 syndrome	ORPHA:99812	UNKNOWN
LILRA3	—		UNKNOWN
LIM2	Cataract 19, multiple types	OMIM:615277	MENDELIAN
LIMK1	Williams syndrome	ORPHA:904	UNKNOWN
LIMS2	Muscular dystrophy, limb-girdle, type 2W	OMIM:616827	MENDELIAN
LIN28A	—		UNKNOWN
LIN28B	Neuroblastoma	ORPHA:635	UNKNOWN
LIN7A	—		UNKNOWN
LINC00271	—		UNKNOWN
LINC00299	—		UNKNOWN
LINC00470	—		UNKNOWN
LINC00523	—		UNKNOWN
LINGO2	—		UNKNOWN
LINS	—		UNKNOWN
LIPA	Lysosomal acid lipase deficiency	OMIM:278000	MENDELIAN
LIPA	Wolman disease	OMIM:620151	MENDELIAN
LIPA	Wolman disease	ORPHA:75233	UNKNOWN
LIPA	Cholesteryl ester storage disease	ORPHA:75234	UNKNOWN
LIPC	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
LIPC	Hepatic lipase deficiency	OMIM:614025	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
LIPC	Hyperlipidemia due to hepatic triacylglycerol lipase deficiency	ORPHA:140905	UNKNOWN
LIPE	Lipodystrophy, familial partial, type 6	OMIM:615980	MENDELIAN
LIPE	LIPE-related familial partial lipodystrophy	ORPHA:435660	UNKNOWN
LIPG	—		UNKNOWN
LIPH	Hypotrichosis 7	OMIM:604379	MENDELIAN
LIPH	Woolly hair	ORPHA:170	UNKNOWN
LIPH	Hypotrichosis simplex	ORPHA:55654	UNKNOWN
LIPI	—		UNKNOWN
LIPN	Ichthyosis, congenital, autosomal recessive 8	OMIM:613943	MENDELIAN
LIPN	Lamellar ichthyosis	ORPHA:313	UNKNOWN
LIPT1	Lipoyltransferase 1 deficiency	OMIM:616299	MENDELIAN
LITAF	Charcot-Marie-Tooth disease, demyelinating, type 1C	OMIM:601098	MENDELIAN
LLGL1	—		UNKNOWN
LMAN1	Combined deficiency of factor V and factor VIII type 1	OMIM:227300	MENDELIAN
LMAN1	Combined deficiency of factor V and factor VIII	ORPHA:35909	UNKNOWN
LMAN2L	Intellectual developmental disorder, autosomal dominant 69	OMIM:617863	MENDELIAN
LMAN2L	Intellectual developmental disorder, autosomal recessive 52	OMIM:616887	MENDELIAN
LMBR1	Laurin-Sandrow syndrome	OMIM:135750	MENDELIAN
LMBR1	Triphalangeal thumb-polysyndactyly syndrome	OMIM:190605	MENDELIAN
LMBR1	Acheiropody	OMIM:200500	MENDELIAN
LMBR1	Syndactyly, type IV	OMIM:186200	MENDELIAN
LMBR1	Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome	ORPHA:988	UNKNOWN
LMBR1	Isolated acheiropodia	ORPHA:931	UNKNOWN
LMBR1	Laurin-Sandrow syndrome	ORPHA:2378	UNKNOWN
LMBR1	Isolated radial hemimelia	ORPHA:93321	UNKNOWN
LMBR1	Syndactyly type 4	ORPHA:93405	UNKNOWN
LMBRD1	Methylmalonic aciduria and homocystinuria, Cblf type	OMIM:277380	MENDELIAN
LMBRD1	Methylmalonic acidemia with homocystinuria type cblF	ORPHA:79284	UNKNOWN
LMF1	Lipase deficiency, combined	OMIM:246650	MENDELIAN
LMNA	Emery-Dreifuss muscular dystrophy 3, autosomal recessive	OMIM:616516	MENDELIAN
LMNA	Heart-hand syndrome, Slovenian type	OMIM:610140	MENDELIAN
LMNA	Malouf syndrome	OMIM:212112	MENDELIAN
LMNA	Charcot-Marie-Tooth disease, axonal, type 2B1	OMIM:605588	MENDELIAN
LMNA	Mandibuloacral dysplasia	OMIM:248370	MENDELIAN
LMNA	Cardiomyopathy, dilated, 1A	OMIM:115200	MENDELIAN
LMNA	Hutchinson-Gilford progeria syndrome	OMIM:176670	MENDELIAN
LMNA	LMNA-related congenital muscular dystrophy	OMIM:613205	MENDELIAN
LMNA	Lipodystrophy, familial partial, type 2	OMIM:151660	MENDELIAN
LMNA	Emery-Dreifuss muscular dystrophy 2, autosomal dominant	OMIM:181350	MENDELIAN
LMNA	Restrictive dermopathy 2	OMIM:619793	MENDELIAN
LMNA	Atypical Werner syndrome	ORPHA:79474	UNKNOWN
LMNA	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
LMNA	Autosomal semi-dominant severe lipodystrophic laminopathy	ORPHA:280365	UNKNOWN
LMNA	Heart-hand syndrome, Slovenian type	ORPHA:168796	UNKNOWN
LMNA	Autosomal recessive Emery-Dreifuss muscular dystrophy	ORPHA:98855	UNKNOWN
LMNA	Charcot-Marie-Tooth disease type 2B1	ORPHA:98856	UNKNOWN
LMNA	Autosomal dominant Emery-Dreifuss muscular dystrophy	ORPHA:98853	UNKNOWN
LMNA	Congenital muscular dystrophy due to LMNA mutation	ORPHA:157973	UNKNOWN
LMNA	Hutchinson-Gilford progeria syndrome	ORPHA:740	UNKNOWN
LMNA	Familial partial lipodystrophy, Dunnigan type	ORPHA:2348	UNKNOWN
LMNA	Mandibuloacral dysplasia with type A lipodystrophy	ORPHA:90153	UNKNOWN
LMNA	Familial dilated cardiomyopathy with conduction defect due to LMNA mutation	ORPHA:300751	UNKNOWN
LMNA	Familial partial lipodystrophy, KÄbberling type	ORPHA:79084	UNKNOWN
LMNA	Restrictive dermopathy	ORPHA:1662	UNKNOWN
LMNA	LMNA-related cardiocutaneous progeria syndrome	ORPHA:363618	UNKNOWN
LMNA	Dilated cardiomyopathy-hypergonadotropic hypogonadism syndrome	ORPHA:2229	UNKNOWN
LMNB1	Microcephaly 26, primary, autosomal dominant	OMIM:619179	MENDELIAN
LMNB1	Adult-onset autosomal dominant leukodystrophy	ORPHA:99027	UNKNOWN
LMNB1	Autosomal dominant primary microcephaly	ORPHA:2514	UNKNOWN
LMNB2	Epilepsy, progressive myoclonic, 9	OMIM:616540	MENDELIAN
LMNB2	Microcephaly 27, primary, autosomal dominant	OMIM:619180	MENDELIAN
LMNB2	Lipodystrophy, partial, acquired, susceptibility to	OMIM:608709	POLYGENIC
LMNB2	Acquired partial lipodystrophy	ORPHA:79087	UNKNOWN
LMO1	Neuroblastoma	ORPHA:635	UNKNOWN
LMO2	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
LMO4	—		UNKNOWN
LMOD3	Nemaline myopathy 10	OMIM:616165	MENDELIAN
LMOD3	Typical nemaline myopathy	ORPHA:171436	UNKNOWN
LMOD3	Severe congenital nemaline myopathy	ORPHA:171430	UNKNOWN
LMTK3	—		UNKNOWN
LMX1B	Focal segmental glomerulosclerosis 10	OMIM:256020	MENDELIAN
LMX1B	Nail-Patella syndrome	OMIM:161200	MENDELIAN
LMX1B	9q33.3q34.11 microdeletion syndrome	ORPHA:495818	UNKNOWN
LMX1B	Nail-patella-like renal disease	ORPHA:2613	UNKNOWN
LMX1B	Nail-patella syndrome	ORPHA:2614	UNKNOWN
LNPK	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum	OMIM:618090	MENDELIAN
LNX2	—		UNKNOWN
loc344967	—		UNKNOWN
LONP1	CODAS syndrome	OMIM:600373	MENDELIAN
LONP1	CODAS syndrome	ORPHA:1458	UNKNOWN
LONP1	Congenital diaphragmatic hernia	ORPHA:2140	UNKNOWN
LONP1	Pyruvate dehydrogenase E1-alpha deficiency	ORPHA:79243	UNKNOWN
LOR	—		UNKNOWN
LOX	Aortic aneurysm, familial thoracic 10	OMIM:617168	MENDELIAN
LOX	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
LOXHD1	Deafness, autosomal recessive 77	OMIM:613079	MENDELIAN
LOXL1	Exfoliation syndrome	OMIM:177650	POLYGENIC
LOXL2	—		UNKNOWN
LPAR1	—		UNKNOWN
LPAR6	Hypotrichosis 8	OMIM:278150	MENDELIAN
LPAR6	Woolly hair	ORPHA:170	UNKNOWN
LPAR6	Hypotrichosis simplex	ORPHA:55654	UNKNOWN
LPIN1	Myoglobinuria, acute recurrent, autosomal recessive	OMIM:268200	MENDELIAN
LPIN1	Genetic recurrent myoglobinuria	ORPHA:99845	UNKNOWN
LPIN2	Majeed syndrome	OMIM:609628	MENDELIAN
LPIN2	Majeed syndrome	ORPHA:77297	UNKNOWN
LPIN3	—		UNKNOWN
LPL	Type I hyperlipoproteinemia	OMIM:238600	MENDELIAN
LPL	Hyperlipidemia, familial combined, 3	OMIM:144250	MENDELIAN
LPP	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
LRAT	Leber congenital amaurosis 14	OMIM:613341	MENDELIAN
LRAT	Retinitis pigmentosa	OMIM:268000	MENDELIAN
LRAT	Leber congenital amaurosis	ORPHA:65	UNKNOWN
LRAT	Retinitis pigmentosa	ORPHA:791	UNKNOWN
LRAT	Severe early-childhood-onset retinal dystrophy	ORPHA:364055	UNKNOWN
LRBA	Immunodeficiency, common variable, 8, with autoimmunity	OMIM:614700	MENDELIAN
LRCH1	—		UNKNOWN
LRFN5	—		UNKNOWN
LRGUK	—		UNKNOWN
LRIG2	Urofacial syndrome 2	OMIM:615112	MENDELIAN
LRIG2	Urofacial syndrome	ORPHA:2704	UNKNOWN
LRIT3	Night blindness, congenital stationary, type 1F	OMIM:615058	MENDELIAN
LRP1	Developmental dysplasia of the hip 3	OMIM:620690	MENDELIAN
LRP1	Keratosis pilaris atrophicans	OMIM:604093	MENDELIAN
LRP1	Keratosis follicularis spinulosa decalvans	ORPHA:2340	UNKNOWN
LRP1	Atrophoderma vermiculata	ORPHA:79100	UNKNOWN
LRP1B	—		UNKNOWN
LRP2	Donnai-Barrow syndrome	OMIM:222448	MENDELIAN
LRP2	Donnai-Barrow syndrome	ORPHA:2143	UNKNOWN
LRP4	Myasthenic syndrome, congenital, 17	OMIM:616304	MENDELIAN
LRP4	Sclerosteosis 2	OMIM:614305	MENDELIAN
LRP4	Cenani-Lenz syndactyly syndrome	OMIM:212780	MENDELIAN
LRP4	Sclerosteosis	ORPHA:3152	UNKNOWN
LRP4	Cenani-Lenz syndrome	ORPHA:3258	UNKNOWN
LRP5	Polycystic liver disease 4 with or without kidney cysts	OMIM:617875	MENDELIAN
LRP5	Osteopetrosis, autosomal dominant 1	OMIM:607634	MENDELIAN
LRP5	Exudative vitreoretinopathy 1	OMIM:133780	MENDELIAN
LRP5	Osteoporosis-pseudoglioma syndrome	OMIM:259770	MENDELIAN
LRP5	Exudative vitreoretinopathy 4	OMIM:601813	MENDELIAN
LRP5	Hyperostosis, endosteal	OMIM:144750	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
LRP5	Endosteal hyperostosis, Worth type	ORPHA:2790	UNKNOWN
LRP5	Osteoporosis-pseudoglioma syndrome	ORPHA:2788	UNKNOWN
LRP5	Osteosclerosis-developmental delay-craniosynostosis syndrome	ORPHA:178377	UNKNOWN
LRP5	Familial exudative vitreoretinopathy	ORPHA:891	UNKNOWN
LRP5	Retinopathy of prematurity	ORPHA:90050	UNKNOWN
LRP5	Isolated polycystic liver disease	ORPHA:2924	UNKNOWN
LRP5	Hyperostosis corticalis generalisata	ORPHA:3416	UNKNOWN
LRP6	Osteopetrosis, autosomal dominant 4	OMIM:621449	MENDELIAN
LRP6	Exudative vitreoretinopathy 8	OMIM:621268	MENDELIAN
LRP6	Tooth agenesis, selective, 7	OMIM:616724	MENDELIAN
LRP6	Coronary artery disease, autosomal dominant 2	OMIM:610947	POLYGENIC
LRP6	Autosomal dominant hypohidrotic ectodermal dysplasia	ORPHA:1810	UNKNOWN
LRP6	Oligodontia	ORPHA:99798	UNKNOWN
LRPAP1	Myopia 23, autosomal recessive	OMIM:615431	MENDELIAN
LRPPRC	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)	OMIM:220111	MENDELIAN
LRPPRC	Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type	ORPHA:70472	UNKNOWN
LRRC29	—		UNKNOWN
LRRC4	—		UNKNOWN
LRRC41	—		UNKNOWN
LRRC4C	—		UNKNOWN
LRRC6	—		UNKNOWN
LRRC69	—		UNKNOWN
LRRC8A	Agammaglobulinemia 5, autosomal dominant	OMIM:613506	MENDELIAN
LRRC8A	Autosomal non-syndromic agammaglobulinemia	ORPHA:33110	UNKNOWN
LRRFIP2	—		UNKNOWN
LRRIQ1	—		UNKNOWN
LRRK2	Parkinson disease 8, autosomal dominant	OMIM:607060	POLYGENIC
LRRK2	Young-onset Parkinson disease	ORPHA:2828	UNKNOWN
LRRK2	Hereditary late-onset Parkinson disease	ORPHA:411602	UNKNOWN
LRRTM1	—		UNKNOWN
LRSAM1	Charcot-Marie-Tooth disease, axonal, type 2P	OMIM:614436	MENDELIAN
LRTOMT	Deafness, autosomal recessive 63	OMIM:611451	MENDELIAN
LTBP1	Cutis laxa, autosomal recessive, type IIE	OMIM:619451	MENDELIAN
LTBP1	Autosomal recessive cutis laxa type 1	ORPHA:90349	UNKNOWN
LTBP2	Glaucoma 3, primary infantile, B	OMIM:600975	MENDELIAN
LTBP2	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma	OMIM:251750	MENDELIAN
LTBP2	Glaucoma 3, primary congenital, D	OMIM:613086	MENDELIAN
LTBP2	Weill-Marchesani syndrome 3	OMIM:614819	MENDELIAN
LTBP2	Congenital glaucoma	ORPHA:98976	UNKNOWN
LTBP2	Weill-Marchesani syndrome	ORPHA:3449	UNKNOWN
LTBP3	Geleophysic dysplasia 3	OMIM:617809	MENDELIAN
LTBP3	Dental anomalies and short stature	OMIM:601216	MENDELIAN
LTBP3	Acromicric dysplasia	ORPHA:969	UNKNOWN
LTBP3	Geleophysic dysplasia	ORPHA:2623	UNKNOWN
LTBP4	Cutis laxa, autosomal recessive, type IC	OMIM:613177	MENDELIAN
LTBP4	Duchenne muscular dystrophy	ORPHA:98896	UNKNOWN
LTBR	—		UNKNOWN
LTC4S	Leukotriene C4 synthase deficiency	OMIM:614037	MENDELIAN
LTF	—		UNKNOWN
LTK	—		UNKNOWN
LTN1	—		UNKNOWN
LUM	—		UNKNOWN
LUZP2	—		UNKNOWN
LUZP4	—		UNKNOWN
LY6G6F	—		UNKNOWN
LY96	—		UNKNOWN
LYL1	—		UNKNOWN
LYN	Autoinflammatory disease, systemic, with vasculitis	OMIM:620376	MENDELIAN
LYRM4	Combined oxidative phosphorylation deficiency 19	OMIM:615595	MENDELIAN
LYRM7	Mitochondrial complex III deficiency, nuclear type 8	OMIM:615838	MENDELIAN
LYST	Chediak-Higashi syndrome	OMIM:214500	MENDELIAN
LYST	ChÃ©diak-Higashi syndrome	ORPHA:167	UNKNOWN
LYST	Attenuated ChÃ©diak-Higashi syndrome	ORPHA:352723	UNKNOWN
LYZ	Amyloidosis, hereditary systemic 5	OMIM:620658	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
LZTFL1	Bardet-Biedl syndrome 17	OMIM:615994	MENDELIAN
LZTFL1	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
LZTR1	Noonan syndrome 10	OMIM:616564	MENDELIAN
LZTR1	Schwannomatosis 2	OMIM:615670	POLYGENIC
LZTR1	Noonan syndrome 2	OMIM:605275	MENDELIAN
LZTR1	Full schwannomatosis	ORPHA:93921	UNKNOWN
LZTR1	Familial isolated café-au-lait macules	ORPHA:2678	UNKNOWN
LZTR1	Noonan syndrome	ORPHA:648	UNKNOWN
LZTS1	Esophageal cancer, somatic	OMIM:133239	MENDELIAN
M1AP	Spermatogenic failure 48	OMIM:619108	MENDELIAN
MAB21L2	Microphthalmia/coloboma and skeletal dysplasia syndrome	OMIM:615877	MENDELIAN
MACC1	—		UNKNOWN
MACF1	Lissencephaly 9 with complex brainstem malformation	OMIM:618325	MENDELIAN
MACF1	Posterior-predominant lissencephaly-broad flat pons and medulla-midline crossing defects syndrome	ORPHA:572013	UNKNOWN
MACROD2	—		UNKNOWN
MAD1L1	Mosaic variegated aneuploidy syndrome 7 with inflammation and tumor predisposition	OMIM:620189	MENDELIAN
MAD1L1	Prostate cancer	OMIM:176807	MENDELIAN
MAD2L1	—		UNKNOWN
MADD	Deeah syndrome	OMIM:619004	MENDELIAN
MADD	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia	OMIM:619005	MENDELIAN
MAEA	—		UNKNOWN
MAEL	—		UNKNOWN
MAF	Cataract 21, multiple types	OMIM:610202	MENDELIAN
MAF	Ayme-Gripp syndrome	OMIM:601088	MENDELIAN
MAF	Aym-Gripp syndrome	ORPHA:1272	UNKNOWN
MAF	Cataract-microcornea syndrome	ORPHA:1377	UNKNOWN
MAFA	Insulinomatosis and diabetes mellitus	OMIM:147630	MENDELIAN
MAFB	Multicentric carpotarsal osteolysis syndrome	OMIM:166300	MENDELIAN
MAFB	Duane retraction syndrome 3 with or without deafness	OMIM:617041	MENDELIAN
MAFB	Multicentric carpo-tarsal osteolysis with or without nephropathy	ORPHA:2774	UNKNOWN
MAFB	Duane retraction syndrome	ORPHA:233	UNKNOWN
MAG	Spastic paraplegia 75, autosomal recessive	OMIM:616680	MENDELIAN
MAG	Autosomal recessive spastic paraplegia type 75	ORPHA:459056	UNKNOWN
MAGEB18	—		UNKNOWN
MAGEB4	—		UNKNOWN
MAGEC3	—		UNKNOWN
MAGEE2	—		UNKNOWN
MAGEL2	Prader-Willi syndrome	OMIM:176270	MENDELIAN
MAGEL2	Schaaf-Yang syndrome	OMIM:615547	MENDELIAN
MAGEL2	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2	ORPHA:177904	UNKNOWN
MAGEL2	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1	ORPHA:177901	UNKNOWN
MAGEL2	Prader-Willi syndrome due to imprinting mutation	ORPHA:177910	UNKNOWN
MAGEL2	Fetal akinesia deformation sequence	ORPHA:994	UNKNOWN
MAGEL2	Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15	ORPHA:98754	UNKNOWN
MAGEL2	Schaaf-Yang syndrome	ORPHA:398069	UNKNOWN
MAGI1	—		UNKNOWN
MAGI2	Nephrotic syndrome, type 15	OMIM:617609	MENDELIAN
MAGI2	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
MAGT1	Congenital disorder of glycosylation, type Icc	OMIM:301031	MENDELIAN
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia	OMIM:300853	MENDELIAN
MAJIN	—		UNKNOWN
MAK	Retinitis pigmentosa 62	OMIM:614181	MENDELIAN
MAK	Retinitis pigmentosa	ORPHA:791	UNKNOWN
MALT1	Immunodeficiency 12	OMIM:615468	MENDELIAN
MALT1	MALT lymphoma	ORPHA:52417	UNKNOWN
MAML2	—		UNKNOWN
MAMLD1	Hypospadias 2, X-linked	OMIM:300758	MENDELIAN
MAMLD1	X-linked myotubular myopathy-abnormal genitalia syndrome	ORPHA:456328	UNKNOWN
MAMLD1	Non-syndromic posterior hypospadias	ORPHA:95706	UNKNOWN
MAN1A2	—		UNKNOWN
MAN1B1	Rafiq syndrome	OMIM:614202	MENDELIAN
MAN1B1	MAN1B1-CDG	ORPHA:397941	UNKNOWN
MAN2A1	—		UNKNOWN
MAN2B1	Alpha-mannosidosis	OMIM:248500	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
MAN2B1	Alpha-mannosidosis, infantile form	ORPHA:309282	UNKNOWN
MAN2B1	Alpha-mannosidosis, adult form	ORPHA:309288	UNKNOWN
MANBA	Mannosidosis, beta	OMIM:248510	MENDELIAN
MANBA	Beta-mannosidosis	ORPHA:118	UNKNOWN
MANBAL	—		UNKNOWN
MANSC1	—		UNKNOWN
MAOA	Brunner syndrome	OMIM:300615	MENDELIAN
MAOA	Monoamine oxidase A deficiency	ORPHA:3057	UNKNOWN
MAP1A	—		UNKNOWN
MAP2	—		UNKNOWN
MAP2K1	Cardiofaciocutaneous syndrome 3	OMIM:615279	MENDELIAN
MAP2K1	Cardiofaciocutaneous syndrome 1	OMIM:115150	MENDELIAN
MAP2K1	Melorheostosis, isolated	OMIM:155950	MENDELIAN
MAP2K1	Noonan syndrome 1	OMIM:163950	MENDELIAN
MAP2K1	Langerhans cell histiocytosis	ORPHA:389	UNKNOWN
MAP2K1	Cardiofaciocutaneous syndrome	ORPHA:1340	UNKNOWN
MAP2K2	Cardiofaciocutaneous syndrome 4	OMIM:615280	MENDELIAN
MAP2K2	Cardiofaciocutaneous syndrome 1	OMIM:115150	MENDELIAN
MAP2K2	Cardiofaciocutaneous syndrome	ORPHA:1340	UNKNOWN
MAP2K2	Neurofibromatosis-Noonan syndrome	ORPHA:638	UNKNOWN
MAP2K3	—		UNKNOWN
MAP2K4	—		UNKNOWN
MAP2K7	—		UNKNOWN
MAP3K1	46,xy sex reversal 6	OMIM:613762	MENDELIAN
MAP3K1	46,XY partial gonadal dysgenesis	ORPHA:251510	UNKNOWN
MAP3K1	46,XY complete gonadal dysgenesis	ORPHA:242	UNKNOWN
MAP3K13	—		UNKNOWN
MAP3K14	Immunodeficiency 112	OMIM:620449	MENDELIAN
MAP3K15	—		UNKNOWN
MAP3K4	—		UNKNOWN
MAP3K8	Lung cancer, susceptibility to	OMIM:211980	MENDELIAN
MAP4	—		UNKNOWN
MAP4K5	—		UNKNOWN
MAP6	—		UNKNOWN
MAP7D1	—		UNKNOWN
MAP7D3	—		UNKNOWN
MAPK1	Noonan syndrome 13	OMIM:619087	MENDELIAN
MAPK1	Distal 22q11.2 microdeletion syndrome	ORPHA:261330	UNKNOWN
MAPK10	Lennox-Gastaut syndrome	ORPHA:2382	UNKNOWN
MAPK14	—		UNKNOWN
MAPK3	—		UNKNOWN
MAPK8IP1	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
MAPRE2	Skin creases, congenital symmetric circumferential, 2	OMIM:616734	MENDELIAN
MAPRE2	Multiple benign circumferential skin creases on limbs	ORPHA:2505	UNKNOWN
MAPT	Pick disease of brain	OMIM:172700	MENDELIAN
MAPT	Supranuclear palsy, progressive atypical	OMIM:260540	MENDELIAN
MAPT	Supranuclear palsy, progressive, 1	OMIM:601104	MENDELIAN
MAPT	Parkinson disease, late-onset	OMIM:168600	POLYGENIC
MAPT	Frontotemporal dementia	OMIM:600274	MENDELIAN
MAPT	Progressive supranuclear palsy-pure akinesia with gait freezing syndrome	ORPHA:240094	UNKNOWN
MAPT	Progressive supranuclear palsy-predominant parkinsonism syndrome	ORPHA:240085	UNKNOWN
MAPT	Classic progressive supranuclear palsy syndrome	ORPHA:240071	UNKNOWN
MAPT	Behavioral variant of frontotemporal dementia	ORPHA:275864	UNKNOWN
MAPT	Progressive non-fluent aphasia	ORPHA:100070	UNKNOWN
MAPT	Semantic dementia	ORPHA:100069	UNKNOWN
MAPT	Progressive supranuclear palsy-progressive non-fluent aphasia syndrome	ORPHA:240112	UNKNOWN
MAPT	Progressive supranuclear palsy-corticobasal syndrome	ORPHA:240103	UNKNOWN
MARCH3	—		UNKNOWN
MARK1	—		UNKNOWN
MARS	—		UNKNOWN
MARS2	Combined oxidative phosphorylation deficiency 25	OMIM:616430	MENDELIAN
MARS2	Spastic ataxia 3, autosomal recessive	OMIM:611390	MENDELIAN
MARS2	Autosomal recessive spastic ataxia with leukoencephalopathy	ORPHA:314603	UNKNOWN
MARVELD2	Deafness, autosomal recessive 49	OMIM:610153	MENDELIAN
MASP1	3MC syndrome 1	OMIM:257920	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
MASP1	3MC syndrome	ORPHA:293843	UNKNOWN
MASP2	Masp2 deficiency	OMIM:613791	MENDELIAN
MAST4	—		UNKNOWN
MAT1A	Methionine adenosyltransferase I/III deficiency	OMIM:250850	MENDELIAN
MAT2A	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
MATN3	Epiphyseal dysplasia, multiple, 5	OMIM:607078	MENDELIAN
MATN3	Osteoarthritis of distal interphalangeal joints	OMIM:140600	POLYGENIC
MATN3	Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type	OMIM:608728	MENDELIAN
MATN3	Spondyloepimetaphyseal dysplasia, matrilin-3 type	ORPHA:156728	UNKNOWN
MATN3	Multiple epiphyseal dysplasia type 5	ORPHA:93311	UNKNOWN
MATN4	—		UNKNOWN
MATR3	Amyotrophic lateral sclerosis 21	OMIM:606070	MENDELIAN
MATR3	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
MATR3	Vocal cord and pharyngeal distal myopathy	ORPHA:600	UNKNOWN
MAVS	—		UNKNOWN
MAX	Pheochromocytoma	OMIM:171300	POLYGENIC
MAX	Polydactyly-macrocephaly syndrome	OMIM:620712	MENDELIAN
MAX	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
MBD1	—		UNKNOWN
MBD3	—		UNKNOWN
MBD4	Tumor predisposition syndrome 2	OMIM:619975	MENDELIAN
MBD4	Uveal melanoma, susceptibility to, 1	OMIM:606660	POLYGENIC
MBD5	Intellectual developmental disorder, autosomal dominant 1	OMIM:156200	MENDELIAN
MBD5	2q23.1 microdeletion syndrome	ORPHA:228402	UNKNOWN
MBD6	—		UNKNOWN
MBL2	Mannose-Binding lectin deficiency	OMIM:614372	POLYGENIC
MBNL1	—		UNKNOWN
MBOAT7	Intellectual developmental disorder, autosomal recessive 57	OMIM:617188	MENDELIAN
MBTPS2	IFAP syndrome with or without BRESHECK syndrome	OMIM:308205	MENDELIAN
MBTPS2	Palmoplantar keratoderma, mutilating, with periorificial keratotic plaques, X-linked	OMIM:300918	MENDELIAN
MBTPS2	Keratosis follicularis spinulosa decalvans, X-linked	OMIM:308800	MENDELIAN
MBTPS2	Osteogenesis imperfecta, type XIX	OMIM:301014	MENDELIAN
MBTPS2	BRESEK syndrome	ORPHA:85284	UNKNOWN
MBTPS2	Keratosis follicularis spinulosa decalvans	ORPHA:2340	UNKNOWN
MBTPS2	Mutilating palmoplantar keratoderma with periorificial keratotic plaques	ORPHA:659	UNKNOWN
MBTPS2	Ichthyosis follicularis-alopecia-photophobia syndrome	ORPHA:2273	UNKNOWN
MC1R	SKIN/HAIR/EYE PIGMENTATION, VARIATION IN, 2	OMIM:266300	POLYGENIC
MC1R	Albinism, oculocutaneous, type II	OMIM:203200	POLYGENIC
MC1R	Oculocutaneous albinism type 2	ORPHA:79432	UNKNOWN
MC1R	Familial melanoma	ORPHA:618	UNKNOWN
MC2R	Glucocorticoid deficiency 1	OMIM:202200	MENDELIAN
MC2R	Familial glucocorticoid deficiency	ORPHA:361	UNKNOWN
MC3R	Obesity, severe, susceptibility to, BMIQ9	OMIM:602025	POLYGENIC
MC4R	Body mass index quantitative trait locus 20	OMIM:618406	MENDELIAN
MC4R	Obesity due to melanocortin 4 receptor deficiency	ORPHA:71529	UNKNOWN
MCC	Colorectal cancer	OMIM:114500	MENDELIAN
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	OMIM:210200	MENDELIAN
MCCC1	3-methylcrotonyl-CoA carboxylase deficiency	ORPHA:6	UNKNOWN
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	OMIM:210210	MENDELIAN
MCCC2	3-methylcrotonyl-CoA carboxylase deficiency	ORPHA:6	UNKNOWN
MCEE	Methylmalonyl-CoA epimerase deficiency	OMIM:251120	MENDELIAN
MCF2	—		UNKNOWN
MCF2L2	—		UNKNOWN
MCFD2	Combined deficiency of factor V and factor VIII type 2	OMIM:613625	MENDELIAN
MCFD2	Combined deficiency of factor V and factor VIII	ORPHA:35909	UNKNOWN
MCHR1	—		UNKNOWN
MCL1	—		UNKNOWN
MCM3AP	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development	OMIM:618124	MENDELIAN
MCM4	Immunodeficiency 54	OMIM:609981	MENDELIAN
MCM5	Meier-Gorlin syndrome 8	OMIM:617564	MENDELIAN
MCM6	Lactase persistence/nonpersistence	OMIM:223100	MENDELIAN
MCM8	Premature ovarian failure 10	OMIM:612885	MENDELIAN
MCM9	Ovarian dysgenesis 4	OMIM:616185	MENDELIAN
MCOLN1	Lisch epithelial corneal dystrophy	OMIM:620763	MENDELIAN
MCOLN1	Mucopolipidosis IV	OMIM:252650	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
MCOLN1	Mucopolipidosis type IV	ORPHA:578	UNKNOWN
MCPH1	Microcephaly, primary autosomal recessive, 1	OMIM:251200	MENDELIAN
MCPH1	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
MCTP2	Distal deletion 15q syndrome	ORPHA:1596	UNKNOWN
MDC1	—		UNKNOWN
MDGA2	Developmental and epileptic encephalopathy 122	OMIM:621608	MENDELIAN
MDGA2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
MDH1	Epileptic encephalopathy, early infantile, 88	OMIM:618959	MENDELIAN
MDH2	Epileptic encephalopathy, early infantile, 51	OMIM:617339	MENDELIAN
MDH2	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
MDK	—		UNKNOWN
MDM2	Lessel-Kubisch syndrome	OMIM:618681	MENDELIAN
MDM2	Well-differentiated liposarcoma	ORPHA:99971	UNKNOWN
MDM2	Li-Fraumeni syndrome	ORPHA:524	UNKNOWN
MDM4	Bone marrow failure syndrome 6	OMIM:618849	MENDELIAN
MDN1	—		UNKNOWN
ME2	—		UNKNOWN
MECOM	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2	OMIM:616738	MENDELIAN
MECOM	Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome	ORPHA:71289	UNKNOWN
MECP2	Intellectual developmental disorder, X-linked syndromic 13	OMIM:300055	MENDELIAN
MECP2	Autism susceptibility, X-linked 3	OMIM:300496	POLYGENIC
MECP2	Intellectual developmental disorder, X-linked syndromic, Lubs type	OMIM:300260	MENDELIAN
MECP2	Rett syndrome	OMIM:312750	MENDELIAN
MECP2	Encephalopathy, neonatal severe, due to mecp2 mutations	OMIM:300673	MENDELIAN
MECP2	Atypical Rett syndrome	ORPHA:3095	UNKNOWN
MECP2	X-linked intellectual disability-psychosis-macroorchidism syndrome	ORPHA:3077	UNKNOWN
MECP2	Rett syndrome	ORPHA:778	UNKNOWN
MECP2	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
MECP2	MECP2-related severe neonatal encephalopathy	ORPHA:209370	UNKNOWN
MECP2	Proximal Xq28 duplication syndrome	ORPHA:1762	UNKNOWN
MED12	Lujan-Fryns syndrome	OMIM:309520	MENDELIAN
MED12	Hardikar syndrome	OMIM:301068	MENDELIAN
MED12	Ohdo syndrome, X-linked	OMIM:300895	MENDELIAN
MED12	Opitz-Kaveggia syndrome	OMIM:305450	MENDELIAN
MED12	Hardikar syndrome	ORPHA:1415	UNKNOWN
MED12	FG syndrome type 1	ORPHA:93932	UNKNOWN
MED12	Blepharophimosis-intellectual disability syndrome, MKB type	ORPHA:293707	UNKNOWN
MED12	Lujan-Fryns syndrome	ORPHA:776	UNKNOWN
MED12L	Nizon-Isidor syndrome	OMIM:618872	MENDELIAN
MED13	Intellectual developmental disorder 61	OMIM:618009	MENDELIAN
MED13L	Impaired intellectual development and distinctive facial features with or without cardiac defects	OMIM:616789	MENDELIAN
MED13L	Developmental delay-facial dysmorphism syndrome due to MED13L deficiency	ORPHA:369891	UNKNOWN
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy	OMIM:613668	MENDELIAN
MED20	—		UNKNOWN
MED23	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy	OMIM:614249	MENDELIAN
MED25	Basel-Vanagaite-Smirin-Yosef syndrome	OMIM:616449	MENDELIAN
MED25	Charcot-Marie-Tooth disease, axonal, type 2B2	OMIM:605589	MENDELIAN
MED25	Basel-Vanagaite-Smirin-Yosef syndrome	ORPHA:464738	UNKNOWN
MEF2A	Coronary artery disease, autosomal dominant, 1	OMIM:608320	POLYGENIC
MEF2B	—		UNKNOWN
MEF2C	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language	OMIM:613443	MENDELIAN
MEF2C	5q14.3 microdeletion syndrome	ORPHA:228384	UNKNOWN
MEFV	Neutrophilic dermatosis, acute febrile	OMIM:608068	MENDELIAN
MEFV	Familial Mediterranean fever, AD	OMIM:134610	MENDELIAN
MEFV	Familial Mediterranean fever, AR	OMIM:249100	MENDELIAN
MEFV	Familial Mediterranean fever	ORPHA:342	UNKNOWN
MEFV	Sweet syndrome	ORPHA:3243	UNKNOWN
MEFV	Intermittent hydrarthrosis	ORPHA:329967	UNKNOWN
MEFV	Behçet disease	ORPHA:117	UNKNOWN
MEG3	Temple syndrome due to paternal 14q32.2 hypomethylation	ORPHA:254531	UNKNOWN
MEG3	Kagami-Ogata syndrome due to maternal 14q32.2 hypermethylation	ORPHA:254534	UNKNOWN
MEG3	Temple syndrome due to paternal 14q32.2 microdeletion	ORPHA:254525	UNKNOWN
MEG3	Kagami-Ogata syndrome due to maternal 14q32.2 microdeletion	ORPHA:254528	UNKNOWN
MEG3	Temple syndrome due to maternal uniparental disomy of chromosome 14	ORPHA:96184	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
MEG3	Kagami-Ogata syndrome due to paternal uniparental disomy of chromosome 14	ORPHA:96334	UNKNOWN
MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	OMIM:614399	MENDELIAN
MEGF10	Congenital myopathy 10B, mild variant	OMIM:620249	MENDELIAN
MEGF11	—		UNKNOWN
MEGF8	Carpenter syndrome 2	OMIM:614976	MENDELIAN
MEGF8	Carpenter syndrome	ORPHA:65759	UNKNOWN
MEI1	Hydatidiform mole, recurrent, 3	OMIM:618431	MENDELIAN
MEIOB	Spermatogenic failure 22	OMIM:617706	MENDELIAN
MEIOB	Premature ovarian failure 23	OMIM:620686	MENDELIAN
MEIOB	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
MEIS1	—		UNKNOWN
MEIS2	Cleft palate, cardiac defects, and impaired intellectual development	OMIM:600987	MENDELIAN
MEIS2	Cleft palate-congenital heart defect-intellectual disability syndrome due to 15q14 microdeletion	ORPHA:261190	UNKNOWN
MELK	—		UNKNOWN
MEN1	Hyperparathyroidism 1	OMIM:145000	MENDELIAN
MEN1	Multiple endocrine neoplasia 1	OMIM:131100	MENDELIAN
MEN1	Familial isolated hyperparathyroidism	ORPHA:99879	UNKNOWN
MEN1	Insulinoma	ORPHA:97279	UNKNOWN
MEN1	Multiple endocrine neoplasia type 1	ORPHA:652	UNKNOWN
MEN1	Prolactinoma	ORPHA:2965	UNKNOWN
MEN1	Pituitary gigantism	ORPHA:99725	UNKNOWN
MEOX1	Klippel-Feil syndrome, autosomal recessive	OMIM:214300	MENDELIAN
MEOX1	Isolated Klippel-Feil syndrome	ORPHA:2345	UNKNOWN
MEP1B	—		UNKNOWN
MEPE	—		UNKNOWN
MERTK	Retinitis pigmentosa 38	OMIM:613862	MENDELIAN
MERTK	Retinitis pigmentosa	ORPHA:791	UNKNOWN
MESDC2	—		UNKNOWN
MESP1	—		UNKNOWN
MESP2	Spondylocostal dysostosis, autosomal recessive 1	OMIM:277300	MENDELIAN
MESP2	Spondylocostal dysostosis 2, autosomal recessive	OMIM:608681	MENDELIAN
MESP2	Autosomal recessive spondylocostal dysostosis	ORPHA:2311	UNKNOWN
MEST	—		UNKNOWN
MET	Osteofibrous dysplasia, susceptibility to	OMIM:607278	POLYGENIC
MET	Arthrogryposis, distal, type 11	OMIM:620019	MENDELIAN
MET	Deafness, autosomal recessive 97	OMIM:616705	MENDELIAN
MET	Renal cell carcinoma, papillary, 1, familial and somatic	OMIM:605074	MENDELIAN
MET	Hepatocellular carcinoma	OMIM:114550	MENDELIAN
MET	Pediatric hepatocellular carcinoma	ORPHA:33402	UNKNOWN
METTL14	—		UNKNOWN
METTL23	Intellectual developmental disorder, autosomal recessive 44	OMIM:615942	MENDELIAN
MFAP4	—		UNKNOWN
MFAP5	Aortic aneurysm, familial thoracic 9	OMIM:616166	MENDELIAN
MFAP5	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
MFF	Encephalopathy due to defective mitochondrial and peroxisomal fission 2	OMIM:617086	MENDELIAN
MFF	MFF-related encephalopathy due to mitochondrial and peroxisomal fission defect	ORPHA:485421	UNKNOWN
MFGE8	—		UNKNOWN
MFHAS1	—		UNKNOWN
MF12	—		UNKNOWN
MFN2	Charcot-marie-tooth disease, axonal, type 2A2A	OMIM:609260	MENDELIAN
MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2B	OMIM:617087	MENDELIAN
MFN2	Hereditary motor and sensory neuropathy VIA	OMIM:601152	MENDELIAN
MFN2	Lipomatosis, multiple symmetric, with or without peripheral neuropathy	OMIM:151800	MENDELIAN
MFN2	Autosomal dominant Charcot-Marie-Tooth disease type 2A2	ORPHA:99947	UNKNOWN
MFN2	Multiple symmetric lipomatosis	ORPHA:2398	UNKNOWN
MFRP	Nanophthalmos 2	OMIM:609549	MENDELIAN
MFRP	Microphthalmia, isolated 5	OMIM:611040	MENDELIAN
MFRP	Isolated complete colobomatous microphthalmia	ORPHA:35612	UNKNOWN
MFSD2A	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities	OMIM:616486	MENDELIAN
MFSD2A	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
MFSD8	Ceroid lipofuscinosis, neuronal, 7	OMIM:610951	MENDELIAN
MFSD8	Macular dystrophy with central cone involvement	OMIM:616170	MENDELIAN
MFSD8	Cone rod dystrophy	ORPHA:1872	UNKNOWN
MGA	Premature ovarian failure 26	OMIM:621065	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
MGAT1	—		UNKNOWN
MGAT2	Congenital disorder of glycosylation, type IIa	OMIM:212066	MENDELIAN
MGAT2	MGAT2-CDG	ORPHA:79329	UNKNOWN
MGAT3	—		UNKNOWN
MGAT4A	—		UNKNOWN
MGAT4B	—		UNKNOWN
MGAT4C	—		UNKNOWN
MGAT5	—		UNKNOWN
MGAT5B	—		UNKNOWN
MGEA5	—		UNKNOWN
MGLL	—		UNKNOWN
MGME1	Mitochondrial DNA depletion syndrome 11	OMIM:615084	MENDELIAN
MGME1	Progressive external ophthalmoplegia-myopathy-emaciation syndrome	ORPHA:352447	UNKNOWN
MGMT	Familial melanoma	ORPHA:618	UNKNOWN
MGP	Keutel syndrome	OMIM:245150	MENDELIAN
MGP	Keutel syndrome	ORPHA:85202	UNKNOWN
MGST2	—		UNKNOWN
MGST3	—		UNKNOWN
MIA3	Ondotochondrodysplasia 2 with hearing loss and diabetes	OMIM:619269	MENDELIAN
MIB1	Left ventricular noncompaction 7	OMIM:615092	MENDELIAN
MICA	—		UNKNOWN
MICAL3	—		UNKNOWN
MICALCL	—		UNKNOWN
MICB	—		UNKNOWN
MICU1	Myopathy with extrapyramidal signs	OMIM:615673	MENDELIAN
MICU1	Proximal myopathy with extrapyramidal signs	ORPHA:401768	UNKNOWN
MID1	Opitz GBBB syndrome	OMIM:300000	MENDELIAN
MID1	Opitz GBBB syndrome	ORPHA:2745	UNKNOWN
MID2	Intellectual developmental disorder, X-linked 101	OMIM:300928	MENDELIAN
MIF	Systemic-onset juvenile idiopathic arthritis	ORPHA:85414	UNKNOWN
MIF	Cystic fibrosis	ORPHA:586	UNKNOWN
MIIP	—		UNKNOWN
MINPP1	Thyroid cancer, nonmedullary, 2	OMIM:188470	POLYGENIC
MINPP1	Pontocerebellar hypoplasia, type 16	OMIM:619527	MENDELIAN
MINPP1	Familial papillary or follicular thyroid carcinoma	ORPHA:319487	UNKNOWN
MIP	Cataract 15, multiple types	OMIM:615274	MENDELIAN
MIPOL1	—		UNKNOWN
MIR106B	—		UNKNOWN
MIR124-1	—		UNKNOWN
MIR125A	—		UNKNOWN
MIR126	—		UNKNOWN
MIR140	Spondyloepiphyseal dysplasia, Nishimura type	OMIM:618618	MENDELIAN
MIR146A	—		UNKNOWN
MIR16-1	—		UNKNOWN
MIR17	—		UNKNOWN
MIR17HG	Feingold syndrome type 2	ORPHA:391646	UNKNOWN
MIR182	—		UNKNOWN
MIR183	—		UNKNOWN
MIR184	Edict syndrome	OMIM:614303	MENDELIAN
MIR191	—		UNKNOWN
MIR196A2	—		UNKNOWN
MIR206	—		UNKNOWN
MIR24-1	—		UNKNOWN
MIR27A	—		UNKNOWN
MIR2861	Bone mineral density QTL 15	OMIM:613418	MENDELIAN
MIR30C1	—		UNKNOWN
MIR34B	—		UNKNOWN
MIR4718	—		UNKNOWN
MIR499A	—		UNKNOWN
MIR502	—		UNKNOWN
MIR510	—		UNKNOWN
MIR890	—		UNKNOWN
MIR892B	—		UNKNOWN
MIR934	—		UNKNOWN
MIR96	Deafness, autosomal dominant 50	OMIM:613074	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
MIRLET7E	—		UNKNOWN
MITF	Tietz albinism-deafness syndrome	OMIM:103500	MENDELIAN
MITF	Melanoma, cutaneous malignant, susceptibility to, 8	OMIM:614456	POLYGENIC
MITF	Coloboma, osteopetrosis, microphthalmia, macrocephaly, albinism, and deafness	OMIM:617306	MENDELIAN
MITF	Waardenburg syndrome, type 2A	OMIM:193510	MENDELIAN
MITF	Waardenburg-Shah syndrome	ORPHA:897	UNKNOWN
MITF	Waardenburg syndrome type 2	ORPHA:895	UNKNOWN
MITF	Tietz syndrome	ORPHA:42665	UNKNOWN
MITF	Familial melanoma	ORPHA:618	UNKNOWN
MKKS	Mckusick-Kaufman syndrome	OMIM:236700	MENDELIAN
MKKS	Bardet-Biedl syndrome 6	OMIM:605231	MENDELIAN
MKKS	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
MKKS	McKusick-Kaufman syndrome	ORPHA:2473	UNKNOWN
MKL1	—		UNKNOWN
MKL2	—		UNKNOWN
MKRN3	Prader-Willi syndrome	OMIM:176270	MENDELIAN
MKRN3	Precocious puberty, central, 2	OMIM:615346	MENDELIAN
MKS1	Bardet-Biedl syndrome 13	OMIM:615990	MENDELIAN
MKS1	Joubert syndrome 28	OMIM:617121	MENDELIAN
MKS1	Meckel syndrome 1	OMIM:249000	MENDELIAN
MKS1	Joubert syndrome with ocular anomaly	ORPHA:220493	UNKNOWN
MKS1	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
MKS1	Meckel syndrome	ORPHA:564	UNKNOWN
MKS1	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
MKX	—		UNKNOWN
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts 1	OMIM:604004	MENDELIAN
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	ORPHA:2478	UNKNOWN
MLF1	—		UNKNOWN
MLH1	Colorectal cancer, hereditary nonpolyposis, type 2	OMIM:609310	MENDELIAN
MLH1	Muir-Torre syndrome	OMIM:158320	MENDELIAN
MLH1	Mismatch repair cancer syndrome 1	OMIM:276300	MENDELIAN
MLH1	Lynch syndrome	ORPHA:144	UNKNOWN
MLH3	Endometrial carcinoma, somatic	OMIM:608089	POLYGENIC
MLH3	Colorectal cancer, hereditary nonpolyposis, type 7	OMIM:614385	MENDELIAN
MLH3	Colorectal cancer	OMIM:114500	MENDELIAN
MLL	—		UNKNOWN
MLLT10	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
MLLT11	—		UNKNOWN
MLPH	Griscelli syndrome, type 3	OMIM:609227	MENDELIAN
MLPH	Griscelli syndrome type 3	ORPHA:79478	UNKNOWN
MLXIPL	Williams-Beuren syndrome	OMIM:194050	MENDELIAN
MLYCD	Malonyl-CoA decarboxylase deficiency	OMIM:248360	MENDELIAN
MLYCD	Malonic aciduria	ORPHA:943	UNKNOWN
MMAA	Methylmalonic aciduria, vitamin B12-responsive, cblA type	OMIM:251100	MENDELIAN
MMAB	Methylmalonic aciduria, vitamin B12-responsive, cblB type	OMIM:251110	MENDELIAN
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	OMIM:277400	MENDELIAN
MMACHC	Methylmalonic acidemia with homocystinuria, type cblC	ORPHA:79282	UNKNOWN
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	OMIM:277410	MENDELIAN
MMADHC	Methylmalonic aciduria, cblD type	OMIM:620953	MENDELIAN
MMADHC	Homocystinuria-megaloblastic anemia, cblD type	OMIM:620952	MENDELIAN
MMADHC	Methylmalonic acidemia with homocystinuria, type cblD	ORPHA:79283	UNKNOWN
MME	Spinocerebellar ataxia 43	OMIM:617018	MENDELIAN
MME	Charcot-Marie-Tooth disease, axonal, type 2T	OMIM:617017	MENDELIAN
MME	Spinocerebellar ataxia type 43	ORPHA:497764	UNKNOWN
MME	Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization	ORPHA:69063	UNKNOWN
MMEL1	Primary biliary cholangitis	ORPHA:186	UNKNOWN
MMP1	Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form	ORPHA:79408	UNKNOWN
MMP10	—		UNKNOWN
MMP12	—		UNKNOWN
MMP13	Metaphyseal chondrodysplasia, Spahr type	OMIM:250400	MENDELIAN
MMP13	Spondyloepimetaphyseal dysplasia, Missouri type	OMIM:602111	MENDELIAN
MMP13	Metaphyseal chondrodysplasia, Spahr type	ORPHA:2501	UNKNOWN
MMP13	Metaphyseal anadysplasia	ORPHA:1040	UNKNOWN
MMP13	Spondyloepimetaphyseal dysplasia, Missouri type	ORPHA:93356	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
MMP14	Winchester syndrome	OMIM:277950	MENDELIAN
MMP14	Multicentric osteolysis-nodulosis-arthropathy spectrum	ORPHA:371428	UNKNOWN
MMP19	Cavitary optic disc anomalies	OMIM:611543	MENDELIAN
MMP2	Multicentric osteolysis, nodulosis, and arthropathy	OMIM:259600	MENDELIAN
MMP2	Multicentric osteolysis-nodulosis-arthropathy spectrum	ORPHA:371428	UNKNOWN
MMP20	Amelogenesis imperfecta, hypomaturation type, iia2	OMIM:612529	MENDELIAN
MMP7	—		UNKNOWN
MMP8	—		UNKNOWN
MMP9	Metaphyseal anadysplasia 2	OMIM:613073	MENDELIAN
MMP9	Metaphyseal anadysplasia	ORPHA:1040	UNKNOWN
MMRN2	—		UNKNOWN
MMS19	—		UNKNOWN
MN1	Meningioma, familial, susceptibility to	OMIM:607174	MENDELIAN
MN1	CEBALID syndrome	OMIM:618774	MENDELIAN
MNAT1	—		UNKNOWN
MND1	—		UNKNOWN
MNX1	Currarino syndrome	OMIM:176450	MENDELIAN
MNX1	Currarino syndrome	ORPHA:1552	UNKNOWN
MOB4	—		UNKNOWN
MOCOS	Xanthinuria, type II	OMIM:603592	MENDELIAN
MOCS1	Molybdenum cofactor deficiency, complementation group A	OMIM:252150	MENDELIAN
MOCS2	Molybdenum cofactor deficiency, complementation group B	OMIM:252160	MENDELIAN
MOG	Narcolepsy 7	OMIM:614250	MENDELIAN
MOG	Narcolepsy type 1	ORPHA:2073	UNKNOWN
MOGS	Congenital disorder of glycosylation, type IIB	OMIM:606056	MENDELIAN
MOGS	MOGS-CDG	ORPHA:79330	UNKNOWN
MOK	—		UNKNOWN
MOV10	—		UNKNOWN
MOV10L1	Spermatogenic failure 73	OMIM:619878	MENDELIAN
MOV10L1	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
MPC1	Mitochondrial pyruvate carrier deficiency	OMIM:614741	MENDELIAN
MPDU1	Congenital disorder of glycosylation, type IF	OMIM:609180	MENDELIAN
MPDU1	MPDU1-CDG	ORPHA:79323	UNKNOWN
MPDZ	Hydrocephalus, congenital, 2, with or without brain or eye anomalies	OMIM:615219	MENDELIAN
MPG	—		UNKNOWN
MPHOSPH8	—		UNKNOWN
MPI	Congenital disorder of glycosylation, type Ib	OMIM:602579	MENDELIAN
MPI	MPI-CDG	ORPHA:79319	UNKNOWN
MPL	Amegakaryocytic thrombocytopenia, congenital	OMIM:604498	MENDELIAN
MPL	Myelofibrosis with myeloid metaplasia, somatic	OMIM:254450	MENDELIAN
MPL	Thrombocythemia 2	OMIM:601977	MENDELIAN
MPL	Familial thrombocytosis	ORPHA:71493	UNKNOWN
MPL	Essential thrombocythemia	ORPHA:3318	UNKNOWN
MPL	Primary myelofibrosis	ORPHA:824	UNKNOWN
MPLKIP	Trichothiodystrophy 4, nonphotosensitive	OMIM:234050	MENDELIAN
MPLKIP	Trichothiodystrophy	ORPHA:33364	UNKNOWN
MPO	Alzheimer disease	OMIM:104300	POLYGENIC
MPO	Myeloperoxidase deficiency	OMIM:254600	MENDELIAN
MPP3	—		UNKNOWN
MPP4	—		UNKNOWN
MPP6	—		UNKNOWN
MPP7	—		UNKNOWN
MPST	—		UNKNOWN
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	OMIM:256810	MENDELIAN
MPV17	Charcot-Marie-Tooth disease, axonal, type 2EE	OMIM:618400	MENDELIAN
MPZ	Roussy-Levy hereditary areflexic dystasia	OMIM:180800	MENDELIAN
MPZ	Charcot-Marie-Tooth disease, axonal, type 2I	OMIM:607677	MENDELIAN
MPZ	Neuropathy, congenital hypomyelinating, 2	OMIM:618184	MENDELIAN
MPZ	Charcot-Marie-Tooth disease, type 1B	OMIM:118200	MENDELIAN
MPZ	Hypertrophic neuropathy of dejerine-sottas	OMIM:145900	MENDELIAN
MPZ	Charcot-Marie-Tooth disease, dominant intermediate D	OMIM:607791	MENDELIAN
MPZ	Charcot-Marie-Tooth disease, type 2J	OMIM:607736	MENDELIAN
MPZ	Charcot-Marie-Tooth disease type 1B	ORPHA:101082	UNKNOWN
MPZ	Roussy-L��vy syndrome	ORPHA:3115	UNKNOWN
MR1	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
MRAP	Glucocorticoid deficiency 2	OMIM:607398	MENDELIAN
MRAP	Familial glucocorticoid deficiency	ORPHA:361	UNKNOWN
MRAP2	Obesity, susceptibility to, BMIQ18	OMIM:615457	POLYGENIC
MRC1	—		UNKNOWN
MRE11	Ataxia-telangiectasia-like disorder 1	OMIM:604391	MENDELIAN
MRE11	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
MRE11	Ataxia-telangiectasia-like disorder	ORPHA:251347	UNKNOWN
MRE11A	—		UNKNOWN
MREG	—		UNKNOWN
MRI1	—		UNKNOWN
MROH9	—		UNKNOWN
MRPL12	Combined oxidative phosphorylation deficiency 45	OMIM:618951	MENDELIAN
MRPL3	Combined oxidative phosphorylation deficiency 9	OMIM:614582	MENDELIAN
MRPL36	—		UNKNOWN
MRPL40	—		UNKNOWN
MRPL44	Combined oxidative phosphorylation deficiency 16	OMIM:615395	MENDELIAN
MRPL48	—		UNKNOWN
MRPS16	Combined oxidative phosphorylation deficiency 2	OMIM:610498	MENDELIAN
MRPS2	Combined oxidative phosphorylation deficiency 36	OMIM:617950	MENDELIAN
MRPS22	Combined oxidative phosphorylation deficiency 5	OMIM:611719	MENDELIAN
MRPS22	Ovarian dysgenesis 7	OMIM:618117	MENDELIAN
MRPS22	46,XX gonadal dysgenesis	ORPHA:243	UNKNOWN
MRRF	—		UNKNOWN
MS4A1	Immunodeficiency, common variable, 5	OMIM:613495	MENDELIAN
MS4A10	—		UNKNOWN
MS4A12	—		UNKNOWN
MS4A2	—		UNKNOWN
MS4A3	—		UNKNOWN
MS4A6A	—		UNKNOWN
MS4A6E	—		UNKNOWN
MSH2	Muir-Torre syndrome	OMIM:158320	MENDELIAN
MSH2	Mismatch repair cancer syndrome 2	OMIM:619096	MENDELIAN
MSH2	Lynch syndrome I	OMIM:120435	MENDELIAN
MSH2	Lynch syndrome	ORPHA:144	UNKNOWN
MSH3	Endometrial carcinoma, somatic	OMIM:608089	MENDELIAN
MSH3	Familial adenomatous polyposis 4	OMIM:617100	MENDELIAN
MSH3	MSH3-related polyposis	ORPHA:480536	UNKNOWN
MSH4	Spermatogenic failure 2	OMIM:108420	MENDELIAN
MSH4	Premature ovarian failure 20	OMIM:619938	MENDELIAN
MSH4	46,XX gonadal dysgenesis	ORPHA:243	UNKNOWN
MSH5	Premature ovarian failure 13	OMIM:617442	MENDELIAN
MSH5	Spermatogenic failure 74	OMIM:619937	MENDELIAN
MSH5	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
MSH6	Endometrial carcinoma, somatic	OMIM:608089	POLYGENIC
MSH6	Colorectal cancer, hereditary nonpolyposis, type 5	OMIM:614350	MENDELIAN
MSH6	Mismatch repair cancer syndrome 3	OMIM:619097	MENDELIAN
MSH6	Lynch syndrome	ORPHA:144	UNKNOWN
MSL3	Basilicata-Akhtar syndrome	OMIM:301032	MENDELIAN
MSMB	—		UNKNOWN
MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis	OMIM:616834	MENDELIAN
MSR1	Barrett esophagus	OMIM:614266	MENDELIAN
MSRA	—		UNKNOWN
MSRB3	Deafness, autosomal recessive 74	OMIM:613718	MENDELIAN
MST1	Primary sclerosing cholangitis	ORPHA:171	UNKNOWN
MST1R	Nasopharyngeal carcinoma, susceptibility to, 3	OMIM:617075	POLYGENIC
MSTN	Muscle hypertrophy	OMIM:614160	MENDELIAN
MSX1	Witkop syndrome	OMIM:189500	MENDELIAN
MSX1	Orofacial cleft 5	OMIM:608874	MENDELIAN
MSX1	Tooth agenesis, selective, 1	OMIM:106600	MENDELIAN
MSX1	Isolated cleft lip	ORPHA:199302	UNKNOWN
MSX1	Cleft lip/palate	ORPHA:199306	UNKNOWN
MSX1	Cleft lip and alveolus	ORPHA:141291	UNKNOWN
MSX1	Oligodontia	ORPHA:99798	UNKNOWN
MSX1	Hypodontia-dysplasia of nails syndrome	ORPHA:2228	UNKNOWN
MSX2	Craniosynostosis 2	OMIM:604757	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
MSX2	Parietal foramina with cleidocranial dysplasia	OMIM:168550	MENDELIAN
MSX2	Parietal foramina	OMIM:168500	MENDELIAN
MSX2	Enlarged parietal foramina	ORPHA:60015	UNKNOWN
MT1A	—		UNKNOWN
MT2A	—		UNKNOWN
MTA1	—		UNKNOWN
MTA2	—		UNKNOWN
MTA3	—		UNKNOWN
MTAP	Diaphyseal medullary stenosis with malignant fibrous histiocytoma	OMIM:112250	MENDELIAN
MTCH2	—		UNKNOWN
MTF1	—		UNKNOWN
MTFMT	Mitochondrial complex I deficiency, nuclear type 27	OMIM:618248	MENDELIAN
MTFMT	Combined oxidative phosphorylation deficiency 15	OMIM:614947	MENDELIAN
MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	OMIM:617780	MENDELIAN
MTHFD1	Neural tube defects, folate-sensitive	OMIM:601634	POLYGENIC
MTHFD1L	—		UNKNOWN
MTHFR	Homocystinuria due to deficiency of n(5,10)-methylenetetrahydrofolatereductase activity	OMIM:236250	MENDELIAN
MTHFR	Thrombophiliavenous thromboembolism, included	OMIM:188050	POLYGENIC
MTHFR	Schizophrenia	OMIM:181500	POLYGENIC
MTHFR	Neural tube defects, folate-sensitive	OMIM:601634	POLYGENIC
MTHFR	Homocystinuria due to methylene tetrahydrofolate reductase deficiency	ORPHA:395	UNKNOWN
MTHFR	Isolated exencephaly	ORPHA:563612	UNKNOWN
MTHFR	Isolated anencephaly	ORPHA:563609	UNKNOWN
MTHFS	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination	OMIM:618367	MENDELIAN
MTM1	Myopathy, centronuclear, X-linked	OMIM:310400	MENDELIAN
MTM1	X-linked myotubular myopathy-abnormal genitalia syndrome	ORPHA:456328	UNKNOWN
MTM1	X-linked centronuclear myopathy	ORPHA:596	UNKNOWN
MTMR12	—		UNKNOWN
MTMR14	Centronuclear myopathy 1	OMIM:160150	POLYGENIC
MTMR14	Autosomal dominant centronuclear myopathy	ORPHA:169189	UNKNOWN
MTMR2	Charcot-Marie-Tooth disease, type 4B1	OMIM:601382	MENDELIAN
MTMR8	—		UNKNOWN
MTMR9	—		UNKNOWN
MTNR1A	—		UNKNOWN
MTNR1B	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
MTO1	Combined oxidative phosphorylation deficiency 10	OMIM:614702	MENDELIAN
MTO1	Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency	ORPHA:314637	UNKNOWN
MTOR	Smith-Kingsmore syndrome	OMIM:616638	MENDELIAN
MTOR	Focal cortical dysplasia of taylor	OMIM:607341	MENDELIAN
MTOR	Hemimegalencephaly	ORPHA:99802	UNKNOWN
MTOR	Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome	ORPHA:457485	UNKNOWN
MTPAP	Spastic ataxia 4, autosomal recessive	OMIM:613672	MENDELIAN
MTPAP	Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome	ORPHA:254343	UNKNOWN
MTR	Homocystinuria-megaloblastic anemia, cbIG complementation type	OMIM:250940	MENDELIAN
MTR	Neural tube defects, folate-sensitive	OMIM:601634	POLYGENIC
MTRR	Homocystinuria-megaloblastic anemia, cbl E type	OMIM:236270	MENDELIAN
MTRR	Neural tube defects, folate-sensitive	OMIM:601634	POLYGENIC
MTRR	Methylcobalamin deficiency type cbIE	ORPHA:2169	UNKNOWN
MTSS1	—		UNKNOWN
MTSS1L	—		UNKNOWN
MTSS2	Intellectual developmental disorder with ocular anomalies and distinctive facial features	OMIM:620086	MENDELIAN
MTTP	Abetalipoproteinemia	OMIM:200100	MENDELIAN
MTTP	Abetalipoproteinemia	ORPHA:14	UNKNOWN
MTUS1	—		UNKNOWN
MUC1	Tubulointerstitial kidney disease, autosomal dominant, 2	OMIM:174000	MENDELIAN
MUC13	—		UNKNOWN
MUC2	—		UNKNOWN
MUC3A	—		UNKNOWN
MUC4	—		UNKNOWN
MUC5B	Pulmonary fibrosis, idiopathic	OMIM:178500	POLYGENIC
MUC5B	Idiopathic pulmonary fibrosis	ORPHA:2032	UNKNOWN
MUC6	—		UNKNOWN
MUC7	Asthma, susceptibility to	OMIM:600807	POLYGENIC
MURC	—		UNKNOWN
MUS81	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
MUSK	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency	OMIM:616325	MENDELIAN
MUSK	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	OMIM:608931	MENDELIAN
MUSK	Fetal akinesia deformation sequence	OMIM:208150	MENDELIAN
MUSK	Fetal akinesia deformation sequence	ORPHA:994	UNKNOWN
MUT	—		UNKNOWN
MUTYH	Familial adenomatous polyposis, 2	OMIM:608456	MENDELIAN
MUTYH	Gastric cancer, somatic	OMIM:613659	MENDELIAN
MUTYH	MUTYH-related polyposis	ORPHA:247798	UNKNOWN
MUTYH	Familial colorectal cancer Type X	ORPHA:440437	UNKNOWN
MVK	Mevalonic aciduria	OMIM:610377	MENDELIAN
MVK	Hyper-Igd syndrome	OMIM:260920	MENDELIAN
MVK	Porokeratosis 3, multiple types	OMIM:175900	MENDELIAN
MVK	Hyperimmunoglobulinemia D with periodic fever	ORPHA:343	UNKNOWN
MVK	Mevalonic aciduria	ORPHA:29	UNKNOWN
MVK	Porokeratosis of Mibelli	ORPHA:735	UNKNOWN
MVK	Disseminated superficial actinic porokeratosis	ORPHA:79152	UNKNOWN
MX1	—		UNKNOWN
MXI1	Prostate cancer	OMIM:176807	MENDELIAN
MXRA5	—		UNKNOWN
MYBBP1A	—		UNKNOWN
MYBL2	—		UNKNOWN
MYBPC1	Arthrogryposis, distal, type 1B	OMIM:614335	MENDELIAN
MYBPC1	Myopathy, congenital, with tremor	OMIM:618524	MENDELIAN
MYBPC1	Lethal congenital contracture syndrome 4	OMIM:614915	MENDELIAN
MYBPC1	Distal arthrogryposis type 1	ORPHA:1146	UNKNOWN
MYBPC3	Cardiomyopathy, familial hypertrophic, 4	OMIM:115197	MENDELIAN
MYBPC3	Left ventricular noncompaction 10	OMIM:615396	MENDELIAN
MYBPC3	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
MYC	Burkitt lymphoma	OMIM:113970	MENDELIAN
MYC	Burkitt lymphoma	ORPHA:543	UNKNOWN
MYCL	—		UNKNOWN
MYCL1	—		UNKNOWN
MYCN	Feingold syndrome 1	OMIM:164280	MENDELIAN
MYCN	Megalencephaly-polydactyly syndrome	OMIM:620748	MENDELIAN
MYCN	Neuroblastoma	ORPHA:635	UNKNOWN
MYCN	Feingold syndrome type 1	ORPHA:391641	UNKNOWN
MYCT1	—		UNKNOWN
MYD88	Macroglobulinemia, Waldenstrom, somatic	OMIM:153600	MENDELIAN
MYD88	Immunodeficiency 68	OMIM:612260	MENDELIAN
MYD88	Waldenström macroglobulinemia	ORPHA:33226	UNKNOWN
MYD88	Transient predisposition to invasive pyogenic bacterial infection	ORPHA:70592	UNKNOWN
MYF5	Ophthalmoplegia, external, with rib and vertebral anomalies	OMIM:618155	MENDELIAN
MYF6	Autosomal dominant centronuclear myopathy	ORPHA:169189	UNKNOWN
MYH11	Visceral myopathy 2	OMIM:619350	MENDELIAN
MYH11	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2	OMIM:619351	MENDELIAN
MYH11	Aortic aneurysm, familial thoracic 4	OMIM:132900	MENDELIAN
MYH11	Familial aortic dissection	ORPHA:229	UNKNOWN
MYH11	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
MYH11	Megacystis-microcolon-intestinal hypoperistalsis syndrome	ORPHA:2241	UNKNOWN
MYH13	—		UNKNOWN
MYH14	Peripheral neuropathy, myopathy, hoarseness, and hearing loss	OMIM:614369	MENDELIAN
MYH14	Deafness, autosomal dominant nonsyndromic sensorineural 4	OMIM:600652	MENDELIAN
MYH14	MYH14-related peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome	ORPHA:397744	UNKNOWN
MYH15	—		UNKNOWN
MYH2	Myopathy, proximal, and ophthalmoplegia	OMIM:605637	MENDELIAN
MYH3	Arthrogryposis, distal, type 2B3	OMIM:618436	MENDELIAN
MYH3	Contractures, pterygia, and spondylcarpostarsal fusion syndrome 1A	OMIM:178110	MENDELIAN
MYH3	Contractures, pterygia, and variable skeletal fusions syndrome 1B	OMIM:618469	MENDELIAN
MYH3	Arthrogryposis, distal, type 2A (Freeman-Sheldon)	OMIM:193700	MENDELIAN
MYH3	Spondylcarpotarsal synostosis	ORPHA:3275	UNKNOWN
MYH3	Freeman-Sheldon syndrome	ORPHA:2053	UNKNOWN
MYH3	Sheldon-Hall syndrome	ORPHA:1147	UNKNOWN
MYH3	Distal arthrogryposis type 1	ORPHA:1146	UNKNOWN
MYH3	Autosomal recessive multiple pterygium syndrome	ORPHA:2990	UNKNOWN
MYH4	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
MYH6	Cardiomyopathy, familial hypertrophic 1	OMIM:192600	MENDELIAN
MYH6	Atrial septal defect 3	OMIM:614089	MENDELIAN
MYH6	Sick sinus syndrome 3	OMIM:614090	POLYGENIC
MYH6	Cardiomyopathy, familial hypertrophic, 14	OMIM:613251	MENDELIAN
MYH6	Cardiomyopathy, dilated, 1EE	OMIM:613252	MENDELIAN
MYH6	Atrial septal defect, ostium secundum type	ORPHA:99103	UNKNOWN
MYH6	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
MYH7	Myopathy, myosin storage, autosomal recessive	OMIM:255160	MENDELIAN
MYH7	Cardiomyopathy, familial hypertrophic 1	OMIM:192600	MENDELIAN
MYH7	Myopathy, distal, 1	OMIM:160500	MENDELIAN
MYH7	Congenital myopathy 7A, myosin storage, autosomal dominant	OMIM:608358	MENDELIAN
MYH7	Cardiomyopathy, dilated, 1S	OMIM:613426	MENDELIAN
MYH7	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
MYH7	Laing distal myopathy	ORPHA:59135	UNKNOWN
MYH7	Ebstein malformation of the tricuspid valve	ORPHA:1880	UNKNOWN
MYH7	Classic multiminicore myopathy	ORPHA:324604	UNKNOWN
MYH8	Arthrogryposis, distal, type 7	OMIM:158300	MENDELIAN
MYH8	Carney complex variant	OMIM:608837	MENDELIAN
MYH8	Trismus-pseudocamptodactyly syndrome	ORPHA:3377	UNKNOWN
MYH9	Deafness, autosomal dominant nonsyndromic sensorineural 17	OMIM:603622	MENDELIAN
MYH9	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss	OMIM:155100	MENDELIAN
MYH9	MYH9-related syndromic thrombocytopenia	ORPHA:182050	UNKNOWN
MYL1	Myopathy, congenital, with fast-twitch (type II) fiber atrophy	OMIM:618414	MENDELIAN
MYL2	Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy	OMIM:619424	MENDELIAN
MYL2	Cardiomyopathy, familial hypertrophic, 10	OMIM:608758	MENDELIAN
MYL2	Congenital fiber-type disproportion myopathy	ORPHA:2020	UNKNOWN
MYL3	Cardiomyopathy, familial hypertrophic, 8	OMIM:608751	MENDELIAN
MYLIP	—		UNKNOWN
MYLK	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1	OMIM:249210	MENDELIAN
MYLK	Aortic aneurysm, familial thoracic 7	OMIM:613780	MENDELIAN
MYLK	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
MYLK	Megacystis-microcolon-intestinal hypoperistalsis syndrome	ORPHA:2241	UNKNOWN
MYLK2	—		UNKNOWN
MYO15A	Deafness, neurosensory, autosomal recessive 3	OMIM:600316	MENDELIAN
MYO18B	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism	OMIM:616549	MENDELIAN
MYO1A	Diarrhea 15, congenital	OMIM:621179	MENDELIAN
MYO1A	Deafness, autosomal dominant 48	OMIM:607841	MENDELIAN
MYO1D	—		UNKNOWN
MYO1E	Focal segmental glomerulosclerosis 6	OMIM:614131	MENDELIAN
MYO1E	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
MYO1F	—		UNKNOWN
MYO3A	Deafness, autosomal recessive 30	OMIM:607101	MENDELIAN
MYO3A	Deafness, autosomal dominant 90	OMIM:620722	MENDELIAN
MYO5A	Griscelli syndrome, type 1	OMIM:214450	MENDELIAN
MYO5A	Griscelli syndrome type 1	ORPHA:79476	UNKNOWN
MYO5A	Griscelli syndrome type 3	ORPHA:79478	UNKNOWN
MYO5A	Neuroectodermal melanolyosomal disease	ORPHA:33445	UNKNOWN
MYO5B	Diarrhea 2, with microvillous atrophy	OMIM:251850	MENDELIAN
MYO5B	Cholestasis, progressive familial intrahepatic, 10	OMIM:619868	MENDELIAN
MYO5B	Microvillus inclusion disease	ORPHA:2290	UNKNOWN
MYO5C	—		UNKNOWN
MYO6	Deafness, autosomal dominant 22	OMIM:606346	MENDELIAN
MYO6	Deafness, autosomal recessive 37	OMIM:607821	MENDELIAN
MYO7A	Usher syndrome, type I	OMIM:276900	MENDELIAN
MYO7A	Deafness, neurosensory, autosomal recessive 2	OMIM:600060	MENDELIAN
MYO7A	Deafness, autosomal dominant nonsyndromic sensorineural 11	OMIM:601317	MENDELIAN
MYO7A	Usher syndrome type 2	ORPHA:231178	UNKNOWN
MYO7A	Usher syndrome type 1	ORPHA:231169	UNKNOWN
MYO7B	—		UNKNOWN
MYO9B	—		UNKNOWN
MYOC	Glaucoma 1, open angle, A	OMIM:137750	MENDELIAN
MYOC	Congenital glaucoma	ORPHA:98976	UNKNOWN
MYOC	Juvenile glaucoma	ORPHA:98977	UNKNOWN
MYOCD	Megabladder, congenital	OMIM:618719	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
MYOD1	Myopathy, congenital, with diaphragmatic defects, respiratory insufficiency, and dysmorphic facies	OMIM:618975	MENDELIAN
MYOD1	Fetal akinesia deformation sequence	ORPHA:994	UNKNOWN
MYOF	Angioedema, hereditary, 7	OMIM:619366	MENDELIAN
MYOM1	—		UNKNOWN
MYOT	Myopathy, myofibrillar, 3	OMIM:609200	MENDELIAN
MYOT	Distal myotilinopathy	ORPHA:98911	UNKNOWN
MYOZ2	Cardiomyopathy, familial hypertrophic, 16	OMIM:613838	MENDELIAN
MYPN	Cardiomyopathy, dilated, 1KK	OMIM:615248	MENDELIAN
MYPN	Nemaline myopathy 11, autosomal recessive	OMIM:617336	MENDELIAN
MYPN	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
MYPN	Familial isolated restrictive cardiomyopathy	ORPHA:75249	UNKNOWN
MYPN	Childhood-onset nemaline myopathy	ORPHA:171439	UNKNOWN
MYPN	Cap myopathy	ORPHA:171881	UNKNOWN
MYT1	—		UNKNOWN
MYT1L	Intellectual developmental disorder, autosomal dominant 39	OMIM:616521	MENDELIAN
MYT1L	MYT1L-related developmental delay-intellectual disability-obesity syndrome	ORPHA:647799	UNKNOWN
NAA10	Microphthalmia, syndromic 1	OMIM:309800	MENDELIAN
NAA10	Ogden syndrome	OMIM:300855	MENDELIAN
NAA10	Ogden syndrome	ORPHA:276432	UNKNOWN
NAA10	Microphthalmia, Lenz type	ORPHA:568	UNKNOWN
NAA15	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities	OMIM:617787	MENDELIAN
NAALADL2	—		UNKNOWN
NAB2	Solitary fibrous tumor	ORPHA:2126	UNKNOWN
NACC1	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination	OMIM:617393	MENDELIAN
NACC1	Severe neurodevelopmental disorder with feeding difficulties-stereotypic hand movement-bilateral cataract	ORPHA:500545	UNKNOWN
NADK2	2,4-dienoyl-CoA reductase deficiency	OMIM:616034	MENDELIAN
NADK2	Progressive encephalopathy with leukodystrophy due to DECR deficiency	ORPHA:431361	UNKNOWN
NAGA	Schindler disease, type I	OMIM:609241	MENDELIAN
NAGA	Kanzaki disease	OMIM:609242	MENDELIAN
NAGA	Alpha-N-acetylgalactosaminidase deficiency type 2	ORPHA:79280	UNKNOWN
NAGA	Alpha-N-acetylgalactosaminidase deficiency type 3	ORPHA:79281	UNKNOWN
NAGA	Alpha-N-acetylgalactosaminidase deficiency type 1	ORPHA:79279	UNKNOWN
NAGLU	Mucopolysaccharidosis type IIIB	OMIM:252920	MENDELIAN
NAGLU	Charcot-Marie-Tooth disease, axonal, type 2V	OMIM:616491	MENDELIAN
NAGPA	—		UNKNOWN
NAGS	N-acetylglutamate synthase deficiency	OMIM:237310	MENDELIAN
NAGS	Hyperammonemia due to N-acetylglutamate synthase deficiency	ORPHA:927	UNKNOWN
NALCN	Congenital contractures of the limbs and face, hypotonia, and developmental delay	OMIM:616266	MENDELIAN
NALCN	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	OMIM:615419	MENDELIAN
NALCN	Freeman-Sheldon syndrome	ORPHA:2053	UNKNOWN
NALCN	Congenital limbs-face contractures-hypotonia-developmental delay syndrome	ORPHA:562528	UNKNOWN
NALCN	Sheldon-Hall syndrome	ORPHA:1147	UNKNOWN
NALCN	Distal arthrogryposis type 1	ORPHA:1146	UNKNOWN
NALP7	—		UNKNOWN
NAMPT	—		UNKNOWN
NANOS1	Spermatogenic failure 12	OMIM:615413	MENDELIAN
NANOS1	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
NANOS2	—		UNKNOWN
NANOS3	—		UNKNOWN
NAPRT1	—		UNKNOWN
NARS2	Combined oxidative phosphorylation deficiency 24	OMIM:616239	MENDELIAN
NARS2	Deafness, autosomal recessive 94	OMIM:618434	MENDELIAN
NARS2	DEND syndrome	ORPHA:79134	UNKNOWN
NAT1	—		UNKNOWN
NAT2	Acetylation, slow	OMIM:243400	MENDELIAN
NAT8L	—		UNKNOWN
NAV1	—		UNKNOWN
NAV2	—		UNKNOWN
NBAS	Infantile liver failure syndrome 2	OMIM:616483	MENDELIAN
NBAS	Short stature, optic nerve atrophy, and pelger-huet anomaly	OMIM:614800	MENDELIAN
NBEA	Neurodevelopmental disorder with or without early-onset generalized epilepsy	OMIM:619157	MENDELIAN
NBEAL2	Gray platelet syndrome	OMIM:139090	MENDELIAN
NBEAL2	Gray platelet syndrome	ORPHA:721	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
NBN	Aplastic anemia	OMIM:609135	MENDELIAN
NBN	Leukemia, acute lymphocytic, susceptibility to, 1	OMIM:613065	MENDELIAN
NBN	Nijmegen breakage syndrome	OMIM:251260	MENDELIAN
NBN	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
NBN	Nijmegen breakage syndrome	ORPHA:647	UNKNOWN
NBPF1	—		UNKNOWN
NCALD	—		UNKNOWN
NCAM1	—		UNKNOWN
NCAM2	—		UNKNOWN
NCAN	—		UNKNOWN
NCAPD2	Microcephaly 21, primary, autosomal recessive	OMIM:617983	MENDELIAN
NCAPG2	Khan-Khan-Katsanis syndrome	OMIM:618460	MENDELIAN
NCF1	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type I	OMIM:233700	MENDELIAN
NCF1	Chronic granulomatous disease	ORPHA:379	UNKNOWN
NCF1	Williams syndrome	ORPHA:904	UNKNOWN
NCF2	Chronic granulomatous disease 2, autosomal recessive	OMIM:233710	MENDELIAN
NCF2	Chronic granulomatous disease	ORPHA:379	UNKNOWN
NCF4	Chronic granulomatous disease 3, autosomal recessive	OMIM:613960	MENDELIAN
NCF4	Chronic granulomatous disease	ORPHA:379	UNKNOWN
NCK2	—		UNKNOWN
NCKAP5	—		UNKNOWN
NCOA1	—		UNKNOWN
NCOA3	—		UNKNOWN
NCOR1	—		UNKNOWN
NCS1	—		UNKNOWN
NCSTN	Acne inversa, familial	OMIM:142690	MENDELIAN
NDE1	Lissencephaly 4	OMIM:614019	MENDELIAN
NDE1	Microhydranencephaly	OMIM:605013	MENDELIAN
NDE1	Lissencephaly syndrome, Norman-Roberts type	ORPHA:89844	UNKNOWN
NDE1	Hydranencephaly	ORPHA:2177	UNKNOWN
NDN	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2	ORPHA:177904	UNKNOWN
NDN	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1	ORPHA:177901	UNKNOWN
NDN	Prader-Willi syndrome due to imprinting mutation	ORPHA:177910	UNKNOWN
NDN	Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15	ORPHA:98754	UNKNOWN
NDNL2	—		UNKNOWN
NDOR1	—		UNKNOWN
NDP	Norrie disease	OMIM:310600	MENDELIAN
NDP	Exudative vitreoretinopathy 2, X-linked	OMIM:305390	MENDELIAN
NDP	Coats disease	ORPHA:190	UNKNOWN
NDP	Familial exudative vitreoretinopathy	ORPHA:891	UNKNOWN
NDP	Retinopathy of prematurity	ORPHA:90050	UNKNOWN
NDP	Persistent hyperplastic primary vitreous	ORPHA:91495	UNKNOWN
NDP	Norrie disease	ORPHA:649	UNKNOWN
NDRG1	Charcot-Marie-Tooth disease, type 4D	OMIM:601455	MENDELIAN
NDRG1	Charcot-Marie-Tooth disease type 4D	ORPHA:99950	UNKNOWN
NDST1	Intellectual developmental disorder, autosomal recessive 46	OMIM:616116	MENDELIAN
NDST2	—		UNKNOWN
NDST3	—		UNKNOWN
NDST4	—		UNKNOWN
NDUFA1	Mitochondrial complex I deficiency, nuclear type 12	OMIM:301020	MENDELIAN
NDUFA1	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFA10	Mitochondrial complex I deficiency, nuclear type 22	OMIM:618243	MENDELIAN
NDUFA11	Mitochondrial complex I deficiency, nuclear type 14	OMIM:618236	MENDELIAN
NDUFA11	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFA12	Mitochondrial complex I deficiency, nuclear type 23	OMIM:618244	MENDELIAN
NDUFA13	Mitochondrial complex I deficiency, nuclear type 28	OMIM:618249	MENDELIAN
NDUFA2	Mitochondrial complex I deficiency, nuclear type 13	OMIM:618235	MENDELIAN
NDUFA3	—		UNKNOWN
NDUFA4	—		UNKNOWN
NDUFA5	—		UNKNOWN
NDUFA6	Mitochondrial complex I deficiency, nuclear type 33	OMIM:618253	MENDELIAN
NDUFA6	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFA7	—		UNKNOWN
NDUFA8	Mitochondrial complex I deficiency, nuclear type 37	OMIM:619272	MENDELIAN
NDUFA9	Mitochondrial complex I deficiency, nuclear type 26	OMIM:618247	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
NDUFA9	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFAB1	—		UNKNOWN
NDUFAF1	Mitochondrial complex I deficiency, nuclear type 11	OMIM:618234	MENDELIAN
NDUFAF1	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10	OMIM:618233	MENDELIAN
NDUFAF2	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFAF3	Mitochondrial complex I deficiency, nuclear type 18	OMIM:618240	MENDELIAN
NDUFAF3	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFAF4	Mitochondrial complex I deficiency, nuclear type 15	OMIM:618237	MENDELIAN
NDUFAF4	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16	OMIM:618238	MENDELIAN
NDUFAF5	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFAF6	Fanconi renotubular syndrome 5	OMIM:618913	MENDELIAN
NDUFAF6	Mitochondrial complex I deficiency, nuclear type 17	OMIM:618239	MENDELIAN
NDUFAF6	Primary Fanconi renotubular syndrome	ORPHA:3337	UNKNOWN
NDUFAF7	—		UNKNOWN
NDUFB1	—		UNKNOWN
NDUFB10	Mitochondrial complex I deficiency, nuclear type 35	OMIM:619003	MENDELIAN
NDUFB10	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFB11	Linear skin defects with multiple congenital anomalies 3	OMIM:300952	MENDELIAN
NDUFB11	Mitochondrial complex I deficiency, nuclear type 30	OMIM:301021	MENDELIAN
NDUFB11	Linear skin defects with multiple congenital anomalies 1	OMIM:309801	MENDELIAN
NDUFB11	Microphthalmia with linear skin defects syndrome	ORPHA:2556	UNKNOWN
NDUFB11	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFB3	Mitochondrial complex I deficiency, nuclear type 25	OMIM:618246	MENDELIAN
NDUFB3	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFB4	—		UNKNOWN
NDUFB6	—		UNKNOWN
NDUFB7	Mitochondrial complex I deficiency, nuclear type 39	OMIM:620135	MENDELIAN
NDUFB8	Mitochondrial complex I deficiency, nuclear type 32	OMIM:618252	MENDELIAN
NDUFB9	Mitochondrial complex I deficiency, nuclear type 24	OMIM:618245	MENDELIAN
NDUFB9	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFC1	—		UNKNOWN
NDUFC2	Mitochondrial complex I deficiency, nuclear type 36	OMIM:619170	MENDELIAN
NDUFS1	Mitochondrial complex I deficiency, nuclear type 5	OMIM:618226	MENDELIAN
NDUFS1	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFS2	Mitochondrial complex I deficiency, nuclear type 6	OMIM:618228	MENDELIAN
NDUFS2	Leber-like hereditary optic neuropathy, autosomal recessive 2	OMIM:620569	MENDELIAN
NDUFS2	Leber hereditary optic neuropathy	ORPHA:104	UNKNOWN
NDUFS2	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFS3	Mitochondrial complex I deficiency, nuclear type 8	OMIM:618230	MENDELIAN
NDUFS3	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1	OMIM:252010	MENDELIAN
NDUFS4	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFS5	—		UNKNOWN
NDUFS6	Mitochondrial complex I deficiency, nuclear type 9	OMIM:618232	MENDELIAN
NDUFS6	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFS7	Mitochondrial complex I deficiency, nuclear type 3	OMIM:618224	MENDELIAN
NDUFS7	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFS8	Mitochondrial complex I deficiency, nuclear type 2	OMIM:618222	MENDELIAN
NDUFS8	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4	OMIM:618225	MENDELIAN
NDUFV1	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFV2	Mitochondrial complex I deficiency, nuclear type 7	OMIM:618229	MENDELIAN
NDUFV2	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NDUFV3	—		UNKNOWN
NEB	Arthrogryposis multiplex congenita 6	OMIM:619334	MENDELIAN
NEB	Nemaline myopathy 2, autosomal recessive	OMIM:256030	MENDELIAN
NEB	Autosomal recessive distal nemalin myopathy	ORPHA:399103	UNKNOWN
NEB	Typical nemaline myopathy	ORPHA:171436	UNKNOWN
NEB	Intermediate nemaline myopathy	ORPHA:171433	UNKNOWN
NEB	Severe congenital nemaline myopathy	ORPHA:171430	UNKNOWN
NEB	Childhood-onset nemaline myopathy	ORPHA:171439	UNKNOWN
NEBL	—		UNKNOWN
NECAP1	Epileptic encephalopathy, early infantile, 21	OMIM:615833	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
NECAP1	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
NEDD4	—		UNKNOWN
NEDD4L	Periventricular nodular heterotopia 7	OMIM:617201	MENDELIAN
NEDD4L	Periventricular nodular heterotopia	ORPHA:98892	UNKNOWN
NEDD9	—		UNKNOWN
NEFH	Amyotrophic lateral sclerosis 1	OMIM:105400	POLYGENIC
NEFH	Charcot-Marie-Tooth disease, axonal, type 2CC	OMIM:616924	MENDELIAN
NEFH	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
NEFL	Charcot-Marie-Tooth disease, dominant intermediate G	OMIM:617882	MENDELIAN
NEFL	Charcot-Marie-Tooth disease, axonal, type 2E	OMIM:607684	MENDELIAN
NEFL	Charcot-Marie-Tooth disease, demyelinating, type 1F	OMIM:607734	MENDELIAN
NEFL	Charcot-Marie-Tooth disease type 1F	ORPHA:101085	UNKNOWN
NEFL	Autosomal dominant Charcot-Marie-Tooth disease type 2E	ORPHA:99939	UNKNOWN
NEFM	—		UNKNOWN
NEGR1	—		UNKNOWN
NEIL1	—		UNKNOWN
NEIL2	—		UNKNOWN
NEIL3	—		UNKNOWN
NEK1	Amyotrophic lateral sclerosis, susceptibility to, 24	OMIM:617892	POLYGENIC
NEK1	Short-Rib thoracic dysplasia 6 with or without polydactyly	OMIM:263520	MENDELIAN
NEK1	Mohr syndrome	OMIM:252100	MENDELIAN
NEK1	Orofaciodigital syndrome type 2	ORPHA:2751	UNKNOWN
NEK1	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
NEK2	Retinitis pigmentosa 67	OMIM:615565	MENDELIAN
NEK2	Retinitis pigmentosa	ORPHA:791	UNKNOWN
NEK8	Renal-Hepatic-Pancreatic dysplasia 2	OMIM:615415	MENDELIAN
NEK8	Nephronophthisis 9	OMIM:613824	MENDELIAN
NEK8	Polycystic kidney disease 8	OMIM:620903	MENDELIAN
NEK8	Renal-hepatic-pancreatic dysplasia	ORPHA:294415	UNKNOWN
NEK8	Autosomal dominant polycystic kidney disease	ORPHA:730	UNKNOWN
NELFA	Wolf-Hirschhorn syndrome	ORPHA:280	UNKNOWN
NELL1	—		UNKNOWN
NEO1	—		UNKNOWN
NEU1	Neuraminidase deficiency	OMIM:256550	MENDELIAN
NEU1	Sialidosis type 1	ORPHA:812	UNKNOWN
NEU1	Juvenile sialidosis type 2	ORPHA:93399	UNKNOWN
NEU1	Congenital sialidosis type 2	ORPHA:93400	UNKNOWN
NEU2	—		UNKNOWN
NEUROD1	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
NEUROD1	Maturity-onset diabetes of the young 6	OMIM:606394	MENDELIAN
NEUROD1	MODY	ORPHA:552	UNKNOWN
NEUROG3	Diarrhea 4, malabsorptive, congenital	OMIM:610370	MENDELIAN
NEUROG3	Enteric anendocrinosis	ORPHA:83620	UNKNOWN
NEXMIF	Intellectual developmental disorder, X-linked 98	OMIM:300912	MENDELIAN
NEXMIF	X-linked intellectual disability, Cantagrel type	ORPHA:85277	UNKNOWN
NEXMIF	Epilepsy with myoclonic-atonic seizures	ORPHA:1942	UNKNOWN
NEXN	Cardiomyopathy, dilated, 2M, autosomal recessive	OMIM:621261	MENDELIAN
NEXN	Cardiomyopathy, dilated, 1cc	OMIM:613122	MENDELIAN
NEXN	Cardiomyopathy, familial hypertrophic, 20	OMIM:613876	MENDELIAN
NEXN	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
NF1	Neurofibromatosis-Noonan syndrome	OMIM:601321	MENDELIAN
NF1	Neurofibromatosis, familial spinal	OMIM:162210	MENDELIAN
NF1	Neurofibromatosis, type 1	OMIM:162200	MENDELIAN
NF1	Watson syndrome	OMIM:193520	MENDELIAN
NF1	Juvenile myelomonocytic leukemia	OMIM:607785	MENDELIAN
NF1	17q11.2 microduplication syndrome	ORPHA:139474	UNKNOWN
NF1	17q11 microdeletion syndrome	ORPHA:97685	UNKNOWN
NF1	Neurofibromatosis-Noonan syndrome	ORPHA:638	UNKNOWN
NF1	Neurofibromatosis type 1 due to NF1 mutation or intragenic deletion	ORPHA:363700	UNKNOWN
NF1	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
NF2	Meningioma, familial, susceptibility to	OMIM:607174	MENDELIAN
NF2	Neurofibromatosis, type II	OMIM:101000	MENDELIAN
NF2	Full schwannomatosis	ORPHA:93921	UNKNOWN
NF2	Full NF2-related schwannomatosis	ORPHA:637	UNKNOWN
NF2	Meningioma	ORPHA:2495	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
NFATC2	Joint contracture, osteochondromas, and B-cell lymphoma	OMIM:620232	MENDELIAN
NFATC3	—		UNKNOWN
NFATC4	—		UNKNOWN
NFE2L1	—		UNKNOWN
NFE2L2	Immunodeficiency, developmental delay, and hypohomocysteinemia	OMIM:617744	MENDELIAN
NFIA	Brain malformations with or without urinary tract defects	OMIM:613735	MENDELIAN
NFIA	1p31p32 microdeletion syndrome	ORPHA:401986	UNKNOWN
NFIB	Macrocephaly, acquired, with impaired intellectual development	OMIM:618286	MENDELIAN
NFIX	Malan syndrome	OMIM:614753	MENDELIAN
NFIX	Marshall-Smith syndrome	OMIM:602535	MENDELIAN
NFIX	19p13.3 microduplication syndrome	ORPHA:447980	UNKNOWN
NFIX	Malan overgrowth syndrome	ORPHA:420179	UNKNOWN
NFIX	Marshall-Smith syndrome	ORPHA:561	UNKNOWN
NFKB1	Immunodeficiency, common variable, 12	OMIM:616576	MENDELIAN
NFKB2	Immunodeficiency, common variable, 10	OMIM:615577	MENDELIAN
NFKB2	Deficiency in anterior pituitary function-variable immunodeficiency syndrome	ORPHA:293978	UNKNOWN
NFKBIA	Ectodermal dysplasia and immunodeficiency 2	OMIM:612132	MENDELIAN
NFKBIA	Hypohidrotic ectodermal dysplasia with immunodeficiency	ORPHA:98813	UNKNOWN
NFKBIB	—		UNKNOWN
NFKBIL1	Rheumatoid arthritis	OMIM:180300	POLYGENIC
NFKBIZ	—		UNKNOWN
NFS1	Combined oxidative phosphorylation deficiency 52	OMIM:619386	MENDELIAN
NFU1	Multiple mitochondrial dysfunctions syndrome 1	OMIM:605711	MENDELIAN
NFU1	Spastic paraplegia 93, autosomal recessive	OMIM:620938	MENDELIAN
NFU1	Multiple mitochondrial dysfunctions syndrome type 1	ORPHA:401869	UNKNOWN
NGEF	—		UNKNOWN
NGF	Neuropathy, hereditary sensory and autonomic, type V	OMIM:608654	MENDELIAN
NGF	Hereditary sensory and autonomic neuropathy type 5	ORPHA:64752	UNKNOWN
NGFR	—		UNKNOWN
NGLY1	Congenital disorder of deglycosylation 1	OMIM:615273	MENDELIAN
NGLY1	Alacrimia-choreoathetosis-liver dysfunction syndrome	ORPHA:404454	UNKNOWN
NHEJ1	Immunodeficiency 124, severe combined	OMIM:611291	MENDELIAN
NHEJ1	Microphthalmia/coloboma 13	OMIM:620968	MENDELIAN
NHEJ1	Cernunnos-XLF deficiency	ORPHA:169079	UNKNOWN
NHLRC1	Myoclonic epilepsy of Lafora 2	OMIM:620681	MENDELIAN
NHLRC1	Lafora disease	ORPHA:501	UNKNOWN
NHP2	Dyskeratosis congenita, autosomal recessive 1	OMIM:224230	MENDELIAN
NHP2	Dyskeratosis congenita, autosomal recessive, 2	OMIM:613987	MENDELIAN
NHP2	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
NHS	Cataract, congenital total, with posterior sutural opacities in heterozygotes	OMIM:302200	MENDELIAN
NHS	Nance-Horan syndrome	OMIM:302350	MENDELIAN
NHS	Nance-Horan syndrome	ORPHA:627	UNKNOWN
NICN1	—		UNKNOWN
NID1	—		UNKNOWN
NIN	Seckel syndrome 7	OMIM:614851	MENDELIAN
NIN	Seckel syndrome	ORPHA:808	UNKNOWN
NIN	Microcephalic primordial dwarfism, Dauber type	ORPHA:319675	UNKNOWN
NINJ1	—		UNKNOWN
NIPA1	Spastic paraplegia 6, autosomal dominant	OMIM:600363	MENDELIAN
NIPA1	Autosomal dominant spastic paraplegia type 6	ORPHA:100988	UNKNOWN
NIPA1	15q11.2 microdeletion syndrome	ORPHA:261183	UNKNOWN
NIPA2	15q11.2 microdeletion syndrome	ORPHA:261183	UNKNOWN
NIPAL4	Ichthyosis, congenital, autosomal recessive 6	OMIM:612281	MENDELIAN
NIPAL4	Lamellar ichthyosis	ORPHA:313	UNKNOWN
NIPAL4	Congenital ichthyosiform erythroderma	ORPHA:79394	UNKNOWN
NIPBL	Cornelia de Lange syndrome 1	OMIM:122470	MENDELIAN
NIPBL	Cornelia de Lange syndrome	ORPHA:199	UNKNOWN
NIPSNAP1	—		UNKNOWN
NIPSNAP3A	—		UNKNOWN
NISCH	—		UNKNOWN
NKAIN2	—		UNKNOWN
NKAIN3	—		UNKNOWN
NKX2-1	Chorea, benign hereditary	OMIM:118700	MENDELIAN
NKX2-1	Thyroid cancer, nonmedullary, 1	OMIM:188550	POLYGENIC
NKX2-1	Choreoathetosis, hypothyroidism, and neonatal respiratory distress	OMIM:610978	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
NKX2-1	Benign hereditary chorea	ORPHA:1429	UNKNOWN
NKX2-1	Brain-lung-thyroid syndrome	ORPHA:209905	UNKNOWN
NKX2-1	Athyreosis	ORPHA:95713	UNKNOWN
NKX2-2	—		UNKNOWN
NKX2-3	—		UNKNOWN
NKX2-5	Hypothyroidism, congenital, nongoitrous, 5	OMIM:225250	MENDELIAN
NKX2-5	Conotruncal heart malformations	OMIM:217095	MENDELIAN
NKX2-5	Tetralogy of Fallot	OMIM:187500	MENDELIAN
NKX2-5	Ventricular septal defect 3	OMIM:614432	MENDELIAN
NKX2-5	Hypoplastic left heart syndrome 2	OMIM:614435	MENDELIAN
NKX2-5	Atrial septal defect 7, with or without AV conduction defects	OMIM:108900	MENDELIAN
NKX2-5	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
NKX2-5	Atrial septal defect-atrioventricular conduction defects syndrome	ORPHA:1479	UNKNOWN
NKX2-5	Atrial septal defect, ostium secundum type	ORPHA:99103	UNKNOWN
NKX2-5	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
NKX2-5	Hereditary progressive cardiac conduction defect	ORPHA:871	UNKNOWN
NKX2-5	Athyreosis	ORPHA:95713	UNKNOWN
NKX2-5	Thyroid ectopia	ORPHA:95712	UNKNOWN
NKX2-5	Deletion 5q35 syndrome	ORPHA:1627	UNKNOWN
NKX2-5	Familial bicuspid aortic valve	ORPHA:402075	UNKNOWN
NKX2-5	Hypoplastic left heart syndrome	ORPHA:2248	UNKNOWN
NKX2-6	Conotruncal heart malformations	OMIM:217095	MENDELIAN
NKX2-6	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
NKX2-6	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
NKX3-1	—		UNKNOWN
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia	OMIM:613330	MENDELIAN
NLGN1	Autism, susceptibility to, 20	OMIM:618830	POLYGENIC
NLGN2	—		UNKNOWN
NLGN3	Autism susceptibility, X-linked 1	OMIM:300425	POLYGENIC
NLGN3	Autism susceptibility 1	OMIM:209850	MENDELIAN
NLGN4X	Autism, susceptibility to, X-linked 2	OMIM:300495	POLYGENIC
NLGN4X	Autism susceptibility 1	OMIM:209850	MENDELIAN
NLGN4Y	—		UNKNOWN
NLRC4	Familial cold autoinflammatory syndrome 4	OMIM:616115	MENDELIAN
NLRC4	Autoinflammation with infantile enterocolitis	OMIM:616050	MENDELIAN
NLRP1	Palmoplantar carcinoma, multiple self-healing	OMIM:615225	MENDELIAN
NLRP1	Respiratory papillomatosis, juvenile recurrent, congenital	OMIM:618803	MENDELIAN
NLRP1	Autoinflammation with arthritis and dyskeratosis	OMIM:617388	MENDELIAN
NLRP1	Vitiligo-Associated multiple autoimmune disease susceptibility 1	OMIM:606579	POLYGENIC
NLRP10	—		UNKNOWN
NLRP11	—		UNKNOWN
NLRP12	Familial cold autoinflammatory syndrome 2	OMIM:611762	MENDELIAN
NLRP13	—		UNKNOWN
NLRP14	—		UNKNOWN
NLRP2	Oocyte/zygote/embryo maturation arrest 18	OMIM:620332	MENDELIAN
NLRP2	Female infertility due to oocyte meiotic arrest	ORPHA:488191	UNKNOWN
NLRP3	Deafness, autosomal dominant 34, with or without inflammation	OMIM:617772	MENDELIAN
NLRP3	Muckle-Wells syndrome	OMIM:191900	MENDELIAN
NLRP3	Cinca syndrome	OMIM:607115	MENDELIAN
NLRP3	Familial cold inflammatory syndrome 1	OMIM:120100	MENDELIAN
NLRP3	Keratoendotheliitis fugax hereditaria	OMIM:148200	MENDELIAN
NLRP3	CINCA syndrome	ORPHA:1451	UNKNOWN
NLRP3	Familial cold urticaria	ORPHA:47045	UNKNOWN
NLRP3	Muckle-Wells syndrome	ORPHA:575	UNKNOWN
NLRP4	—		UNKNOWN
NLRP5	Oocyte/zygote/embryo maturation arrest 19	OMIM:620333	MENDELIAN
NLRP5	Female infertility due to oocyte meiotic arrest	ORPHA:488191	UNKNOWN
NLRP6	—		UNKNOWN
NLRP7	Hydatidiform mole, recurrent, 1	OMIM:231090	MENDELIAN
NLRP7	Oocyte/zygote/embryo maturation arrest 25	OMIM:621471	MENDELIAN
NLRP7	Female infertility due to oocyte meiotic arrest	ORPHA:488191	UNKNOWN
NLRP8	—		UNKNOWN
NLRP9	—		UNKNOWN
NLRX1	—		UNKNOWN
NMB	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
NME1	—		UNKNOWN
NME5	Ciliary dyskinesia, primary, 48, without situs inversus	OMIM:620032	MENDELIAN
NME5	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
NME8	Ciliary dyskinesia, primary, 6	OMIM:610852	MENDELIAN
NME8	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
NMNAT1	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis	OMIM:619260	MENDELIAN
NMNAT1	Leber congenital amaurosis 9	OMIM:608553	MENDELIAN
NMNAT1	Leber congenital amaurosis	ORPHA:65	UNKNOWN
NMNAT1	Cone rod dystrophy	ORPHA:1872	UNKNOWN
NMT2	—		UNKNOWN
NMU	—		UNKNOWN
NNMT	—		UNKNOWN
NNT	Glucocorticoid deficiency 4 with or without mineralocorticoid deficiency	OMIM:614736	MENDELIAN
NNT	Familial glucocorticoid deficiency	ORPHA:361	UNKNOWN
NOBOX	Premature ovarian failure 5	OMIM:611548	MENDELIAN
NOD1	—		UNKNOWN
NOD2	Inflammatory bowel disease 1, Crohn disease	OMIM:266600	POLYGENIC
NOD2	Blau syndrome	OMIM:186580	MENDELIAN
NOD2	Yao syndrome	OMIM:617321	POLYGENIC
NOD2	Blau syndrome	ORPHA:90340	UNKNOWN
NODAL	Heterotaxy, visceral, 5	OMIM:270100	MENDELIAN
NODAL	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
NODAL	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
NODAL	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
NODAL	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
NODAL	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
NODAL	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
NOG	Symphalangism, proximal, 1A	OMIM:185800	MENDELIAN
NOG	Brachydactyly, type B2	OMIM:611377	MENDELIAN
NOG	Stapes ankylosis with broad thumb and toes	OMIM:184460	MENDELIAN
NOG	Tarsal-Carpal coalition syndrome	OMIM:186570	MENDELIAN
NOG	Multiple synostoses syndrome 1	OMIM:186500	MENDELIAN
NOG	Proximal symphalangism	ORPHA:3250	UNKNOWN
NOG	Multiple synostoses syndrome	ORPHA:3237	UNKNOWN
NOG	Tarsal-carpal coalition syndrome	ORPHA:1412	UNKNOWN
NOG	Brachydactyly type B2	ORPHA:140908	UNKNOWN
NOL3	Myoclonus, familial cortical	OMIM:614937	MENDELIAN
NONO	Intellectual developmental disorder, X-linked syndromic 34	OMIM:300967	MENDELIAN
NONO	Macrocephaly-intellectual disability-left ventricular non compaction syndrome	ORPHA:466791	UNKNOWN
NOP10	Dyskeratosis congenita, autosomal recessive 1	OMIM:224230	MENDELIAN
NOP10	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9	OMIM:620400	MENDELIAN
NOP10	Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2	OMIM:620425	MENDELIAN
NOP10	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
NOP56	Spinocerebellar ataxia 36	OMIM:614153	MENDELIAN
NOP56	Spinocerebellar ataxia type 36	ORPHA:276198	UNKNOWN
NOS1	Idiopathic achalasia	ORPHA:930	UNKNOWN
NOS1AP	Nephrotic syndrome, type 22	OMIM:619155	MENDELIAN
NOS3	Moyamoya disease 8	OMIM:621469	MENDELIAN
NOTCH1	Aortic valve disease 1	OMIM:109730	MENDELIAN
NOTCH1	Adams-Oliver syndrome 5	OMIM:616028	MENDELIAN
NOTCH1	Adams-Oliver syndrome	ORPHA:974	UNKNOWN
NOTCH1	Familial bicuspid aortic valve	ORPHA:402075	UNKNOWN
NOTCH2	Alagille syndrome 2	OMIM:610205	MENDELIAN
NOTCH2	Hajdu-Cheney syndrome	OMIM:102500	MENDELIAN
NOTCH2	Hajdu-Cheney syndrome	ORPHA:955	UNKNOWN
NOTCH3	Myofibromatosis, infantile 2	OMIM:615293	MENDELIAN
NOTCH3	Lateral meningocele syndrome	OMIM:130720	MENDELIAN
NOTCH3	Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 1	OMIM:621295	MENDELIAN
NOTCH3	Cerebral arteriopathy, autosomal dominant, with subcortical infarctsand leukoencephalopathy	OMIM:125310	MENDELIAN
NOTCH3	Lipodystrophy, familial partial, type 1	OMIM:608600	MENDELIAN
NOTCH3	Lateral meningocele syndrome	ORPHA:2789	UNKNOWN
NOTCH3	CADASIL	ORPHA:136	UNKNOWN
NOTCH3	Infantile myofibromatosis	ORPHA:2591	UNKNOWN
NOTCH4	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
NOVA2	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities	OMIM:618859	MENDELIAN
NOX3	—		UNKNOWN
NPAP1	Prader-Willi syndrome	OMIM:176270	MENDELIAN
NPAS2	—		UNKNOWN
NPAS3	—		UNKNOWN
NPAS4	—		UNKNOWN
NPAT	Familial colorectal cancer Type X	ORPHA:440437	UNKNOWN
NPC1	Niemann-pick disease, type C1	OMIM:257220	MENDELIAN
NPC1L1	—		UNKNOWN
NPC2	Niemann-pick disease, type C2	OMIM:607625	MENDELIAN
NPFFR2	—		UNKNOWN
NPHP1	Joubert syndrome 4	OMIM:609583	MENDELIAN
NPHP1	Senior-Loken syndrome 1	OMIM:266900	MENDELIAN
NPHP1	Nephronophthisis 1	OMIM:256100	MENDELIAN
NPHP1	Joubert syndrome with renal defect	ORPHA:220497	UNKNOWN
NPHP1	Senior-Loken syndrome	ORPHA:3156	UNKNOWN
NPHP1	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
NPHP3	Meckel syndrome, type 7	OMIM:267010	MENDELIAN
NPHP3	Nephronophthisis 3	OMIM:604387	MENDELIAN
NPHP3	Renal-hepatic-pancreatic dysplasia 1	OMIM:208540	MENDELIAN
NPHP3	Senior-Loken syndrome	ORPHA:3156	UNKNOWN
NPHP3	NPHP3-related Meckel-like syndrome	ORPHA:3032	UNKNOWN
NPHP3	Renal-hepatic-pancreatic dysplasia	ORPHA:294415	UNKNOWN
NPHP4	Senior-Loken syndrome 4	OMIM:606996	MENDELIAN
NPHP4	Nephronophthisis 4	OMIM:606966	MENDELIAN
NPHP4	Senior-Loken syndrome	ORPHA:3156	UNKNOWN
NPHS1	Nephrotic syndrome, type 1	OMIM:256300	MENDELIAN
NPHS1	Congenital nephrotic syndrome, Finnish type	ORPHA:839	UNKNOWN
NPHS1	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
NPHS2	Nephrotic syndrome, type 2	OMIM:600995	MENDELIAN
NPHS2	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
NPL	—		UNKNOWN
NPM1	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
NPM1	Acute promyelocytic leukemia	ORPHA:520	UNKNOWN
NPM1	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
NPPA	Atrial standstill 2	OMIM:615745	MENDELIAN
NPPA	Atrial fibrillation, familial, 6	OMIM:612201	MENDELIAN
NPPA	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
NPPA	Isolated atrial standstill	ORPHA:1344	UNKNOWN
NPPB	—		UNKNOWN
NPPC	—		UNKNOWN
NPR1	—		UNKNOWN
NPR2	Short stature with nonspecific skeletal abnormalities	OMIM:616255	MENDELIAN
NPR2	Epiphyseal chondrodysplasia, Miura type	OMIM:615923	MENDELIAN
NPR2	Acromesomelic dysplasia, Maroteaux type	OMIM:602875	MENDELIAN
NPR2	Acromesomelic dysplasia, Maroteaux type	ORPHA:40	UNKNOWN
NPR3	Boudin-Mortier syndrome	OMIM:619543	MENDELIAN
NPSR1	Short sleep, familial natural, 3	OMIM:621336	MENDELIAN
NPTN	—		UNKNOWN
NPY	—		UNKNOWN
NPY1R	—		UNKNOWN
NPY2R	—		UNKNOWN
NQO1	—		UNKNOWN
NQO2	Breast cancer	OMIM:114480	POLYGENIC
NR0B1	Dosage-Sensitive sex reversal	OMIM:300018	MENDELIAN
NR0B1	Adrenal hypoplasia, congenital	OMIM:300200	MENDELIAN
NR0B1	46,XX testicular difference of sex development	ORPHA:393	UNKNOWN
NR0B1	46,XY partial gonadal dysgenesis	ORPHA:251510	UNKNOWN
NR0B1	46,XY complete gonadal dysgenesis	ORPHA:242	UNKNOWN
NR0B1	X-linked adrenal hypoplasia congenita	ORPHA:95702	UNKNOWN
NR0B2	Obesity	OMIM:601665	MENDELIAN
NR1H2	—		UNKNOWN
NR1H3	—		UNKNOWN
NR1H4	Cholestasis, progressive familial intrahepatic, 5	OMIM:617049	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
NR1H4	Intrahepatic cholestasis of pregnancy	ORPHA:69665	UNKNOWN
NR1I2	—		UNKNOWN
NR1I3	—		UNKNOWN
NR2E1	—		UNKNOWN
NR2E3	Retinitis pigmentosa 37	OMIM:611131	MENDELIAN
NR2E3	Enhanced S-cone syndrome	OMIM:268100	MENDELIAN
NR2E3	Retinitis pigmentosa	ORPHA:791	UNKNOWN
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome	OMIM:615722	MENDELIAN
NR2F1	Optic atrophy-intellectual disability syndrome	ORPHA:401777	UNKNOWN
NR2F2	46,XX sex reversal 5	OMIM:618901	MENDELIAN
NR2F2	Congenital heart defects, multiple types, 4	OMIM:615779	MENDELIAN
NR3C1	Glucocorticoid resistance	OMIM:615962	MENDELIAN
NR3C1	Cushing disease	ORPHA:96253	UNKNOWN
NR3C1	Generalized glucocorticoid resistance syndrome	ORPHA:786	UNKNOWN
NR3C2	Pseudohypoadosteronism, type I, autosomal dominant	OMIM:177735	MENDELIAN
NR3C2	Hypertension, early-onset, autosomal dominant, with severe exacerbationin pregnancy	OMIM:605115	MENDELIAN
NR4A1	—		UNKNOWN
NR4A2	Parkinson disease, late-onset	OMIM:168600	MENDELIAN
NR4A2	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism	OMIM:619911	MENDELIAN
NR4A2	Autosomal dominant dopa-responsive dystonia	ORPHA:98808	UNKNOWN
NR4A2	Developmental delay-language impairment-dopa responsive dystonia-parkinsonism syndrome due to 2q24 microdeletion	ORPHA:1617	UNKNOWN
NR4A3	Chondrosarcoma, extraskeletal myxoid	OMIM:612237	MENDELIAN
NR5A1	Premature ovarian failure 7	OMIM:612964	MENDELIAN
NR5A1	46XY sex reversal 3	OMIM:612965	MENDELIAN
NR5A1	46,xx sex reversal 4	OMIM:617480	MENDELIAN
NR5A1	Spermatogenic failure 8	OMIM:613957	MENDELIAN
NR5A1	46,XX testicular difference of sex development	ORPHA:393	UNKNOWN
NR5A1	46,XY partial gonadal dysgenesis	ORPHA:251510	UNKNOWN
NR5A1	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
NR5A1	46,XX gonadal dysgenesis	ORPHA:243	UNKNOWN
NR5A1	46,XY complete gonadal dysgenesis	ORPHA:242	UNKNOWN
NR5A1	46,XX ovotesticular difference of sex development	ORPHA:2138	UNKNOWN
NRAS	Epidermal nevus, somatic	OMIM:162900	MENDELIAN
NRAS	Neurocutaneous melanosis, somatic	OMIM:249400	MENDELIAN
NRAS	Thyroid cancer, nonmedullary, 2	OMIM:188470	MENDELIAN
NRAS	Schimmelpenning-Feuerstein-Mims syndrome	OMIM:163200	MENDELIAN
NRAS	Noonan syndrome 6	OMIM:613224	MENDELIAN
NRAS	Colorectal cancer	OMIM:114500	MENDELIAN
NRAS	RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic	OMIM:614470	MENDELIAN
NRAS	Melanocytic nevus syndrome, congenital	OMIM:137550	MENDELIAN
NRAS	Langerhans cell histiocytosis	ORPHA:389	UNKNOWN
NRAS	Linear nevus sebaceus syndrome	ORPHA:2612	UNKNOWN
NRAS	Large/giant congenital melanocytic nevus	ORPHA:626	UNKNOWN
NRAS	Noonan syndrome	ORPHA:648	UNKNOWN
NRCAM	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities	OMIM:619833	MENDELIAN
NRG1	—		UNKNOWN
NRG3	—		UNKNOWN
NRIP1	Congenital anomalies of kidney and urinary tract 3	OMIM:618270	MENDELIAN
NRL	Enhanced S-cone syndrome 2	OMIM:621371	MENDELIAN
NRL	Retinitis pigmentosa 27	OMIM:613750	MENDELIAN
NRL	Retinitis pigmentosa	ORPHA:791	UNKNOWN
NRN1	—		UNKNOWN
NRP2	—		UNKNOWN
NRTN	Hirschsprung disease	ORPHA:388	UNKNOWN
NRXN1	Pitt-Hopkins-Like syndrome 2	OMIM:614325	MENDELIAN
NRXN1	Schizophrenia, susceptibility to, 17	OMIM:621407	POLYGENIC
NRXN2	—		UNKNOWN
NRXN3	—		UNKNOWN
NSD1	Sotos syndrome	OMIM:117550	MENDELIAN
NSD1	Sotos syndrome	ORPHA:821	UNKNOWN
NSD1	Deletion 5q35 syndrome	ORPHA:1627	UNKNOWN
NSD1	Weaver syndrome	ORPHA:3447	UNKNOWN
NSD1	5q35 microduplication syndrome	ORPHA:228415	UNKNOWN
NSD2	Wolf-Hirschhorn syndrome	OMIM:194190	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
NSD2	Rauch-Steindl syndrome	OMIM:619695	MENDELIAN
NSD2	Wolf-Hirschhorn syndrome	ORPHA:280	UNKNOWN
NSDHL	Congenital hemidysplasia with ichthyosiform erythroderma and limb defects	OMIM:308050	MENDELIAN
NSDHL	CK syndrome	OMIM:300831	MENDELIAN
NSDHL	CHILD syndrome	ORPHA:139	UNKNOWN
NSDHL	CK syndrome	ORPHA:251383	UNKNOWN
NSMF	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
NSMF	Hypogonadotropic hypogonadism 9 with or without anosmia	OMIM:614838	MENDELIAN
NSMF	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
NSUN2	Intellectual developmental disorder, autosomal recessive 5	OMIM:611091	MENDELIAN
NSUN7	—		UNKNOWN
NT5C1B	—		UNKNOWN
NT5C2	Spastic paraplegia 45, autosomal recessive	OMIM:613162	MENDELIAN
NT5C2	Autosomal recessive spastic paraplegia type 45	ORPHA:320396	UNKNOWN
NT5C3A	Uridine 5-prime monophosphate hydrolase deficiency, hemolytic anemiadue to	OMIM:266120	MENDELIAN
NT5E	Calcification of joints and arteries	OMIM:211800	MENDELIAN
NT5E	Hereditary arterial and articular multiple calcification syndrome	ORPHA:289601	UNKNOWN
NTAN1	—		UNKNOWN
NTF3	—		UNKNOWN
NTF4	Glaucoma 1, open angle, O	OMIM:613100	MENDELIAN
NTHL1	Familial adenomatous polyposis 3	OMIM:616415	MENDELIAN
NTHL1	NTHL1-related polyposis	ORPHA:454840	UNKNOWN
NTM	—		UNKNOWN
NTNG1	Atypical Rett syndrome	ORPHA:3095	UNKNOWN
NTNG2	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia	OMIM:618718	MENDELIAN
NTRK1	Insensitivity to pain, congenital, with anhidrosis	OMIM:256800	MENDELIAN
NTRK1	Thyroid carcinoma, familial medullary	OMIM:155240	MENDELIAN
NTRK1	Hereditary sensory and autonomic neuropathy type 5	ORPHA:64752	UNKNOWN
NTRK1	Hereditary sensory and autonomic neuropathy type 4	ORPHA:642	UNKNOWN
NTRK2	Developmental and epileptic encephalopathy 58	OMIM:617830	MENDELIAN
NTRK2	Obesity, hyperphagia, and developmental delay	OMIM:613886	MENDELIAN
NTRK2	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
NTRK2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
NUAK1	—		UNKNOWN
NUB1	—		UNKNOWN
NUBPL	Mitochondrial complex I deficiency, nuclear type 21	OMIM:618242	MENDELIAN
NUBPL	Isolated complex I deficiency	ORPHA:2609	UNKNOWN
NUCB2	—		UNKNOWN
NUDC	—		UNKNOWN
NUDT1	—		UNKNOWN
NUDT15	Thiopurines, poor metabolism of, 2	OMIM:616903	POLYGENIC
NUDT6	—		UNKNOWN
NUDT7	—		UNKNOWN
NUMA1	Acute promyelocytic leukemia	ORPHA:520	UNKNOWN
NUMBL	—		UNKNOWN
NUP107	Nephrotic syndrome, type 11	OMIM:616730	MENDELIAN
NUP107	Galloway-Mowat syndrome 7	OMIM:618348	MENDELIAN
NUP107	Ovarian dysgenesis 6	OMIM:618078	MENDELIAN
NUP107	46,XX gonadal dysgenesis	ORPHA:243	UNKNOWN
NUP107	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
NUP107	Galloway-Mowat syndrome	ORPHA:2065	UNKNOWN
NUP155	Atrial fibrillation, familial, 15	OMIM:615770	MENDELIAN
NUP155	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
NUP214	Leukemia, acute lymphocytic, susceptibility to, 1	OMIM:613065	MENDELIAN
NUP214	Encephalopathy, acute, infection-induced, susceptibility to, 9	OMIM:618426	POLYGENIC
NUP214	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
NUP62	Striatonigral degeneration, infantile	OMIM:271930	MENDELIAN
NUP62	Familial infantile bilateral striatal necrosis	ORPHA:225154	UNKNOWN
NUP93	Nephrotic syndrome, type 12	OMIM:616892	MENDELIAN
NUP93	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
NUTM1	NUT midline carcinoma	ORPHA:443167	UNKNOWN
NXF2	—		UNKNOWN
NXF3	—		UNKNOWN
NXF5	—		UNKNOWN
NXN	Robinow syndrome, autosomal recessive 2	OMIM:618529	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
NXN	Autosomal recessive Robinow syndrome	ORPHA:1507	UNKNOWN
NXNL1	—		UNKNOWN
NYX	Night blindness, congenital stationary, type 1A	OMIM:310500	MENDELIAN
OAS1	Pulmonary alveolar proteinosis with hypogammaglobulinemia	OMIM:618042	MENDELIAN
OAS2	Autoinflammation and autoimmunity, systemic, with immune dysregulation 2	OMIM:621409	MENDELIAN
OAS3	—		UNKNOWN
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia	OMIM:258870	MENDELIAN
OAT	Gyrate atrophy of choroid and retina	ORPHA:414	UNKNOWN
OAZ1	—		UNKNOWN
OBSCN	Rhabdomyolysis, susceptibility to, 1	OMIM:620235	POLYGENIC
OBSCN	Genetic recurrent myoglobinuria	ORPHA:99845	UNKNOWN
OBSL1	3-M syndrome 2	OMIM:612921	MENDELIAN
OBSL1	3M syndrome	ORPHA:2616	UNKNOWN
OCA2	Albinism, oculocutaneous, type II	OMIM:203200	MENDELIAN
OCA2	Oculocutaneous albinism type 2	ORPHA:79432	UNKNOWN
OCA2	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2	ORPHA:177904	UNKNOWN
OCA2	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1	ORPHA:177901	UNKNOWN
OCA2	Angelman syndrome due to maternal 15q11q13 deletion	ORPHA:98794	UNKNOWN
OCA2	Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15	ORPHA:98754	UNKNOWN
OCLN	Band-Like calcification with simplified gyration and polymicrogyria	OMIM:251290	MENDELIAN
OCLN	Pseudo-TORCH syndrome type 1	ORPHA:1229	UNKNOWN
OCRL	Dent disease 2	OMIM:300555	MENDELIAN
OCRL	Lowe syndrome	OMIM:309000	MENDELIAN
OCRL	Oculocerebrorenal syndrome of Lowe	ORPHA:534	UNKNOWN
ODC1	Neurodevelopmental disorder with alopecia and brain abnormalities	OMIM:619075	MENDELIAN
ODC1	Global developmental delay-alopecia-macrocephaly-facial dysmorphism-structural brain anomalies syndrome	ORPHA:544488	UNKNOWN
OFD1	Retinitis pigmentosa 23	OMIM:300424	MENDELIAN
OFD1	Simpson-Golabi-Behmel syndrome, type 2	OMIM:300209	MENDELIAN
OFD1	Joubert syndrome 10	OMIM:300804	MENDELIAN
OFD1	Orofaciodigital syndrome I	OMIM:311200	MENDELIAN
OFD1	Orofaciodigital syndrome type 1	ORPHA:2750	UNKNOWN
OFD1	Orofaciodigital syndrome type 6	ORPHA:2754	UNKNOWN
OFD1	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
OFD1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
OFD1	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
OGDH	Oxoglutarate dehydrogenase deficiency	OMIM:203740	MENDELIAN
OGDH	Oxoglutaric aciduria	ORPHA:31	UNKNOWN
OGG1	Renal cell carcinoma, nonpapillary	OMIM:144700	MENDELIAN
OGT	Intellectual developmental disorder, X-linked 106	OMIM:300997	MENDELIAN
OLFM2	—		UNKNOWN
OLIG2	—		UNKNOWN
OMG	—		UNKNOWN
OOEP	—		UNKNOWN
OPA1	Mitochondrial DNA depletion syndrome 14A (encephalomyopathic type)	OMIM:621481	MENDELIAN
OPA1	Optic atrophy 1	OMIM:165500	MENDELIAN
OPA1	Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type)	OMIM:616896	MENDELIAN
OPA1	Behr syndrome	OMIM:210000	MENDELIAN
OPA1	Optic atrophy with or without deafness, ophthalmoplegia, myopathy, ataxia, and neuropathy	OMIM:125250	MENDELIAN
OPA1	Autosomal dominant optic atrophy, classic form	ORPHA:98673	UNKNOWN
OPA1	Autosomal dominant optic atrophy plus syndrome	ORPHA:1215	UNKNOWN
OPA3	Optic atrophy 3, autosomal dominant	OMIM:165300	MENDELIAN
OPA3	3-methylglutaconic aciduria, type III	OMIM:258501	MENDELIAN
OPA3	Autosomal dominant optic atrophy and cataract	ORPHA:67036	UNKNOWN
OPA3	3-methylglutaconic aciduria type 3	ORPHA:67047	UNKNOWN
OPCML	Ovarian cancer	OMIM:167000	MENDELIAN
OPHN1	Intellectual developmental disorder, X-linked syndromic, Billuart type	OMIM:300486	MENDELIAN
OPHN1	X-linked intellectual disability-cerebellar hypoplasia syndrome	ORPHA:137831	UNKNOWN
OPLAH	5-@oxoprolinase deficiency	OMIM:260005	MENDELIAN
OPLAH	5-oxoprolinase deficiency	ORPHA:33572	UNKNOWN
OPN1LW	Colorblindness, partial, protan series	OMIM:303900	MENDELIAN
OPN1LW	Blue cone monochromacy	OMIM:303700	MENDELIAN
OPN1LW	Blue cone monochromatism	ORPHA:16	UNKNOWN
OPN1LW	Cone rod dystrophy	ORPHA:1872	UNKNOWN
OPN1MW	Colorblindness, deutan	OMIM:303800	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
OPN1MW	Blue cone monochromacy	OMIM:303700	MENDELIAN
OPN1MW	Blue cone monochromatism	ORPHA:16	UNKNOWN
OPN1MW	Cone rod dystrophy	ORPHA:1872	UNKNOWN
OPN1SW	Colorblindness, tritan	OMIM:190900	MENDELIAN
OPN1SW	Tritanopia	ORPHA:88629	UNKNOWN
OPN4	—		UNKNOWN
OPRD1	—		UNKNOWN
OPRK1	—		UNKNOWN
OPRL1	—		UNKNOWN
OPRM1	—		UNKNOWN
OPTC	—		UNKNOWN
OPTN	Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia	OMIM:613435	MENDELIAN
OPTN	Glaucoma, primary open angle	OMIM:137760	MENDELIAN
OPTN	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
OR10V1	—		UNKNOWN
OR10X1	—		UNKNOWN
OR13G1	—		UNKNOWN
OR13H1	—		UNKNOWN
OR1B1	—		UNKNOWN
OR1J1	—		UNKNOWN
OR2M2	—		UNKNOWN
OR4M2	—		UNKNOWN
OR51G1	—		UNKNOWN
OR52H1	—		UNKNOWN
OR52N4	—		UNKNOWN
OR5AC2	—		UNKNOWN
OR5H6	—		UNKNOWN
OR7D4	—		UNKNOWN
OR8H1	—		UNKNOWN
OR8K3	—		UNKNOWN
ORAI1	Immunodeficiency 9	OMIM:612782	MENDELIAN
ORAI1	Myopathy, tubular aggregate, 2	OMIM:615883	MENDELIAN
ORAI1	Stormorken-Sjaastad-Langslet syndrome	ORPHA:3204	UNKNOWN
ORAI1	Tubular aggregate myopathy	ORPHA:2593	UNKNOWN
ORC1	Meier-Gorlin syndrome 1	OMIM:224690	MENDELIAN
ORC1	Ear-patella-short stature syndrome	ORPHA:2554	UNKNOWN
ORC4	Meier-Gorlin syndrome 2	OMIM:613800	MENDELIAN
ORC4	Ear-patella-short stature syndrome	ORPHA:2554	UNKNOWN
ORC6	Meier-Gorlin syndrome 3	OMIM:613803	MENDELIAN
ORC6	Ear-patella-short stature syndrome	ORPHA:2554	UNKNOWN
ORM1	—		UNKNOWN
ORMDL3	—		UNKNOWN
OSBPL2	Deafness, autosomal dominant 67	OMIM:616340	MENDELIAN
OSBPL2	Dyschromatosis, ichthyosis, deafness, and atopic disease	OMIM:621400	MENDELIAN
OSBPL5	—		UNKNOWN
OSGIN1	—		UNKNOWN
OSMR	Amyloidosis, primary localized cutaneous, 1	OMIM:105250	MENDELIAN
OSR1	—		UNKNOWN
OSTM1	Osteopetrosis, autosomal recessive 5	OMIM:259720	MENDELIAN
OSTM1	Autosomal recessive malignant osteopetrosis	ORPHA:667	UNKNOWN
OTC	Ornithine transcarbamylase deficiency, hyperammonemia due to	OMIM:311250	MENDELIAN
OTC	Ornithine transcarbamylase deficiency	ORPHA:664	UNKNOWN
OTOA	Deafness, autosomal recessive 22	OMIM:607039	MENDELIAN
OTOF	Deafness, autosomal recessive 9	OMIM:601071	MENDELIAN
OTOG	Deafness, autosomal recessive 18B	OMIM:614945	MENDELIAN
OTOGL	Deafness, autosomal recessive 84B	OMIM:614944	MENDELIAN
OTOR	—		UNKNOWN
OTUD4	—		UNKNOWN
OTX2	Microphthalmia, syndromic 5	OMIM:610125	MENDELIAN
OTX2	Pituitary hormone deficiency, combined, 6	OMIM:613986	MENDELIAN
OTX2	Septo-optic dysplasia spectrum	ORPHA:3157	UNKNOWN
OTX2	Leber congenital amaurosis	ORPHA:65	UNKNOWN
OTX2	Combined pituitary hormone deficiencies, genetic forms	ORPHA:95494	UNKNOWN
OTX2	Agnathia-holoprosencephaly-situs inversus syndrome	ORPHA:990	UNKNOWN
OTX2	Isolated complete colobomatous microphthalmia	ORPHA:35612	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
OTX2	Severe early-childhood-onset retinal dystrophy	ORPHA:364055	UNKNOWN
OVCH2	—		UNKNOWN
OVGP1	—		UNKNOWN
OXA1L	—		UNKNOWN
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency	OMIM:245050	MENDELIAN
OXCT1	Succinyl-CoA:3-oxoacid CoA transferase deficiency	ORPHA:832	UNKNOWN
OXTR	—		UNKNOWN
P2RX1	—		UNKNOWN
P2RX2	Deafness, autosomal dominant 41	OMIM:608224	MENDELIAN
P2RX4	—		UNKNOWN
P2RX5	—		UNKNOWN
P2RY1	—		UNKNOWN
P2RY11	Narcolepsy type 1	ORPHA:2073	UNKNOWN
P2RY12	Bleeding disorder, platelet-type, 8	OMIM:609821	MENDELIAN
P2RY4	—		UNKNOWN
PABPC1	—		UNKNOWN
PABPC4L	—		UNKNOWN
PABPN1	Oculopharyngeal muscular dystrophy	OMIM:164300	MENDELIAN
PABPN1	Oculopharyngeal muscular dystrophy	ORPHA:270	UNKNOWN
PACRG	—		UNKNOWN
PACS1	Schuurs-Hoeijmakers syndrome	OMIM:615009	MENDELIAN
PACS1	Schuurs-Hoeijmakers syndrome	ORPHA:329224	UNKNOWN
PACS2	Epileptic encephalopathy, early infantile, 66	OMIM:618067	MENDELIAN
PACS2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
PADI4	—		UNKNOWN
PADI6	Preimplantation embryonic lethality 2	OMIM:617234	MENDELIAN
PADI6	Female infertility due to oocyte meiotic arrest	ORPHA:488191	UNKNOWN
PAFAH1B1	Lissencephaly 1	OMIM:607432	MENDELIAN
PAFAH1B1	17p13.3 microduplication syndrome	ORPHA:217385	UNKNOWN
PAFAH1B1	Miller-Dieker syndrome	ORPHA:531	UNKNOWN
PAFAH1B1	Lissencephaly due to LIS1 mutation	ORPHA:95232	UNKNOWN
PAFAH1B3	—		UNKNOWN
PAH	Phenylketonuria	OMIM:261600	MENDELIAN
PAH	Maternal phenylketonuria syndrome	ORPHA:2209	UNKNOWN
PAK1	Intellectual developmental disorder with macrocephaly, seizures, and speech delay	OMIM:618158	MENDELIAN
PAK3	Intellectual developmental disorder, X-linked 30	OMIM:300558	MENDELIAN
PAK7	—		UNKNOWN
PALB2	Fanconi anemia, complementation group N	OMIM:610832	MENDELIAN
PALB2	Pancreatic cancer, susceptibility to, 3	OMIM:613348	POLYGENIC
PALB2	Breast-ovarian cancer, familial, susceptibility to, 5	OMIM:620442	POLYGENIC
PALB2	Fanconi anemia	ORPHA:84	UNKNOWN
PALB2	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
PALB2	Familial pancreatic carcinoma	ORPHA:1333	UNKNOWN
PALLD	Pancreatic cancer, susceptibility to, 1	OMIM:606856	POLYGENIC
PALLD	Familial pancreatic carcinoma	ORPHA:1333	UNKNOWN
PAM16	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melki type	OMIM:613320	MENDELIAN
PAMR1	—		UNKNOWN
PANK2	Neurodegeneration with brain iron accumulation 1	OMIM:234200	MENDELIAN
PANK2	Atypical pantothenate kinase-associated neurodegeneration	ORPHA:216873	UNKNOWN
PANK2	Classic pantothenate kinase-associated neurodegeneration	ORPHA:216866	UNKNOWN
PAPD7	—		UNKNOWN
PAPOLA	—		UNKNOWN
PAPSS2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes	OMIM:612847	MENDELIAN
PARD3B	—		UNKNOWN
PARD6A	—		UNKNOWN
PARD6G	—		UNKNOWN
PARK2	—		UNKNOWN
PARK7	Parkinson disease 7, autosomal recessive early-onset	OMIM:606324	MENDELIAN
PARK7	Young-onset Parkinson disease	ORPHA:2828	UNKNOWN
PARL	—		UNKNOWN
PARN	Dyskeratosis congenita, autosomal recessive 6	OMIM:616353	MENDELIAN
PARN	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4	OMIM:616371	MENDELIAN
PARN	Idiopathic pulmonary fibrosis	ORPHA:2032	UNKNOWN
PARN	Hoyeraal-Hreidarsson syndrome	ORPHA:3322	UNKNOWN
PARN	Dyskeratosis congenita	ORPHA:1775	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
PARP1	—		UNKNOWN
PARP2	—		UNKNOWN
PARP3	—		UNKNOWN
PARP4	—		UNKNOWN
PARS2	Epileptic encephalopathy, early infantile, 75	OMIM:618437	MENDELIAN
PARS2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
PASD1	—		UNKNOWN
PASK	—		UNKNOWN
PATL2	Oocyte maturation defect 4	OMIM:617743	MENDELIAN
PATL2	Female infertility due to oocyte meiotic arrest	ORPHA:488191	UNKNOWN
PAWR	—		UNKNOWN
PAX1	Otofaciocervical syndrome 2	OMIM:615560	MENDELIAN
PAX2	Papillorenal syndrome	OMIM:120330	MENDELIAN
PAX2	Focal segmental glomerulosclerosis 7	OMIM:616002	MENDELIAN
PAX2	Renal coloboma syndrome	ORPHA:1475	UNKNOWN
PAX2	Renal hypoplasia, bilateral	ORPHA:97362	UNKNOWN
PAX2	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
PAX3	Waardenburg syndrome, type 3	OMIM:148820	MENDELIAN
PAX3	Craniofacial-deafness-hand syndrome	OMIM:122880	MENDELIAN
PAX3	Waardenburg syndrome, type 1	OMIM:193500	MENDELIAN
PAX3	Rhabdomyosarcoma 2, alveolar	OMIM:268220	MENDELIAN
PAX3	Craniofacial-deafness-hand syndrome	ORPHA:1529	UNKNOWN
PAX3	Waardenburg syndrome type 3	ORPHA:896	UNKNOWN
PAX3	Waardenburg syndrome type 1	ORPHA:894	UNKNOWN
PAX4	Diabetes mellitus, noninsulin-dependent	OMIM:125853	MENDELIAN
PAX4	Maturity-onset diabetes of the young, type IX	OMIM:612225	MENDELIAN
PAX4	Diabetes mellitus, ketosis-prone	OMIM:612227	POLYGENIC
PAX4	MODY	ORPHA:552	UNKNOWN
PAX6	Foveal hypoplasia and presenile cataract syndromefoveal hypoplasia, isolated, included	OMIM:136520	MENDELIAN
PAX6	Anterior segment dysgenesis 5, multiple subtypes	OMIM:604229	MENDELIAN
PAX6	Optic nerve hypoplasia	OMIM:165550	MENDELIAN
PAX6	Wilms tumor, aniridia, genitourinary anomalies and impaired intellectual development syndrome	OMIM:194072	MENDELIAN
PAX6	Coloboma, ocular	OMIM:120200	MENDELIAN
PAX6	Coloboma of optic nerveoptic nerve head pits, bilateral congenital, included	OMIM:120430	MENDELIAN
PAX6	Aniridia	OMIM:106210	MENDELIAN
PAX6	Keratitis, hereditary	OMIM:148190	MENDELIAN
PAX6	Isolated aniridia	ORPHA:250923	UNKNOWN
PAX6	WAGR syndrome	ORPHA:893	UNKNOWN
PAX6	Aniridia-cerebellar ataxia-intellectual disability syndrome	ORPHA:1065	UNKNOWN
PAX6	Autosomal dominant keratitis	ORPHA:2334	UNKNOWN
PAX6	Peters anomaly	ORPHA:708	UNKNOWN
PAX6	Morning glory disc anomaly	ORPHA:35737	UNKNOWN
PAX6	Foveal hypoplasia-presenile cataract syndrome	ORPHA:2253	UNKNOWN
PAX7	Myopathy, congenital, progressive, with scoliosis	OMIM:618578	MENDELIAN
PAX7	Rhabdomyosarcoma 2, alveolar	OMIM:268220	MENDELIAN
PAX8	Hypothyroidism, congenital, nongoitrous, 2	OMIM:218700	MENDELIAN
PAX8	Thyroid hypoplasia	ORPHA:95720	UNKNOWN
PAX8	Athyreosis	ORPHA:95713	UNKNOWN
PAX8	Thyroid ectopia	ORPHA:95712	UNKNOWN
PAX9	Tooth agenesis, selective, 3	OMIM:604625	MENDELIAN
PAX9	Oligodontia	ORPHA:99798	UNKNOWN
PAXIP1	—		UNKNOWN
PBRM1	Renal cell carcinoma, nonpapillary	OMIM:144700	MENDELIAN
PBX1	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay	OMIM:617641	MENDELIAN
PBX1	Renal hypoplasia, bilateral	ORPHA:97362	UNKNOWN
PBX3	—		UNKNOWN
PBX4	—		UNKNOWN
PC	Pyruvate carboxylase deficiency	OMIM:266150	MENDELIAN
PCBD1	Hyperphenylalaninemia, BH4-deficient, D	OMIM:264070	MENDELIAN
PCBD1	Pterin-4 alpha-carbinolamine dehydratase deficiency	ORPHA:1578	UNKNOWN
PCBP3	—		UNKNOWN
PCCA	Propionic acidemia	OMIM:606054	MENDELIAN
PCCA	Propionic acidemia	ORPHA:35	UNKNOWN
PCCB	Propionic acidemia	OMIM:606054	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
PCCB	Propionic acidemia	ORPHA:35	UNKNOWN
PCDH11X	—		UNKNOWN
PCDH15	Usher syndrome, type ID	OMIM:601067	MENDELIAN
PCDH15	Deafness, autosomal recessive 23	OMIM:609533	MENDELIAN
PCDH15	Usher syndrome, type IF	OMIM:602083	MENDELIAN
PCDH15	Usher syndrome type 1	ORPHA:231169	UNKNOWN
PCDH18	—		UNKNOWN
PCDH19	Epileptic encephalopathy, early infantile, 9	OMIM:300088	MENDELIAN
PCDH19	PCDH19 clustering epilepsy	ORPHA:714652	UNKNOWN
PCDH19	Dravet syndrome	ORPHA:33069	UNKNOWN
PCDH9	—		UNKNOWN
PCDHA1	—		UNKNOWN
PCDHA10	—		UNKNOWN
PCDHA13	—		UNKNOWN
PCDHA3	—		UNKNOWN
PCDHB16	—		UNKNOWN
PCDHB3	—		UNKNOWN
PCDHB4	—		UNKNOWN
PCDHGB4	—		UNKNOWN
PCGF2	Turnpenny-Fry syndrome	OMIM:618371	MENDELIAN
PCK1	Phosphoenolpyruvate carboxykinase deficiency, cytosolic	OMIM:261680	MENDELIAN
PCK1	Phosphoenolpyruvate carboxykinase deficiency	ORPHA:2880	UNKNOWN
PCK2	Phosphoenolpyruvate carboxykinase 2, mitochondrial	OMIM:261650	MENDELIAN
PCK2	Phosphoenolpyruvate carboxykinase deficiency	ORPHA:2880	UNKNOWN
PCLO	Pontocerebellar hypoplasia, type 3	OMIM:608027	MENDELIAN
PCMT1	—		UNKNOWN
PCNA	Ataxia-telangiectasia-like disorder 2	OMIM:615919	MENDELIAN
PCNA	PCNA-related progressive neurodegenerative photosensitivity syndrome	ORPHA:438134	UNKNOWN
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	OMIM:210720	MENDELIAN
PCNT	Microcephalic osteodysplastic primordial dwarfism type II	ORPHA:2637	UNKNOWN
PCNT	Seckel syndrome	ORPHA:808	UNKNOWN
PCOLCE	—		UNKNOWN
PCP4	—		UNKNOWN
PCSK1	Proprotein convertase 1/3 deficiency	OMIM:600955	MENDELIAN
PCSK1	Obesity due to prohormone convertase I deficiency	ORPHA:71528	UNKNOWN
PCSK2	—		UNKNOWN
PCSK5	—		UNKNOWN
PCSK7	—		UNKNOWN
PCSK9	Hypercholesterolemia, familial, 3	OMIM:603776	MENDELIAN
PCSK9	Homozygous familial hypercholesterolemia	ORPHA:391665	UNKNOWN
PCYT1A	Lipodystrophy, congenital generalized, type 5	OMIM:620680	MENDELIAN
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy	OMIM:608940	MENDELIAN
PCYT1A	Leber congenital amaurosis	ORPHA:65	UNKNOWN
PCYT1A	Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome	ORPHA:85167	UNKNOWN
PDAP1	—		UNKNOWN
PDCD1	Autoimmune disease with susceptibility to mycobacterium tuberculosis	OMIM:621004	MENDELIAN
PDCD1	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
PDCD10	Cerebral cavernous malformations 3	OMIM:603285	MENDELIAN
PDCD10	Cerebral cavernous malformations 1	OMIM:116860	MENDELIAN
PDCD10	Familial cerebral cavernous malformation	ORPHA:221061	UNKNOWN
PDCD1LG2	—		UNKNOWN
PDCD5	—		UNKNOWN
PDE10A	Striatal degeneration, autosomal dominant 2	OMIM:616922	MENDELIAN
PDE10A	Dyskinesia, limb and orofacial, infantile-onset	OMIM:616921	MENDELIAN
PDE10A	Infantile-onset generalized dyskinesia with orofacial involvement	ORPHA:494526	UNKNOWN
PDE10A	Childhood-onset benign chorea with striatal involvement	ORPHA:494541	UNKNOWN
PDE11A	Pigmented nodular adrenocortical disease, primary, 2	OMIM:610475	MENDELIAN
PDE11A	Carney complex	ORPHA:1359	UNKNOWN
PDE1A	—		UNKNOWN
PDE1C	Deafness, autosomal dominant 74	OMIM:618140	MENDELIAN
PDE4B	—		UNKNOWN
PDE4D	Acrodysostosis 2 with or without hormone resistance	OMIM:614613	MENDELIAN
PDE4D	PDE4D haploinsufficiency syndrome	ORPHA:439822	UNKNOWN
PDE4D	Acrodysostosis	ORPHA:950	UNKNOWN
PDE6A	Retinitis pigmentosa 43	OMIM:613810	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
PDE6A	Retinitis pigmentosa	ORPHA:791	UNKNOWN
PDE6B	Night blindness, congenital stationary, autosomal dominant 2	OMIM:163500	MENDELIAN
PDE6B	Retinitis pigmentosa 40	OMIM:613801	MENDELIAN
PDE6B	Retinitis pigmentosa	ORPHA:791	UNKNOWN
PDE6C	Cone dystrophy 4	OMIM:613093	MENDELIAN
PDE6C	Progressive cone dystrophy	ORPHA:1871	UNKNOWN
PDE6C	Achromatopsia	ORPHA:49382	UNKNOWN
PDE6D	Joubert syndrome 22	OMIM:615665	MENDELIAN
PDE6D	Orofaciodigital syndrome type 6	ORPHA:2754	UNKNOWN
PDE6D	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
PDE6G	Retinitis pigmentosa	OMIM:268000	MENDELIAN
PDE6G	Retinitis pigmentosa 57	OMIM:613582	MENDELIAN
PDE6G	Retinitis pigmentosa	ORPHA:791	UNKNOWN
PDE6H	Retinal cone dystrophy 3A	OMIM:610024	MENDELIAN
PDE6H	Achromatopsia	ORPHA:49382	UNKNOWN
PDE7B	—		UNKNOWN
PDE8B	Striatal degeneration, autosomal dominant	OMIM:609161	MENDELIAN
PDE8B	Pigmented nodular adrenocortical disease, primary, 3	OMIM:614190	MENDELIAN
PDE8B	Autosomal dominant striatal neurodegeneration	ORPHA:228169	UNKNOWN
PDGFB	Meningioma, familial, susceptibility to	OMIM:607174	MENDELIAN
PDGFB	Basal ganglia calcification, idiopathic, 1	OMIM:213600	MENDELIAN
PDGFB	Basal ganglia calcification, idiopathic, 5	OMIM:615483	MENDELIAN
PDGFB	Dermatofibrosarcoma protuberans	ORPHA:31112	UNKNOWN
PDGFB	Bilateral striopallidodentate calcinosis	ORPHA:1980	UNKNOWN
PDGFB	Meningioma	ORPHA:2495	UNKNOWN
PDGFC	—		UNKNOWN
PDGFRA	Hypereosinophilic syndrome, idiopathic	OMIM:607685	MENDELIAN
PDGFRA	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial	OMIM:175510	MENDELIAN
PDGFRA	Cleft lip/palate	ORPHA:199306	UNKNOWN
PDGFRA	Gastrointestinal stromal tumor	ORPHA:44890	UNKNOWN
PDGFRB	Kosaki overgrowth syndrome	OMIM:616592	MENDELIAN
PDGFRB	Ocular pterygium-digital keloid dysplasia syndrome	OMIM:621091	MENDELIAN
PDGFRB	Basal ganglia calcification, idiopathic, 4	OMIM:615007	MENDELIAN
PDGFRB	Basal ganglia calcification, idiopathic, 1	OMIM:213600	MENDELIAN
PDGFRB	Myofibromatosis, infantile, 1	OMIM:228550	MENDELIAN
PDGFRB	Premature aging syndrome, Penttinen type	OMIM:601812	MENDELIAN
PDGFRB	Infantile myofibromatosis	ORPHA:2591	UNKNOWN
PDGFRB	Bilateral striopallidodentate calcinosis	ORPHA:1980	UNKNOWN
PDGFRL	Colorectal cancer	OMIM:114500	MENDELIAN
PDGFRL	Hepatocellular carcinoma	OMIM:114550	MENDELIAN
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	OMIM:312170	MENDELIAN
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	ORPHA:79243	UNKNOWN
PDHA2	Spermatogenic failure 70	OMIM:619828	MENDELIAN
PDHA2	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
PDHB	Pyruvate dehydrogenase E1-beta deficiency	OMIM:614111	MENDELIAN
PDHB	Pyruvate dehydrogenase E1-beta deficiency	ORPHA:255138	UNKNOWN
PDHX	Pyruvate dehydrogenase e3-binding protein deficiency	OMIM:245349	MENDELIAN
PDHX	Pyruvate dehydrogenase E3-binding protein deficiency	ORPHA:255182	UNKNOWN
PDIA4	—		UNKNOWN
PDK1	—		UNKNOWN
PDK2	—		UNKNOWN
PDK3	Charcot-Marie-Tooth disease, X-linked dominant, 6	OMIM:300905	MENDELIAN
PDK3	X-linked Charcot-Marie-Tooth disease type 6	ORPHA:352675	UNKNOWN
PDK4	—		UNKNOWN
PDLIM3	—		UNKNOWN
PDLIM4	Osteoporosis	OMIM:166710	POLYGENIC
PDLIM5	—		UNKNOWN
PDP1	Pyruvate dehydrogenase phosphatase deficiency	OMIM:608782	MENDELIAN
PDP1	Pyruvate dehydrogenase phosphatase deficiency	ORPHA:79246	UNKNOWN
PDPK1	—		UNKNOWN
PDPR	—		UNKNOWN
PDSS1	Coenzyme Q10 deficiency, primary, 2	OMIM:614651	MENDELIAN
PDSS2	Coenzyme Q10 deficiency, primary, 3	OMIM:614652	MENDELIAN
PDX1	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
PDX1	Pancreatic agenesis, congenital	OMIM:260370	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
PDX1	Maturity-onset diabetes of the young, type 4	OMIM:606392	MENDELIAN
PDX1	Isolated permanent neonatal diabetes mellitus	ORPHA:99885	UNKNOWN
PDX1	MODY	ORPHA:552	UNKNOWN
PDXK	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy	OMIM:618511	MENDELIAN
PDYN	Spinocerebellar ataxia 23	OMIM:610245	MENDELIAN
PDYN	Spinocerebellar ataxia type 23	ORPHA:101108	UNKNOWN
PDZD7	Usher syndrome, type IIA	OMIM:276901	POLYGENIC
PDZD7	Deafness, autosomal recessive 57	OMIM:618003	MENDELIAN
PDZD7	Usher syndrome, type IIC	OMIM:605472	MENDELIAN
PDZD7	Usher syndrome type 2	ORPHA:231178	UNKNOWN
PDZD8	Intellectual developmental disorder with autism and dysmorphic facies	OMIM:620021	MENDELIAN
PEAR1	—		UNKNOWN
PECAM1	—		UNKNOWN
PECR	—		UNKNOWN
PEMT	—		UNKNOWN
PENK	—		UNKNOWN
PEPD	Prolidase deficiency	OMIM:170100	MENDELIAN
PEPD	Prolidase deficiency	ORPHA:742	UNKNOWN
PER1	—		UNKNOWN
PER2	Advanced sleep phase syndrome, familial, 1	OMIM:604348	MENDELIAN
PER3	Advanced sleep phase syndrome, familial, 3	OMIM:616882	MENDELIAN
PET100	Mitochondrial complex IV deficiency, nuclear type 12	OMIM:619055	MENDELIAN
PEX1	Peroxisome biogenesis disorder 1A (Zellweger)	OMIM:214100	MENDELIAN
PEX1	Heimler syndrome 1	OMIM:234580	MENDELIAN
PEX1	Peroxisome biogenesis disorder 1B	OMIM:601539	MENDELIAN
PEX1	Deafness-enamel hypoplasia-nail defects syndrome	ORPHA:3220	UNKNOWN
PEX1	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX1	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX1	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX10	Peroxisome biogenesis disorder 6A (Zellweger)	OMIM:614870	MENDELIAN
PEX10	Peroxisome biogenesis disorder 6B	OMIM:614871	MENDELIAN
PEX10	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX10	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX10	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX10	Autosomal recessive ataxia due to PEX10 deficiency	ORPHA:247815	UNKNOWN
PEX11B	Peroxisome biogenesis disorder 14B	OMIM:614920	MENDELIAN
PEX11B	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX11B	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX11B	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX12	Peroxisome biogenesis disorder 3B	OMIM:266510	MENDELIAN
PEX12	Peroxisome biogenesis disorder 3A (Zellweger)	OMIM:614859	MENDELIAN
PEX12	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX12	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX12	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX13	Peroxisome biogenesis disorder 11B	OMIM:614885	MENDELIAN
PEX13	Peroxisome biogenesis disorder 11A (Zellweger)	OMIM:614883	MENDELIAN
PEX13	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX13	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX13	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX14	Peroxisome biogenesis disorder 13A (Zellweger)	OMIM:614887	MENDELIAN
PEX14	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX14	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX14	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX16	Peroxisome biogenesis disorder 8B	OMIM:614877	MENDELIAN
PEX16	Peroxisome biogenesis disorder 8A (Zellweger)	OMIM:614876	MENDELIAN
PEX16	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX16	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX16	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX19	Peroxisome biogenesis disorder 12A (Zellweger)	OMIM:614886	MENDELIAN
PEX19	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX19	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX19	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)	OMIM:614866	MENDELIAN
PEX2	Peroxisome biogenesis disorder 5B	OMIM:614867	MENDELIAN
PEX2	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
PEX2	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX2	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX26	Peroxisome biogenesis disorder 7B	OMIM:614873	MENDELIAN
PEX26	Peroxisome biogenesis disorder 7A (zellweger)	OMIM:614872	MENDELIAN
PEX26	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX26	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX26	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX3	Peroxisome biogenesis disorder 10B	OMIM:617370	MENDELIAN
PEX3	Peroxisome biogenesis disorder 10A (Zellweger)	OMIM:614882	MENDELIAN
PEX3	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX3	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX3	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX5	Rhizomelic chondrodysplasia punctata, type 5	OMIM:616716	MENDELIAN
PEX5	Peroxisome biogenesis disorder 2A (Zellweger)	OMIM:214110	MENDELIAN
PEX5	Peroxisome biogenesis disorder 2B	OMIM:202370	MENDELIAN
PEX5	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX5	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX5	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX6	Heimler syndrome 2	OMIM:616617	MENDELIAN
PEX6	Peroxisome biogenesis disorder 4A (Zellweger)	OMIM:614862	MENDELIAN
PEX6	Peroxisome biogenesis disorder 4B	OMIM:614863	MENDELIAN
PEX6	Autosomal recessive spinocerebellar ataxia-blindness-deafness syndrome	ORPHA:95433	UNKNOWN
PEX6	Deafness-enamel hypoplasia-nail defects syndrome	ORPHA:3220	UNKNOWN
PEX6	Neonatal adrenoleukodystrophy	ORPHA:44	UNKNOWN
PEX6	Zellweger syndrome	ORPHA:912	UNKNOWN
PEX6	Infantile Refsum disease	ORPHA:772	UNKNOWN
PEX7	Rhizomelic chondrodysplasia punctata, type 1	OMIM:215100	MENDELIAN
PEX7	Refsum disease	OMIM:266500	MENDELIAN
PEX7	Peroxisome biogenesis disorder 9B	OMIM:614879	MENDELIAN
PEX7	Adult Refsum disease	ORPHA:773	UNKNOWN
PFKFB1	—		UNKNOWN
PFKL	—		UNKNOWN
PFKM	Glycogen storage disease VII	OMIM:232800	MENDELIAN
PFKM	Glycogen storage disease due to muscle phosphofructokinase deficiency	ORPHA:371	UNKNOWN
PFN1	Amyotrophic lateral sclerosis 18	OMIM:614808	MENDELIAN
PFN1	Paget disease of bone 7, early-onset	OMIM:621600	MENDELIAN
PFN1	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
PGAM1	—		UNKNOWN
PGAM2	Phosphoglycerate mutase, muscle, deficiency of	OMIM:261670	MENDELIAN
PGAM2	Glycogen storage disease due to phosphoglycerate mutase deficiency	ORPHA:97234	UNKNOWN
PGAM5	—		UNKNOWN
PGAP1	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities	OMIM:615802	MENDELIAN
PGAP1	Autosomal recessive spastic paraplegia type 67	ORPHA:401820	UNKNOWN
PGAP2	Hyperphosphatasia with impaired intellectual development syndrome 3	OMIM:614207	MENDELIAN
PGAP2	Hyperphosphatasia-intellectual disability syndrome	ORPHA:247262	UNKNOWN
PGAP3	Hyperphosphatasia with impaired intellectual development syndrome 4	OMIM:615716	MENDELIAN
PGAP3	Hyperphosphatasia-intellectual disability syndrome	ORPHA:247262	UNKNOWN
PGBD1	—		UNKNOWN
PGBD3	—		UNKNOWN
PGBD4	—		UNKNOWN
PGC	—		UNKNOWN
PGD	—		UNKNOWN
PGK1	Phosphoglycerate kinase 1 deficiency	OMIM:300653	MENDELIAN
PGK1	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency	ORPHA:713	UNKNOWN
PGM1	Congenital disorder of glycosylation, type It	OMIM:614921	MENDELIAN
PGM3	Immunodeficiency 23	OMIM:615816	MENDELIAN
PGM3	PGM3-CDG	ORPHA:443811	UNKNOWN
PGR	Progesterone resistance	OMIM:264080	MENDELIAN
PHACTR1	Developmental and epileptic encephalopathy 70	OMIM:618298	MENDELIAN
PHACTR1	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
PHB	—		UNKNOWN
PHB2	—		UNKNOWN
PHC1	Microcephaly 11, primary, autosomal recessive	OMIM:615414	MENDELIAN
PHC1	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
PHEX	Hypophosphatemic rickets, X-linked dominant	OMIM:307800	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
PHEX	X-linked hypophosphatemia	ORPHA:89936	UNKNOWN
PHF11	—		UNKNOWN
PHF12	—		UNKNOWN
PHF2	—		UNKNOWN
PHF21A	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures	OMIM:618725	MENDELIAN
PHF21A	Potocki-Shaffer syndrome	ORPHA:52022	UNKNOWN
PHF3	—		UNKNOWN
PHF6	Borjeson-Forssman-Lehmann syndrome	OMIM:301900	MENDELIAN
PHF6	Borjeson-Forssman-Lehmann syndrome	ORPHA:127	UNKNOWN
PHF8	Intellectual developmental disorder, X-linked syndromic, Siderius type	OMIM:300263	MENDELIAN
PHF8	X-linked intellectual disability, Siderius type	ORPHA:85287	UNKNOWN
PHGDH	Neu-Laxova syndrome 1	OMIM:256520	MENDELIAN
PHGDH	Phosphoglycerate dehydrogenase deficiency	OMIM:601815	MENDELIAN
PHGDH	3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form	ORPHA:79351	UNKNOWN
PHIP	Developmental delay, intellectual disability, obesity, and dysmorphic features	OMIM:617991	MENDELIAN
PHIP	PHIP-related behavioral problems-intellectual disability-obesity-dysmorphic features syndrome	ORPHA:589905	UNKNOWN
PHKA1	Muscle glycogenosis, X-linked	OMIM:300559	MENDELIAN
PHKA1	Glycogen storage disease due to muscle phosphorylase kinase deficiency	ORPHA:715	UNKNOWN
PHKA2	Glycogen storage disease, type IXa1	OMIM:306000	MENDELIAN
PHKA2	Glycogen storage disease due to liver phosphorylase kinase deficiency	ORPHA:264580	UNKNOWN
PHKB	Glycogen storage disease type IXb	OMIM:261750	MENDELIAN
PHKB	Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency	ORPHA:79240	UNKNOWN
PHKG2	Glycogen storage disease IXc	OMIM:613027	MENDELIAN
PHKG2	Glycogen storage disease due to liver phosphorylase kinase deficiency	ORPHA:264580	UNKNOWN
PHLDA2	—		UNKNOWN
PHLPP2	—		UNKNOWN
PHOX2A	Fibrosis of extraocular muscles, congenital, 2	OMIM:602078	MENDELIAN
PHOX2A	Congenital fibrosis of extraocular muscles	ORPHA:45358	UNKNOWN
PHOX2B	Neuroblastoma, susceptibility to, 2	OMIM:613013	MENDELIAN
PHOX2B	Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease	OMIM:209880	MENDELIAN
PHOX2B	Haddad syndrome	ORPHA:99803	UNKNOWN
PHOX2B	Congenital central hypoventilation syndrome	ORPHA:661	UNKNOWN
PHOX2B	Neuroblastoma	ORPHA:635	UNKNOWN
PHOX2B	Hirschsprung disease-ganglioneuroblastoma syndrome	ORPHA:2151	UNKNOWN
PHYH	Refsum disease	OMIM:266500	MENDELIAN
PHYH	Adult Refsum disease	ORPHA:773	UNKNOWN
PHYKPL	Phosphohydroxylysineuria	OMIM:615011	MENDELIAN
PI3	—		UNKNOWN
PI4KA	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis	OMIM:616531	MENDELIAN
PI4KA	Spastic paraplegia 84, autosomal recessive	OMIM:619621	MENDELIAN
PI4KA	Gastrointestinal defects and immunodeficiency syndrome 2	OMIM:619708	MENDELIAN
PI4KA	Bilateral perisylvian polymicrogyria	ORPHA:98889	UNKNOWN
PI4KA	Combined immunodeficiency-multiple intestinal atresia	ORPHA:436252	UNKNOWN
PIAS1	—		UNKNOWN
PIBF1	Joubert syndrome 33	OMIM:617767	MENDELIAN
PIBF1	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
PICALM	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
PID1	—		UNKNOWN
PIEZO1	Lymphedema, hereditary, III	OMIM:616843	MENDELIAN
PIEZO1	Dehydrated hereditary stomatocytosis 1 with or without pseudohyperkalemia and/or perinatal edema	OMIM:194380	MENDELIAN
PIEZO1	Dehydrated hereditary stomatocytosis	ORPHA:3202	UNKNOWN
PIEZO2	Arthrogryposis, distal, type 5	OMIM:108145	MENDELIAN
PIEZO2	Arthrogryposis, distal, type 3	OMIM:114300	MENDELIAN
PIEZO2	Marden-Walker syndrome	OMIM:248700	MENDELIAN
PIEZO2	Arthrogryposis, distal, with impaired proprioception and touch	OMIM:617146	MENDELIAN
PIEZO2	Gordon syndrome	ORPHA:376	UNKNOWN
PIEZO2	Arthrogryposis-oculomotor limitation-electroretinal anomalies syndrome	ORPHA:1154	UNKNOWN
PIEZO2	Marden-Walker syndrome	ORPHA:2461	UNKNOWN
PIF1	—		UNKNOWN
PIGA	Neurodevelopmental disorder with epilepsy and hemochromatosis	OMIM:301072	MENDELIAN
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2	OMIM:300868	MENDELIAN
PIGA	Paroxysmal nocturnal hemoglobinuria	OMIM:300818	MENDELIAN
PIGA	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
PIGA	Epilepsy of infancy with migrating focal seizures	ORPHA:293181	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
PIGA	Paroxysmal nocturnal hemoglobinuria	ORPHA:447	UNKNOWN
PIGL	CHIME syndrome	OMIM:280000	MENDELIAN
PIGL	Hyperphosphatasia-intellectual disability syndrome	ORPHA:247262	UNKNOWN
PIGL	CHIME syndrome	ORPHA:3474	UNKNOWN
PIGM	Glycosylphosphatidylinositol deficiency	OMIM:610293	MENDELIAN
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1	OMIM:614080	MENDELIAN
PIGN	Fryns syndrome	ORPHA:2059	UNKNOWN
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome	ORPHA:280633	UNKNOWN
PIGO	Hyperphosphatasia with impaired intellectual development syndrome 2	OMIM:614749	MENDELIAN
PIGO	Hyperphosphatasia-intellectual disability syndrome	ORPHA:247262	UNKNOWN
PIGQ	Epileptic encephalopathy, early infantile, 77	OMIM:618548	MENDELIAN
PIGQ	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
PIGR	—		UNKNOWN
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3	OMIM:615398	MENDELIAN
PIGT	Paroxysmal nocturnal hemoglobinuria 2	OMIM:615399	MENDELIAN
PIGT	Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome	ORPHA:369837	UNKNOWN
PIGV	Hyperphosphatasia with impaired intellectual development syndrome 1	OMIM:239300	MENDELIAN
PIGV	Hyperphosphatasia-intellectual disability syndrome	ORPHA:247262	UNKNOWN
PIGW	Glycosylphosphatidylinositol biosynthesis defect 11	OMIM:616025	MENDELIAN
PIGW	Hyperphosphatasia-intellectual disability syndrome	ORPHA:247262	UNKNOWN
PIGZ	—		UNKNOWN
PIK3C2B	—		UNKNOWN
PIK3C2G	—		UNKNOWN
PIK3C3	—		UNKNOWN
PIK3CA	Epidermal nevus, somatic	OMIM:162900	MENDELIAN
PIK3CA	Congenital lipomatous overgrowth, vascular malformations, and epidermal nevi	OMIM:612918	MENDELIAN
PIK3CA	Cowden syndrome 5	OMIM:615108	MENDELIAN
PIK3CA	Capillary malformation of the lower lip, lymphatic malformation of face and neck, asymmetry of face and limbs, and partial/generalized overgrowth	OMIM:613089	MENDELIAN
PIK3CA	Keratosis, seborrheic, somatic	OMIM:182000	MENDELIAN
PIK3CA	Breast cancer	OMIM:114480	MENDELIAN
PIK3CA	Cerebral cavernous malformations 4	OMIM:619538	MENDELIAN
PIK3CA	Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic	OMIM:602501	MENDELIAN
PIK3CA	Colorectal cancer	OMIM:114500	MENDELIAN
PIK3CA	Ovarian cancer	OMIM:167000	MENDELIAN
PIK3CA	Hepatocellular carcinoma	OMIM:114550	MENDELIAN
PIK3CA	Hemifacial myohyperplasia, somatic	OMIM:606773	MENDELIAN
PIK3CA	Lung cancer, susceptibility to	OMIM:211980	MENDELIAN
PIK3CA	Gastric cancer, somatic	OMIM:613659	MENDELIAN
PIK3CA	Macroductyly, somatic	OMIM:155500	MENDELIAN
PIK3CA	Congenital infiltrating lipomatosis of the face	ORPHA:583097	UNKNOWN
PIK3CA	Hemimegalencephaly	ORPHA:99802	UNKNOWN
PIK3CA	CLAPO syndrome	ORPHA:168984	UNKNOWN
PIK3CA	Megalencephaly-capillary malformation-polymicrogyria syndrome	ORPHA:60040	UNKNOWN
PIK3CA	Hemihyperplasia-multiple lipomatosis syndrome	ORPHA:276280	UNKNOWN
PIK3CA	Lynch syndrome	ORPHA:144	UNKNOWN
PIK3CA	Capillary-lymphatic-venous malformation with segmental distribution	ORPHA:90308	UNKNOWN
PIK3CA	CLOVES syndrome	ORPHA:140944	UNKNOWN
PIK3CA	Meningioma	ORPHA:2495	UNKNOWN
PIK3CA	Segmental progressive overgrowth syndrome with fibroadipose hyperplasia	ORPHA:314662	UNKNOWN
PIK3CA	Familial cerebral cavernous malformation	ORPHA:221061	UNKNOWN
PIK3CB	—		UNKNOWN
PIK3CD	Immunodeficiency 14	OMIM:615513	MENDELIAN
PIK3CD	Roifman-Chitayat syndrome	OMIM:613328	MENDELIAN
PIK3CD	Immunodeficiency 14B, autosomal recessive	OMIM:619281	MENDELIAN
PIK3CD	Activated PI3K-delta syndrome 1	ORPHA:693661	UNKNOWN
PIK3CD	Combined immunodeficiency with facio-oculo-skeletal anomalies	ORPHA:221139	UNKNOWN
PIK3CD	Autosomal non-syndromic agammaglobulinemia	ORPHA:33110	UNKNOWN
PIK3CG	Immunodeficiency 97 with autoinflammation	OMIM:619802	MENDELIAN
PIK3R1	Agammaglobulinemia 7, autosomal recessive	OMIM:615214	MENDELIAN
PIK3R1	Short syndrome	OMIM:269880	MENDELIAN
PIK3R1	Immunodeficiency 36	OMIM:616005	MENDELIAN
PIK3R1	SHORT syndrome	ORPHA:3163	UNKNOWN
PIK3R1	Activated PI3K-delta syndrome 2	ORPHA:693681	UNKNOWN
PIK3R1	Autosomal non-syndromic agammaglobulinemia	ORPHA:33110	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
PIK3R2	Megalencephaly-Polymicrogyria-Polydactyly-Hydrocephalus syndrome	OMIM:603387	MENDELIAN
PIK3R2	Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome	ORPHA:83473	UNKNOWN
PIK3R3	—		UNKNOWN
PIK3R4	—		UNKNOWN
PIK3R5	Ataxia-Oculomotor apraxia 3	OMIM:615217	MENDELIAN
PIK3R5	Spinocerebellar ataxia with axonal neuropathy type 2	ORPHA:64753	UNKNOWN
PIKFYVE	Corneal fleck dystrophy	OMIM:121850	MENDELIAN
PIM1	—		UNKNOWN
PIN1	—		UNKNOWN
PINK1	Parkinson disease 6, autosomal recessive early-onset	OMIM:605909	MENDELIAN
PINK1	Young-onset Parkinson disease	ORPHA:2828	UNKNOWN
PIP5K1B	—		UNKNOWN
PIP5K1C	Lethal congenital contracture syndrome 3	OMIM:611369	MENDELIAN
PIR	—		UNKNOWN
PITPNA	—		UNKNOWN
PITPNM3	Cone-Rod dystrophy 5	OMIM:600977	MENDELIAN
PITPNM3	Cone rod dystrophy	ORPHA:1872	UNKNOWN
PITX1	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly	OMIM:119800	MENDELIAN
PITX1	Brachydactyly-elbow wrist dysplasia syndrome	ORPHA:1275	UNKNOWN
PITX2	Ring dermoid of cornea	OMIM:180550	MENDELIAN
PITX2	Axenfeld-Rieger syndrome, type 1	OMIM:180500	MENDELIAN
PITX2	Iridogoniodysgenesis, type 2	OMIM:137600	MENDELIAN
PITX2	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
PITX2	Axenfeld-Rieger syndrome	ORPHA:782	UNKNOWN
PITX2	Peters anomaly	ORPHA:708	UNKNOWN
PITX3	Cataract 11, multiple types	OMIM:610623	MENDELIAN
PITX3	Anterior segment dysgenesis 1	OMIM:107250	MENDELIAN
PIWIL1	—		UNKNOWN
PIWIL2	—		UNKNOWN
PIWIL3	—		UNKNOWN
PIWIL4	—		UNKNOWN
PJA1	—		UNKNOWN
PKD1	Polycystic kidneys	OMIM:173900	MENDELIAN
PKD1	Autosomal dominant polycystic kidney disease	ORPHA:730	UNKNOWN
PKD1L1	Heterotaxy, visceral, 8, autosomal	OMIM:617205	MENDELIAN
PKD2	Polycystic kidney disease 2	OMIM:613095	MENDELIAN
PKD2	Autosomal dominant polycystic kidney disease	ORPHA:730	UNKNOWN
PKHD1	Polycystic kidney disease 4 with or without polycystic liver disease	OMIM:263200	MENDELIAN
PKHD1	Caroli disease	ORPHA:53035	UNKNOWN
PKHD1	Autosomal recessive polycystic kidney disease	ORPHA:731	UNKNOWN
PKHD1L1	Deafness, autosomal recessive 124	OMIM:620794	MENDELIAN
PKLR	Pyruvate kinase deficiency of red cells	OMIM:266200	MENDELIAN
PKLR	Adenosine triphosphate, elevated, of erythrocytes	OMIM:102900	MENDELIAN
PKLR	Hemolytic anemia due to red cell pyruvate kinase deficiency	ORPHA:766	UNKNOWN
PKM	—		UNKNOWN
PKN3	—		UNKNOWN
PKNOX1	—		UNKNOWN
PKP1	Ectodermal dysplasia/skin fragility syndrome	OMIM:604536	MENDELIAN
PKP1	Ectodermal dysplasia-skin fragility syndrome	ORPHA:158668	UNKNOWN
PKP2	Arrhythmogenic right ventricular dysplasia, familial, 9	OMIM:609040	MENDELIAN
PKP2	Brugada syndrome	ORPHA:130	UNKNOWN
PLA2G10	—		UNKNOWN
PLA2G2A	Colorectal cancer	OMIM:114500	POLYGENIC
PLA2G2D	—		UNKNOWN
PLA2G4A	Gastrointestinal ulceration, recurrent, with dysfunctional platelets	OMIM:618372	MENDELIAN
PLA2G4C	—		UNKNOWN
PLA2G5	Fleck retina, familial benign	OMIM:228980	MENDELIAN
PLA2G6	Parkinson disease 14, autosomal recessive	OMIM:612953	MENDELIAN
PLA2G6	Neurodegeneration with brain iron accumulation 2B	OMIM:610217	MENDELIAN
PLA2G6	Neurodegeneration with brain iron accumulation 2A	OMIM:256600	MENDELIAN
PLA2G6	PLA2G6-related neurodegeneration, adult-onset	ORPHA:199351	UNKNOWN
PLA2G6	PLA2G6-related neurodegeneration, infantile-onset	ORPHA:35069	UNKNOWN
PLA2G7	Platelet-Activating factor acetylhydrolase deficiency	OMIM:614278	MENDELIAN
PLAG1	Silver-Russell syndrome 4	OMIM:618907	MENDELIAN
PLAG1	Salivary gland adenoma, pleomorphic	OMIM:181030	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
PLAG1	Silver-Russell syndrome due to a point mutation	ORPHA:397590	UNKNOWN
PLAGL1	Diabetes mellitus, transient neonatal, 1	OMIM:601410	POLYGENIC
PLAGL1	Transient neonatal diabetes mellitus	ORPHA:99886	UNKNOWN
PLAGL1	Paternal uniparental disomy of chromosome 6 syndrome	ORPHA:96191	UNKNOWN
PLAU	Alzheimer disease	OMIM:104300	POLYGENIC
PLAU	Quebec platelet disorder	OMIM:601709	MENDELIAN
PLAUR	—		UNKNOWN
PLCB1	Developmental and epileptic encephalopathy 12	OMIM:613722	MENDELIAN
PLCB1	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
PLCB1	Epilepsy of infancy with migrating focal seizures	ORPHA:293181	UNKNOWN
PLCB2	—		UNKNOWN
PLCB4	Auriculocondylar syndrome 2	OMIM:614669	MENDELIAN
PLCB4	Auriculocondylar syndrome 2B	OMIM:620458	MENDELIAN
PLCB4	Auriculocondylar syndrome	ORPHA:137888	UNKNOWN
PLCD1	Nail disorder, nonsyndromic congenital, 3	OMIM:151600	MENDELIAN
PLCD1	Leukonychia totalis	ORPHA:2387	UNKNOWN
PLCD4	—		UNKNOWN
PLCE1	Nephrotic syndrome, type 3	OMIM:610725	MENDELIAN
PLCE1	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
PLCG1	Immune dysregulation, autoimmunity, and autoinflammation	OMIM:620514	MENDELIAN
PLCG2	Familial cold autoinflammatory syndrome 3	OMIM:614468	MENDELIAN
PLCG2	Autoinflammation, antibody deficiency, and immune dysregulation, plc2-associated	OMIM:614878	MENDELIAN
PLCZ1	Spermatogenic failure 17	OMIM:617214	MENDELIAN
PLD2	—		UNKNOWN
PLD3	Spinocerebellar ataxia 46	OMIM:617770	MENDELIAN
PLD6	—		UNKNOWN
PLEC	Epidermolysis bullosa simplex with muscular dystrophy	OMIM:226670	MENDELIAN
PLEC	Epidermolysis bullosa simplex with nail dystrophy	OMIM:616487	MENDELIAN
PLEC	Epidermolysis bullosa, junctional, with pyloric atresia	OMIM:226730	MENDELIAN
PLEC	Epidermolysis bullosa simplex, Ogna type	OMIM:131950	MENDELIAN
PLEC	Epidermolysis bullosa simplex with pyloric atresia	OMIM:612138	MENDELIAN
PLEC	Muscular dystrophy, limb-girdle, type 2Q	OMIM:613723	MENDELIAN
PLEC	PLEC-related intermediate epidermolysis bullosa simplex without extracutaneous involvement	ORPHA:79401	UNKNOWN
PLEC	Epidermolysis bullosa simplex with muscular dystrophy	ORPHA:257	UNKNOWN
PLEC	Epidermolysis bullosa simplex with pyloric atresia	ORPHA:158684	UNKNOWN
PLEC	Plectin-related limb-girdle muscular dystrophy R17	ORPHA:254361	UNKNOWN
PLEC	Aplasia cutis congenita	ORPHA:1114	UNKNOWN
PLEKHA1	—		UNKNOWN
PLEKHG4	Spinocerebellar ataxia type 4	ORPHA:98765	UNKNOWN
PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate C	OMIM:615376	MENDELIAN
PLEKHG5	Spinal muscular atrophy, distal, autosomal recessive, 4	OMIM:611067	MENDELIAN
PLEKHM1	Osteopetrosis, autosomal recessive 6	OMIM:611497	MENDELIAN
PLEKHM1	Osteopetrosis, autosomal dominant 3	OMIM:618107	MENDELIAN
PLEKHM1	Intermediate osteopetrosis	ORPHA:210110	UNKNOWN
PLG	Plasminogen deficiency, type I	OMIM:217090	MENDELIAN
PLG	Angioedema, hereditary, 4	OMIM:619360	MENDELIAN
PLG	Hypoplasminogenemia	ORPHA:722	UNKNOWN
PLIN1	Lipodystrophy, familial partial, type 4	OMIM:613877	MENDELIAN
PLIN1	PLIN1-related familial partial lipodystrophy	ORPHA:280356	UNKNOWN
PLIN4	Vacuolar neuromyopathy	OMIM:601846	MENDELIAN
PLK1	—		UNKNOWN
PLK2	—		UNKNOWN
PLK4	Microcephaly and chorioretinopathy, autosomal recessive, 2	OMIM:616171	MENDELIAN
PLK4	Autosomal recessive chorioretinopathy-microcephaly syndrome	ORPHA:2518	UNKNOWN
PLK4	Seckel syndrome	ORPHA:808	UNKNOWN
PLN	Cardiomyopathy, dilated, 1P	OMIM:609909	MENDELIAN
PLN	Cardiomyopathy, familial hypertrophic, 18	OMIM:613874	MENDELIAN
PLN	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	OMIM:225400	MENDELIAN
PLOD1	Kyphoscoliotic Ehlers-Danlos syndrome due to lysyl hydroxylase 1 deficiency	ORPHA:1900	UNKNOWN
PLOD2	Bruck syndrome 2	OMIM:609220	MENDELIAN
PLOD2	Bruck syndrome	ORPHA:2771	UNKNOWN
PLOD3	Bone fragility with contractures, arterial rupture, and deafness	OMIM:612394	MENDELIAN
PLP1	Spastic paraplegia 2, X-linked	OMIM:312920	MENDELIAN
PLP1	Pelizaeus-Merzbacher disease	OMIM:312080	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
PLP1	Spastic paraplegia type 2	ORPHA:99015	UNKNOWN
PLP1	Pelizaeus-Merzbacher disease, connatal form	ORPHA:280210	UNKNOWN
PLP1	Pelizaeus-Merzbacher disease, transitional form	ORPHA:280224	UNKNOWN
PLP1	Pelizaeus-Merzbacher disease, classic form	ORPHA:280219	UNKNOWN
PLP1	Null syndrome	ORPHA:280234	UNKNOWN
PLP1	Pelizaeus-Merzbacher disease in female carriers	ORPHA:280229	UNKNOWN
PLP2	—		UNKNOWN
PLRG1	—		UNKNOWN
PLS3	Hernia, anterior diaphragmatic	OMIM:306950	MENDELIAN
PLSCR3	—		UNKNOWN
PLTP	—		UNKNOWN
PLXNA1	Dworschak-Punetha neurodevelopmental syndrome	OMIM:619955	MENDELIAN
PLXNA2	—		UNKNOWN
PLXNA3	—		UNKNOWN
PLXNA4	—		UNKNOWN
PLXNB1	—		UNKNOWN
PLXNB3	—		UNKNOWN
PLXND1	Congenital heart defects, multiple types, 9	OMIM:620294	MENDELIAN
PLXND1	Common arterial trunk	ORPHA:3384	UNKNOWN
PLXND1	Moebius syndrome	ORPHA:570	UNKNOWN
PMAIP1	—		UNKNOWN
PMFBP1	Spermatogenic failure 31	OMIM:618112	MENDELIAN
PMFBP1	Male infertility due to acephalic spermatozoa	ORPHA:529970	UNKNOWN
PML	Acute promyelocytic leukemia	ORPHA:520	UNKNOWN
PMM2	Congenital disorder of glycosylation, type Ia	OMIM:212065	MENDELIAN
PMM2	PMM2-CDG	ORPHA:79318	UNKNOWN
PMP22	Roussy-Levy hereditary areflexic dystasia	OMIM:180800	MENDELIAN
PMP22	Neuropathy, inflammatory demyelinating	OMIM:139393	MENDELIAN
PMP22	Charcot-Marie-Tooth disease, demyelinating, type 1A	OMIM:118220	MENDELIAN
PMP22	Neuropathy, hereditary, with liability to pressure palsies	OMIM:162500	MENDELIAN
PMP22	Charcot-Marie-Tooth disease and deafness	OMIM:118300	MENDELIAN
PMP22	Hypertrophic neuropathy of dejerine-sottas	OMIM:145900	MENDELIAN
PMP22	Charcot-Marie-Tooth disease type 1E	ORPHA:90658	UNKNOWN
PMP22	Charcot-Marie-Tooth disease type 1A	ORPHA:101081	UNKNOWN
PMP22	Roussy-L��vy syndrome	ORPHA:3115	UNKNOWN
PMP22	Acute inflammatory demyelinating polyradiculoneuropathy	ORPHA:98916	UNKNOWN
PMP22	Hereditary neuropathy with liability to pressure palsies	ORPHA:640	UNKNOWN
PMPCA	Spinocerebellar ataxia, autosomal recessive 2	OMIM:213200	MENDELIAN
PMPCA	Autosomal recessive cerebelloparenchymal disorder type 3	ORPHA:1170	UNKNOWN
PMS1	Lynch syndrome I	OMIM:120435	MENDELIAN
PMS1	Lynch syndrome	ORPHA:144	UNKNOWN
PMS2	Colorectal cancer, hereditary nonpolyposis, type 4	OMIM:614337	MENDELIAN
PMS2	Mismatch repair cancer syndrome 4	OMIM:619101	MENDELIAN
PMS2	Lynch syndrome	ORPHA:144	UNKNOWN
PMS2P3	—		UNKNOWN
PNKD	Paroxysmal nonkinesigenic dyskinesia 1	OMIM:118800	MENDELIAN
PNKD	Paroxysmal non-kinesigenic dyskinesia	ORPHA:98810	UNKNOWN
PNKP	Ataxia-oculomotor apraxia 4	OMIM:616267	MENDELIAN
PNKP	Charcot-Marie-Tooth disease, axonal, type 2B2	OMIM:605589	MENDELIAN
PNKP	Microcephaly, seizures, and developmental delay	OMIM:613402	MENDELIAN
PNKP	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
PNKP	Ataxia-oculomotor apraxia type 4	ORPHA:459033	UNKNOWN
PNLDC1	Spermatogenic failure 57	OMIM:619528	MENDELIAN
PNLDC1	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
PNLIP	Pancreatic lipase deficiency	OMIM:614338	MENDELIAN
PNMT	—		UNKNOWN
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency	OMIM:613179	MENDELIAN
PNP	Purine nucleoside phosphorylase deficiency	ORPHA:760	UNKNOWN
PNPLA1	Ichthyosis, congenital, autosomal recessive 10	OMIM:615024	MENDELIAN
PNPLA1	Congenital ichthyosiform erythroderma	ORPHA:79394	UNKNOWN
PNPLA2	Neutral lipid storage disease with myopathy	OMIM:610717	MENDELIAN
PNPLA2	Primary triglyceride deposit cardiomyovasculopathy	ORPHA:565612	UNKNOWN
PNPLA2	Neutral lipid storage disease with myopathy	ORPHA:98908	UNKNOWN
PNPLA3	—		UNKNOWN
PNPLA6	Laurence-Moon syndrome	OMIM:245800	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
PNPLA6	Spastic paraplegia 39, autosomal recessive	OMIM:612020	MENDELIAN
PNPLA6	Boucher-Neuhauser syndrome	OMIM:215470	MENDELIAN
PNPLA6	Oliver-McFarlane syndrome	OMIM:275400	MENDELIAN
PNPLA6	Trichomegaly-retina pigmentary degeneration-dwarfism syndrome	ORPHA:3363	UNKNOWN
PNPLA6	Autosomal recessive spastic paraplegia type 39	ORPHA:139480	UNKNOWN
PNPLA6	Laurence-Moon syndrome	ORPHA:2377	UNKNOWN
PNPLA6	Ataxia-hypogonadism-choroidal dystrophy syndrome	ORPHA:1180	UNKNOWN
PNPLA6	Cerebellar ataxia-hypogonadism syndrome	ORPHA:1173	UNKNOWN
PNPO	Pyridoxamine 5-prime-phosphate oxidase deficiency	OMIM:610090	MENDELIAN
PNPO	Pyridoxamine-5-phosphate deficiency-developmental and epileptic encephalopathy	ORPHA:79096	UNKNOWN
PNPT1	Spinocerebellar ataxia 25	OMIM:608703	MENDELIAN
PNPT1	Combined oxidative phosphorylation deficiency 13	OMIM:614932	MENDELIAN
PNPT1	Deafness, autosomal recessive 70	OMIM:614934	MENDELIAN
PNPT1	Combined oxidative phosphorylation defect type 13	ORPHA:319514	UNKNOWN
PNPT1	Spinocerebellar ataxia type 25	ORPHA:101111	UNKNOWN
PNRC1	—		UNKNOWN
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	OMIM:614813	MENDELIAN
POC1B	Cone-Rod dystrophy 20	OMIM:615973	MENDELIAN
POC1B	Cone rod dystrophy	ORPHA:1872	UNKNOWN
PODXL	Young-onset Parkinson disease	ORPHA:2828	UNKNOWN
PODXL	Atypical juvenile parkinsonism	ORPHA:391411	UNKNOWN
POF1B	Premature ovarian failure 2B	OMIM:300604	MENDELIAN
POFUT1	Dowling-Degos disease 2	OMIM:615327	MENDELIAN
POFUT1	Dowling-Degos disease	ORPHA:79145	UNKNOWN
POFUT2	—		UNKNOWN
POGLUT1	Dowling-Degos disease 4	OMIM:615696	MENDELIAN
POGLUT1	Muscular dystrophy, limb-girdle, autosomal recessive 21	OMIM:617232	MENDELIAN
POGLUT1	Dowling-Degos disease	ORPHA:79145	UNKNOWN
POGZ	White-Sutton syndrome	OMIM:616364	MENDELIAN
POGZ	White-Sutton syndrome	ORPHA:468678	UNKNOWN
POLA1	Pigmentary disorder, reticulate, with systemic manifestations, X-linked	OMIM:301220	MENDELIAN
POLA1	Van Esch-O'Driscoll syndrome	OMIM:301030	MENDELIAN
POLA1	X-linked intellectual disability, Van Esch type	ORPHA:163976	UNKNOWN
POLB	—		UNKNOWN
POLD1	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome	OMIM:615381	MENDELIAN
POLD1	Colorectal cancer, susceptibility to, 10	OMIM:612591	POLYGENIC
POLD1	Immunodeficiency 120	OMIM:620836	MENDELIAN
POLD1	Familial colorectal cancer Type X	ORPHA:440437	UNKNOWN
POLD1	Polymerase proofreading-related polyposis	ORPHA:447877	UNKNOWN
POLD3	Immunodeficiency 122	OMIM:620869	MENDELIAN
POLD4	—		UNKNOWN
POLE	Facial dysmorphism, immunodeficiency, livedo, and short stature	OMIM:615139	MENDELIAN
POLE	Colorectal cancer, susceptibility to, 12	OMIM:615083	POLYGENIC
POLE	Intrauterine growth retardation, metaphyseal dysplasia, adrenal hypoplasia congenita, genital anomalies, and immunodeficiency	OMIM:618336	MENDELIAN
POLE	Familial colorectal cancer Type X	ORPHA:440437	UNKNOWN
POLE	IMAGe syndrome	ORPHA:85173	UNKNOWN
POLE	Polymerase proofreading-related polyposis	ORPHA:447877	UNKNOWN
POLE2	—		UNKNOWN
POLE3	—		UNKNOWN
POLE4	—		UNKNOWN
POLG	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	OMIM:603041	MENDELIAN
POLG	Progressive external ophthalmoplegia with mitochondrial DNA deletions,autosomal recessive	OMIM:258450	MENDELIAN
POLG	Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis	OMIM:607459	MENDELIAN
POLG	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 1	OMIM:157640	MENDELIAN
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type)	OMIM:203700	MENDELIAN
POLG	Mitochondrial DNA depletion syndrome 4B (mngie type)	OMIM:613662	MENDELIAN
POLG	Recessive mitochondrial ataxia syndrome	ORPHA:94125	UNKNOWN
POLG	Mitochondrial neurogastrointestinal encephalomyopathy	ORPHA:298	UNKNOWN
POLG	Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome	ORPHA:70595	UNKNOWN
POLG	Alpers-Huttenlocher syndrome	ORPHA:726	UNKNOWN
POLG	Autosomal dominant progressive external ophthalmoplegia	ORPHA:254892	UNKNOWN
POLG	Spinocerebellar ataxia with epilepsy	ORPHA:254881	UNKNOWN
POLG	Autosomal recessive progressive external ophthalmoplegia	ORPHA:254886	UNKNOWN
POLG2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4	OMIM:610131	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
POLG2	Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type)	OMIM:619425	MENDELIAN
POLG2	Mitochondrial DNA depletion syndrome 16 (hepatic type)	OMIM:618528	MENDELIAN
POLG2	Autosomal dominant progressive external ophthalmoplegia	ORPHA:254892	UNKNOWN
POLH	Xeroderma pigmentosum, variant type	OMIM:278750	MENDELIAN
POLH	Xeroderma pigmentosum variant	ORPHA:90342	UNKNOWN
POLI	—		UNKNOWN
POLK	—		UNKNOWN
POLL	—		UNKNOWN
POLM	—		UNKNOWN
POLN	—		UNKNOWN
POLQ	—		UNKNOWN
POLR1C	Leukodystrophy, hypomyelinating, 11	OMIM:616494	MENDELIAN
POLR1C	Treacher Collins syndrome 3	OMIM:248390	MENDELIAN
POLR1C	Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome	ORPHA:88637	UNKNOWN
POLR1C	Treacher-Collins syndrome	ORPHA:861	UNKNOWN
POLR1D	Treacher collins syndrome 2	OMIM:613717	MENDELIAN
POLR1D	Treacher-Collins syndrome	ORPHA:861	UNKNOWN
POLR2A	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities	OMIM:618603	MENDELIAN
POLR2B	—		UNKNOWN
POLR2C	—		UNKNOWN
POLR2D	—		UNKNOWN
POLR2E	—		UNKNOWN
POLR2F	—		UNKNOWN
POLR2G	—		UNKNOWN
POLR2H	—		UNKNOWN
POLR2I	—		UNKNOWN
POLR2J	—		UNKNOWN
POLR2J3	—		UNKNOWN
POLR2K	—		UNKNOWN
POLR2L	—		UNKNOWN
POLR2M	—		UNKNOWN
POLR3A	Wiedemann-Rautenstrauch syndrome	OMIM:264090	MENDELIAN
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/orhypogonadotropic hypogonadism	OMIM:607694	MENDELIAN
POLR3A	Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome	ORPHA:88637	UNKNOWN
POLR3A	Wiedemann-Rautenstrauch syndrome	ORPHA:3455	UNKNOWN
POLR3A	Tremor-ataxia-central hypomyelination syndrome	ORPHA:447896	UNKNOWN
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	OMIM:614381	MENDELIAN
POLR3B	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/orhypogonadotropic hypogonadism	OMIM:607694	MENDELIAN
POLR3B	Charcot-Marie-Tooth disease, demyelinating, type 11	OMIM:619742	MENDELIAN
POLR3B	Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome	ORPHA:88637	UNKNOWN
POLRMT	Combined oxidative phosphorylation deficiency 55	OMIM:619743	MENDELIAN
POMC	Obesity, early-onset, with adrenal insufficiency and red hair	OMIM:609734	MENDELIAN
POMC	Obesity	OMIM:601665	POLYGENIC
POMC	Obesity due to pro-opiomelanocortin deficiency	ORPHA:71526	UNKNOWN
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	OMIM:253280	MENDELIAN
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3	OMIM:613151	MENDELIAN
POMGNT1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3	OMIM:613157	MENDELIAN
POMGNT1	Retinitis pigmentosa 76	OMIM:617123	MENDELIAN
POMGNT1	Congenital muscular dystrophy with cerebellar involvement	ORPHA:370959	UNKNOWN
POMGNT1	Walker-Warburg syndrome	ORPHA:899	UNKNOWN
POMGNT1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
POMGNT1	Muscle-eye-brain disease	ORPHA:588	UNKNOWN
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12	OMIM:615249	MENDELIAN
POMK	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12	OMIM:616094	MENDELIAN
POMK	Congenital muscular dystrophy with cerebellar involvement	ORPHA:370959	UNKNOWN
POMK	Walker-Warburg syndrome	ORPHA:899	UNKNOWN
POMP	Proteasome-Associated autoinflammatory syndrome 2	OMIM:618048	MENDELIAN
POMP	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	OMIM:601952	MENDELIAN
POMP	Keratosis linearis-ichthyosis congenita-sclerosing keratoderma syndrome	ORPHA:281201	UNKNOWN
POMT1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1	OMIM:609308	MENDELIAN
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	OMIM:236670	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1	OMIM:613155	MENDELIAN
POMT1	Congenital muscular dystrophy with intellectual disability	ORPHA:370968	UNKNOWN
POMT1	Congenital muscular dystrophy without intellectual disability	ORPHA:370980	UNKNOWN
POMT1	Congenital muscular dystrophy with cerebellar involvement	ORPHA:370959	UNKNOWN
POMT1	Walker-Warburg syndrome	ORPHA:899	UNKNOWN
POMT1	POMT1-related limb-girdle muscular dystrophy R11	ORPHA:86812	UNKNOWN
POMT1	Muscle-eye-brain disease	ORPHA:588	UNKNOWN
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	OMIM:236670	MENDELIAN
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	OMIM:613150	MENDELIAN
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2	OMIM:613156	MENDELIAN
POMT2	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	OMIM:613158	MENDELIAN
POMT2	Congenital muscular dystrophy with intellectual disability	ORPHA:370968	UNKNOWN
POMT2	Congenital muscular dystrophy with cerebellar involvement	ORPHA:370959	UNKNOWN
POMT2	Walker-Warburg syndrome	ORPHA:899	UNKNOWN
POMT2	POMT2-related limb-girdle muscular dystrophy R14	ORPHA:206559	UNKNOWN
POMT2	Muscle-eye-brain disease	ORPHA:588	UNKNOWN
PON1	—		UNKNOWN
PON2	—		UNKNOWN
PON3	—		UNKNOWN
POP1	Anauxetic dysplasia 2	OMIM:617396	MENDELIAN
POR	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis	OMIM:207410	MENDELIAN
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	OMIM:201750	MENDELIAN
POR	Disordered steroidogenesis due to cytochrome P450 oxidoreductase	OMIM:613571	MENDELIAN
POR	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	ORPHA:95699	UNKNOWN
PORCN	Focal dermal hypoplasia	OMIM:305600	MENDELIAN
PORCN	Focal dermal hypoplasia	ORPHA:2092	UNKNOWN
POSTN	—		UNKNOWN
POT1	Melanoma, cutaneous malignant, susceptibility to, 10	OMIM:615848	MENDELIAN
POT1	Cerebroretinal microangiopathy with calcifications and cysts 3	OMIM:620368	MENDELIAN
POT1	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8	OMIM:620367	MENDELIAN
POT1	Familial melanoma	ORPHA:618	UNKNOWN
POTEJ	—		UNKNOWN
POU1F1	Pituitary hormone deficiency, combined, 1	OMIM:613038	MENDELIAN
POU1F1	Combined pituitary hormone deficiencies, genetic forms	ORPHA:95494	UNKNOWN
POU1F1	Hypothyroidism due to deficient transcription factors involved in pituitary development or function	ORPHA:226307	UNKNOWN
POU2F1	—		UNKNOWN
POU3F3	Snijders Blok-Fisher syndrome	OMIM:618604	MENDELIAN
POU3F4	Deafness, X-linked 2	OMIM:304400	MENDELIAN
POU3F4	Xq21 microdeletion syndrome	ORPHA:1435	UNKNOWN
POU4F2	—		UNKNOWN
POU4F3	Deafness, autosomal dominant 15	OMIM:602459	MENDELIAN
POU5F1	—		UNKNOWN
POU5F1B	—		UNKNOWN
POU6F2	Wilms tumor 5	OMIM:601583	POLYGENIC
POU6F2	Nephroblastoma	ORPHA:654	UNKNOWN
PPARA	—		UNKNOWN
PPARD	—		UNKNOWN
PPARG	Diabetes mellitus, noninsulin-dependent	OMIM:125853	MENDELIAN
PPARG	Lipodystrophy, familial partial, type 3	OMIM:604367	MENDELIAN
PPARG	Obesity	OMIM:601665	MENDELIAN
PPARG	PPARG-related familial partial lipodystrophy	ORPHA:79083	UNKNOWN
PPARGC1A	—		UNKNOWN
PPARGC1B	—		UNKNOWN
PPAT	—		UNKNOWN
PPFIA3	Paul-Chao neurodevelopmental syndrome	OMIM:621122	MENDELIAN
PPIA	—		UNKNOWN
PPIB	Osteogenesis imperfecta, type IX	OMIM:259440	MENDELIAN
PPIB	Autosomal dominant optic atrophy, classic form	ORPHA:98673	UNKNOWN
PPIG	—		UNKNOWN
PPM1B	2p21 microdeletion syndrome	ORPHA:163693	UNKNOWN
PPM1D	Breast cancer	OMIM:114480	MENDELIAN
PPM1D	Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold	OMIM:617450	MENDELIAN
PPM1G	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
PPM1K	Maple syrup urine disease, mild variant	OMIM:615135	MENDELIAN
PPOX	Porphyria variegata	OMIM:176200	MENDELIAN
PPOX	Variegate porphyria, childhood-onset	OMIM:620483	MENDELIAN
PPOX	Variegate porphyria	ORPHA:79473	UNKNOWN
PPP1R12B	—		UNKNOWN
PPP1R15B	Microcephaly, short stature, and impaired glucose metabolism 2	OMIM:616817	MENDELIAN
PPP1R15B	Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome	ORPHA:391408	UNKNOWN
PPP1R17	Hypercholesterolemia, familial, 1	OMIM:143890	POLYGENIC
PPP1R1A	—		UNKNOWN
PPP1R3A	Diabetes mellitus, noninsulin-dependent	OMIM:125853	MENDELIAN
PPP1R3C	—		UNKNOWN
PPP1R8	—		UNKNOWN
PPP1R9B	—		UNKNOWN
PPP2CA	Neurodevelopmental disorder and language delay with or without structural brain abnormalities	OMIM:618354	MENDELIAN
PPP2R1A	Houge-Janssens syndrome 2	OMIM:616362	MENDELIAN
PPP2R1A	Houge-Janssens syndrome type 2	ORPHA:457284	UNKNOWN
PPP2R1B	Lung cancer, susceptibility to	OMIM:211980	MENDELIAN
PPP2R2A	—		UNKNOWN
PPP2R2B	Spinocerebellar ataxia 12	OMIM:604326	MENDELIAN
PPP2R2B	Spinocerebellar ataxia type 12	ORPHA:98762	UNKNOWN
PPP2R2C	—		UNKNOWN
PPP2R5B	—		UNKNOWN
PPP2R5C	Houge-Janssens syndrome 4	OMIM:621185	MENDELIAN
PPP2R5D	Houge-Janssens syndrome 1	OMIM:616355	MENDELIAN
PPP2R5D	Houge-Janssens syndrome type 1	ORPHA:457279	UNKNOWN
PPP3CA	Developmental and epileptic encephalopathy 91	OMIM:617711	MENDELIAN
PPP3CA	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development	OMIM:618265	MENDELIAN
PPP3CA	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
PPP3R1	—		UNKNOWN
PPP4C	—		UNKNOWN
PPP4R1	—		UNKNOWN
PPP4R2	—		UNKNOWN
PPP4R4	—		UNKNOWN
PPP5C	—		UNKNOWN
PPP6C	—		UNKNOWN
PPP6R1	—		UNKNOWN
PPP6R2	—		UNKNOWN
PPP6R3	—		UNKNOWN
PPT1	Ceroid lipofuscinosis, neuronal, 1	OMIM:256730	MENDELIAN
PQBP1	Renpenning syndrome	OMIM:309500	MENDELIAN
PQBP1	X-linked intellectual disability, Sutherland-Haan type	ORPHA:93950	UNKNOWN
PQBP1	X-linked intellectual disability, Porteous type	ORPHA:93945	UNKNOWN
PQBP1	Hamel cerebro-palato-cardiac syndrome	ORPHA:93946	UNKNOWN
PQBP1	X-linked intellectual disability, Golabi-Ito-Hall type	ORPHA:93947	UNKNOWN
PRAF2	—		UNKNOWN
PRAM1	—		UNKNOWN
PRAMEF2	—		UNKNOWN
PRB1	—		UNKNOWN
PRB3	—		UNKNOWN
PRB4	—		UNKNOWN
PRCC	Renal cell carcinoma, papillary, 1, familial and somatic	OMIM:605074	MENDELIAN
PRCD	Retinitis pigmentosa 36	OMIM:610599	MENDELIAN
PRCD	Retinitis pigmentosa	ORPHA:791	UNKNOWN
PRCP	—		UNKNOWN
PRDM1	—		UNKNOWN
PRDM16	Left ventricular noncompaction 8	OMIM:615373	MENDELIAN
PRDM16	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
PRDM16	1p36 deletion syndrome	ORPHA:1606	UNKNOWN
PRDM2	—		UNKNOWN
PRDM5	Brittle cornea syndrome 2	OMIM:614170	MENDELIAN
PRDM5	Brittle cornea syndrome	ORPHA:90354	UNKNOWN
PRDM9	—		UNKNOWN
PRDX4	—		UNKNOWN
PREPL	Myasthenic syndrome, congenital, 22	OMIM:616224	MENDELIAN
PREPL	Hypotonia-cystinuria syndrome	ORPHA:163690	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
PREPL	2p21 microdeletion syndrome	ORPHA:163693	UNKNOWN
PREX2	—		UNKNOWN
PRF1	Aplastic anemia	OMIM:609135	MENDELIAN
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	OMIM:603553	MENDELIAN
PRF1	Lymphoma, non-hodgkin, familial	OMIM:605027	MENDELIAN
PRF1	Idiopathic aplastic anemia	ORPHA:88	UNKNOWN
PRF1	Familial hemophagocytic lymphohistiocytosis	ORPHA:540	UNKNOWN
PRG4	Camptodactyly-Arthropathy-Coxa vara-pericarditis syndrome	OMIM:208250	MENDELIAN
PRG4	Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome	ORPHA:2848	UNKNOWN
PRICKLE1	Epilepsy, progressive myoclonic 1B	OMIM:612437	MENDELIAN
PRICKLE1	Progressive myoclonic epilepsy type 1	ORPHA:308	UNKNOWN
PRIMPOL	Myopia 22, autosomal dominant	OMIM:615420	MENDELIAN
PRKAA2	—		UNKNOWN
PRKACA	Cardioacrofacial dysplasia 1	OMIM:619142	MENDELIAN
PRKACA	Pigmented nodular adrenocortical disease, primary, 4	OMIM:615830	MENDELIAN
PRKACA	Ellis-Van Creveld syndrome	ORPHA:289	UNKNOWN
PRKACG	Bleeding disorder, platelet-type, 19	OMIM:616176	MENDELIAN
PRKAG2	Cardiomyopathy, familial hypertrophic, 6	OMIM:600858	MENDELIAN
PRKAG2	Wolff-Parkinson-White syndrome	OMIM:194200	MENDELIAN
PRKAG2	Glycogen storage disease of heart, lethal congenital	OMIM:261740	MENDELIAN
PRKAR1A	Acrodysostosis 1, with or without hormone resistance	OMIM:101800	MENDELIAN
PRKAR1A	Carney complex, type 1	OMIM:160980	MENDELIAN
PRKAR1A	Pigmented nodular adrenocortical disease, primary, 1	OMIM:610489	MENDELIAN
PRKAR1A	Myxoma, intracardiac	OMIM:255960	MENDELIAN
PRKAR1A	Adrenocortical carcinoma	ORPHA:1501	UNKNOWN
PRKAR1A	Acrodysostosis	ORPHA:950	UNKNOWN
PRKAR1A	Carney complex	ORPHA:1359	UNKNOWN
PRKAR1A	Familial atrial myxoma	ORPHA:615	UNKNOWN
PRKAR1A	Acute promyelocytic leukemia	ORPHA:520	UNKNOWN
PRKCA	—		UNKNOWN
PRKCB	—		UNKNOWN
PRKCD	Autoimmune lymphoproliferative syndrome, type III	OMIM:615559	MENDELIAN
PRKCG	Spinocerebellar ataxia 14	OMIM:605361	MENDELIAN
PRKCG	Spinocerebellar ataxia type 14	ORPHA:98763	UNKNOWN
PRKCH	—		UNKNOWN
PRKCI	—		UNKNOWN
PRKCSH	Polycystic liver disease 1 with or without kidney cysts	OMIM:174050	MENDELIAN
PRKCSH	Isolated polycystic liver disease	ORPHA:2924	UNKNOWN
PRKD1	Congenital heart defects and ectodermal dysplasia	OMIM:617364	MENDELIAN
PRKD3	—		UNKNOWN
PRKDC	Immunodeficiency 26 with or without neurologic abnormalities	OMIM:615966	MENDELIAN
PRKG1	Aortic aneurysm, familial thoracic 8	OMIM:615436	MENDELIAN
PRKG1	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
PRKG2	Spondylometaphyseal dysplasia, Pagnamenta type	OMIM:619638	MENDELIAN
PRKG2	Acromesomelic dysplasia 4	OMIM:619636	MENDELIAN
PRKRA	Dystonia 16	OMIM:612067	MENDELIAN
PRKRA	Dystonia 16	ORPHA:210571	UNKNOWN
PRL	—		UNKNOWN
PRLH	—		UNKNOWN
PRLHR	—		UNKNOWN
PRLR	Hyperprolactinemia	OMIM:615555	MENDELIAN
PRLR	Multiple fibroadenomas of the breast	OMIM:615554	MENDELIAN
PRLR	Familial hyperprolactinemia	ORPHA:397685	UNKNOWN
PRM1	—		UNKNOWN
PRM2	—		UNKNOWN
PRMT10	—		UNKNOWN
PRMT2	—		UNKNOWN
PRMT3	—		UNKNOWN
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures	OMIM:617157	MENDELIAN
PRMT7	Short stature-brachydactyly-obesity-global developmental delay syndrome	ORPHA:464288	UNKNOWN
PRND	—		UNKNOWN
PRNP	Huntington disease-like 1	OMIM:603218	MENDELIAN
PRNP	Creutzfeldt-Jakob disease	OMIM:123400	MENDELIAN
PRNP	Fatal familial insomnia	OMIM:600072	MENDELIAN
PRNP	Kuru, susceptibility to	OMIM:245300	POLYGENIC

Gene_symbol	Disease_name	Disease_ID	Association_type
PRNP	Spongiform encephalopathy with neuropsychiatric features	OMIM:606688	MENDELIAN
PRNP	Gerstmann-Straussler disease	OMIM:137440	MENDELIAN
PRNP	Gerstmann-Straussler-Scheinker syndrome	ORPHA:356	UNKNOWN
PRNP	Familial Alzheimer-like prion disease	ORPHA:280397	UNKNOWN
PRNP	Inherited Creutzfeldt-Jakob disease	ORPHA:282166	UNKNOWN
PRNP	Huntington disease-like 1	ORPHA:157941	UNKNOWN
PRNP	Fatal familial insomnia	ORPHA:466	UNKNOWN
PROC	Thrombophilia, hereditary, due to protein C deficiency, autosomaldominant	OMIM:176860	MENDELIAN
PROC	Thrombophilia due to protein C deficiency, autosomal recessive	OMIM:612304	MENDELIAN
PROC	Severe hereditary thrombophilia due to congenital protein C deficiency	ORPHA:745	UNKNOWN
PROCR	—		UNKNOWN
PRODH	Schizophrenia 4	OMIM:600850	POLYGENIC
PRODH	Hyperprolinemia, type I	OMIM:239500	MENDELIAN
PRODH	Hyperprolinemia type 1	ORPHA:419	UNKNOWN
PROK1	—		UNKNOWN
PROK2	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
PROK2	Hypogonadotropic hypogonadism 4 with or without anosmia	OMIM:610628	MENDELIAN
PROK2	Kallmann syndrome	ORPHA:478	UNKNOWN
PROK2	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
PROKR1	—		UNKNOWN
PROKR2	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
PROKR2	Hypogonadotropic hypogonadism 3 with or without anosmia	OMIM:244200	MENDELIAN
PROKR2	Septo-optic dysplasia spectrum	ORPHA:3157	UNKNOWN
PROKR2	Pituitary stalk interruption syndrome	ORPHA:95496	UNKNOWN
PROKR2	Kallmann syndrome	ORPHA:478	UNKNOWN
PROKR2	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
PROM1	Macular dystrophy, retinal, 2	OMIM:608051	MENDELIAN
PROM1	Cone-rod dystrophy 12	OMIM:612657	MENDELIAN
PROM1	Retinitis pigmentosa 41	OMIM:612095	MENDELIAN
PROM1	Stargardt disease 4	OMIM:603786	MENDELIAN
PROM1	Cone rod dystrophy	ORPHA:1872	UNKNOWN
PROM1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
PROM1	Stargardt disease	ORPHA:827	UNKNOWN
PROP1	Pituitary hormone deficiency, combined, 2	OMIM:262600	MENDELIAN
PROP1	Non-acquired panhypopituitarism	ORPHA:90695	UNKNOWN
PROP1	Combined pituitary hormone deficiencies, genetic forms	ORPHA:95494	UNKNOWN
PROP1	Hypothyroidism due to deficient transcription factors involved in pituitary development or function	ORPHA:226307	UNKNOWN
PROS1	Thrombophilia due to protein S deficiency, autosomal recessive	OMIM:614514	MENDELIAN
PROS1	Thrombophilia, hereditary, due to protein S deficiency, autosomal	OMIM:612336	MENDELIAN
PROS1	Severe hereditary thrombophilia due to congenital protein S deficiency	ORPHA:743	UNKNOWN
PROX1	—		UNKNOWN
PROX2	—		UNKNOWN
PRPF19	—		UNKNOWN
PRPF3	Retinitis pigmentosa 18	OMIM:601414	MENDELIAN
PRPF3	Retinitis pigmentosa	ORPHA:791	UNKNOWN
PRPF31	Retinitis pigmentosa 11	OMIM:600138	MENDELIAN
PRPF31	Retinitis pigmentosa	ORPHA:791	UNKNOWN
PRPF39	—		UNKNOWN
PRPF4	Retinitis pigmentosa 70	OMIM:615922	MENDELIAN
PRPF4	Retinitis pigmentosa	ORPHA:791	UNKNOWN
PRPF6	Retinitis pigmentosa 60	OMIM:613983	MENDELIAN
PRPF6	Retinitis pigmentosa	ORPHA:791	UNKNOWN
PRPF8	Retinitis pigmentosa 13	OMIM:600059	MENDELIAN
PRPF8	Retinitis pigmentosa	ORPHA:791	UNKNOWN
PRPH	Amyotrophic lateral sclerosis 1	OMIM:105400	POLYGENIC
PRPH	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
PRPH2	Retinitis pigmentosa 7	OMIM:608133	MENDELIAN
PRPH2	Fundus albipunctatus	OMIM:136880	MENDELIAN
PRPH2	Macular dystrophy, patterned, 1	OMIM:169150	MENDELIAN
PRPH2	Macular dystrophy, vitelliform, 3	OMIM:608161	MENDELIAN
PRPH2	Choroidal dystrophy, central areolar 2	OMIM:613105	MENDELIAN
PRPH2	Adult-onset foveomacular vitelliform dystrophy	ORPHA:99000	UNKNOWN
PRPH2	Leber congenital amaurosis	ORPHA:65	UNKNOWN
PRPH2	Retinitis punctata albescens	ORPHA:52427	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
PRPH2	Central areolar choroidal dystrophy	ORPHA:75377	UNKNOWN
PRPH2	Cone rod dystrophy	ORPHA:1872	UNKNOWN
PRPH2	Retinitis pigmentosa	ORPHA:791	UNKNOWN
PRPH2	Stargardt disease	ORPHA:827	UNKNOWN
PRPH2	Severe early-childhood-onset retinal dystrophy	ORPHA:364055	UNKNOWN
PRPS1	Deafness, X-linked 1	OMIM:304500	MENDELIAN
PRPS1	Charcot-Marie-Tooth disease, X-linked recessive, 5	OMIM:311070	MENDELIAN
PRPS1	Arts syndrome	OMIM:301835	MENDELIAN
PRPS1	Phosphoribosylpyrophosphate synthetase superactivity	OMIM:300661	MENDELIAN
PRPS1	X-linked Charcot-Marie-Tooth disease type 5	ORPHA:99014	UNKNOWN
PRPS1	X-linked intellectual disability-limb spasticity-retinal dystrophy-arginine vasopressin deficiency	ORPHA:423479	UNKNOWN
PRPS1	Severe phosphoribosylpyrophosphate synthetase superactivity	ORPHA:411543	UNKNOWN
PRPS1	Mild phosphoribosylpyrophosphate synthetase superactivity	ORPHA:411536	UNKNOWN
PRPS1	Lethal ataxia with deafness and optic atrophy	ORPHA:1187	UNKNOWN
PRR12	Neuroocular syndrome	OMIM:619539	MENDELIAN
PRR14L	—		UNKNOWN
PRR16	—		UNKNOWN
PRRC2A	—		UNKNOWN
PRRT2	Episodic kinesigenic dyskinesia 1	OMIM:128200	MENDELIAN
PRRT2	Seizures, benign familial infantile, 2	OMIM:605751	MENDELIAN
PRRT2	Convulsions, familial infantile, with paroxysmal choreoathetosis	OMIM:602066	MENDELIAN
PRRT2	Self-limited infantile epilepsy	ORPHA:306	UNKNOWN
PRRT2	Genetic epilepsy with febrile seizure plus	ORPHA:36387	UNKNOWN
PRRT2	Paroxysmal exertion-induced dyskinesia	ORPHA:98811	UNKNOWN
PRRT2	Paroxysmal non-kinesigenic dyskinesia	ORPHA:98810	UNKNOWN
PRRT2	Paroxysmal kinesigenic dyskinesia	ORPHA:98809	UNKNOWN
PRRT2	Familial or sporadic hemiplegic migraine	ORPHA:569	UNKNOWN
PRRX1	Agnathia-Otocephaly complex	OMIM:202650	MENDELIAN
PRRX1	Agnathia-holoprosencephaly-situs inversus syndrome	ORPHA:990	UNKNOWN
PRSS1	Pancreatitis, hereditary	OMIM:167800	MENDELIAN
PRSS1	Autosomal dominant hereditary chronic pancreatitis	ORPHA:676	UNKNOWN
PRSS12	Intellectual developmental disorder, autosomal recessive 1	OMIM:249500	MENDELIAN
PRSS2	Pancreatitis, hereditary	OMIM:167800	POLYGENIC
PRSS2	Autosomal dominant hereditary chronic pancreatitis	ORPHA:676	UNKNOWN
PRSS3P2	—		UNKNOWN
PRSS56	Microphthalmia, isolated 6	OMIM:613517	MENDELIAN
PRSS56	Isolated complete colobomatous microphthalmia	ORPHA:35612	UNKNOWN
PRSS8	—		UNKNOWN
PRTG	—		UNKNOWN
PRX	Hypertrophic neuropathy of dejerine-sottas	OMIM:145900	MENDELIAN
PRX	Charcot-Marie-Tooth disease, demyelinating, type 4F	OMIM:614895	MENDELIAN
PRY	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
PRY2	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
PSAP	Combined saposin deficiency	OMIM:611721	MENDELIAN
PSAP	Krabbe disease, atypical, due to saposin A deficiency	OMIM:611722	MENDELIAN
PSAP	Metachromatic leukodystrophy due to saposin B deficiency	OMIM:249900	MENDELIAN
PSAP	Gaucher disease, atypical	OMIM:610539	MENDELIAN
PSAP	Parkinson disease 24, autosomal dominant, susceptibility to	OMIM:619491	POLYGENIC
PSAP	Encephalopathy due to prosaposin deficiency	ORPHA:139406	UNKNOWN
PSAP	Infantile Krabbe disease	ORPHA:206436	UNKNOWN
PSAP	Metachromatic leukodystrophy, adult form	ORPHA:309271	UNKNOWN
PSAP	Metachromatic leukodystrophy, juvenile form	ORPHA:309263	UNKNOWN
PSAP	Atypical Gaucher disease due to saposin C deficiency	ORPHA:309252	UNKNOWN
PSAP	Metachromatic leukodystrophy, late infantile form	ORPHA:309256	UNKNOWN
PSAT1	Phosphoserine aminotransferase deficiency	OMIM:610992	MENDELIAN
PSAT1	Neu-Laxova syndrome 2	OMIM:616038	MENDELIAN
PSAT1	Phosphoserine aminotransferase deficiency, infantile/juvenile form	ORPHA:284417	UNKNOWN
PSCA	—		UNKNOWN
PSEN1	Pick disease of brain	OMIM:172700	MENDELIAN
PSEN1	Acne inversa, familial, 3	OMIM:613737	MENDELIAN
PSEN1	Alzheimer disease 3	OMIM:607822	MENDELIAN
PSEN1	Cardiomyopathy, dilated, 1U	OMIM:613694	MENDELIAN
PSEN1	Frontotemporal dementia	OMIM:600274	MENDELIAN
PSEN1	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
PSEN1	Behavioral variant of frontotemporal dementia	ORPHA:275864	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
PSEN1	Early-onset autosomal dominant Alzheimer disease	ORPHA:1020	UNKNOWN
PSEN1	Progressive non-fluent aphasia	ORPHA:100070	UNKNOWN
PSEN1	Semantic dementia	ORPHA:100069	UNKNOWN
PSEN2	Alzheimer disease-4	OMIM:606889	MENDELIAN
PSEN2	Cardiomyopathy, dilated, 1V	OMIM:613697	MENDELIAN
PSEN2	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
PSEN2	Early-onset autosomal dominant Alzheimer disease	ORPHA:1020	UNKNOWN
PSENEN	Acne inversa, familial, 2, with or without dowing-degos disease	OMIM:613736	MENDELIAN
PSENEN	Dowling-Degos disease	ORPHA:79145	UNKNOWN
PSMA4	—		UNKNOWN
PSMA7	—		UNKNOWN
PSMB8	Proteasome-associated autoinflammatory syndrome 1 and digenic forms	OMIM:256040	MENDELIAN
PSMB9	Proteasome-associated autoinflammatory syndrome 6	OMIM:620796	MENDELIAN
PSMC3IP	Ovarian dysgenesis 3	OMIM:614324	MENDELIAN
PSMC3IP	46,XX gonadal dysgenesis	ORPHA:243	UNKNOWN
PSMD11	—		UNKNOWN
PSMD12	Stankiewicz-Isidor syndrome	OMIM:617516	MENDELIAN
PSMD12	17q24.2 microdeletion syndrome	ORPHA:529962	UNKNOWN
PSMD6	—		UNKNOWN
PSMD7	—		UNKNOWN
PSPH	Phosphoserine phosphatase deficiency	OMIM:614023	MENDELIAN
PSPH	3-phosphoserine phosphatase deficiency, infantile/juvenile form	ORPHA:79350	UNKNOWN
PSPN	—		UNKNOWN
PSTPIP1	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne	OMIM:604416	MENDELIAN
PSTPIP1	Hyperzincemia with functional zinc depletion	OMIM:601979	MENDELIAN
PSTPIP1	PAPA syndrome	ORPHA:69126	UNKNOWN
PSTPIP2	—		UNKNOWN
PTAFR	—		UNKNOWN
PTCD1	—		UNKNOWN
PTCH1	Holoprosencephaly 7	OMIM:610828	MENDELIAN
PTCH1	Basal cell nevus syndrome	OMIM:109400	MENDELIAN
PTCH1	Basal cell carcinoma, susceptibility to, 1	OMIM:605462	MENDELIAN
PTCH1	Gorlin syndrome	ORPHA:377	UNKNOWN
PTCH1	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
PTCH1	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
PTCH1	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
PTCH1	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
PTCH1	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
PTCH1	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
PTCH1	Monosomy 9q22.3 syndrome	ORPHA:77301	UNKNOWN
PTCH2	Basal cell carcinoma, susceptibility to, 1	OMIM:605462	MENDELIAN
PTCH2	Medulloblastoma	OMIM:155255	MENDELIAN
PTCH2	Gorlin syndrome	ORPHA:377	UNKNOWN
PTCHD1	Autism, susceptibility to, X-linked 4	OMIM:300830	POLYGENIC
PTCHD3	—		UNKNOWN
PTCSC3	—		UNKNOWN
PTDSS1	Lenz-Majewski hyperostotic dwarfism	OMIM:151050	MENDELIAN
PTDSS1	Lenz-Majewski hyperostotic dysplasia	ORPHA:2658	UNKNOWN
PTEN	Meningioma, familial, susceptibility to	OMIM:607174	POLYGENIC
PTEN	Glioma susceptibility 2	OMIM:613028	POLYGENIC
PTEN	Cowden syndrome 1	OMIM:158350	MENDELIAN
PTEN	Prostate cancer	OMIM:176807	MENDELIAN
PTEN	Macrocephaly/autism syndrome	OMIM:605309	MENDELIAN
PTEN	Cowden syndrome	ORPHA:201	UNKNOWN
PTEN	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
PTEN	Bannayan-Riley-Ruvalcaba syndrome	ORPHA:109	UNKNOWN
PTEN	Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome	ORPHA:137608	UNKNOWN
PTEN	Lhermitte-Duclos disease	ORPHA:65285	UNKNOWN
PTEN	Macrocephaly-intellectual disability-autism syndrome	ORPHA:210548	UNKNOWN
PTEN	Juvenile polyposis of infancy	ORPHA:79076	UNKNOWN
PTEN	Proteus-like syndrome	ORPHA:2969	UNKNOWN
PTF1A	Pancreatic and cerebellar agenesis	OMIM:609069	MENDELIAN
PTF1A	Pancreatic agenesis 2	OMIM:615935	MENDELIAN
PTF1A	Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome	ORPHA:65288	UNKNOWN
PTGDR2	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
PTGDS	—		UNKNOWN
PTGER2	Asthma, nasal polyps, and aspirin intolerance	OMIM:208550	POLYGENIC
PTGER4	—		UNKNOWN
PTGES2	—		UNKNOWN
PTGIR	—		UNKNOWN
PTGIS	Hypertension, essential	OMIM:145500	MENDELIAN
PTGS1	—		UNKNOWN
PTGS2	—		UNKNOWN
PTH	Hypoparathyroidism, familial isolated	OMIM:146200	MENDELIAN
PTH1R	Chondrodysplasia, Blomstrand type	OMIM:215045	MENDELIAN
PTH1R	Eiken syndrome	OMIM:600002	MENDELIAN
PTH1R	Metaphyseal chondrodysplasia, Jansen type	OMIM:156400	MENDELIAN
PTH1R	Dental noneruption	OMIM:125350	MENDELIAN
PTH1R	Ollier disease	ORPHA:296	UNKNOWN
PTH1R	Blomstrand lethal chondrodysplasia	ORPHA:50945	UNKNOWN
PTH1R	Eiken syndrome	ORPHA:79106	UNKNOWN
PTH1R	Metaphyseal chondrodysplasia, Jansen type	ORPHA:33067	UNKNOWN
PTH2R	—		UNKNOWN
PTHLH	Brachydactyly, type E2	OMIM:613382	MENDELIAN
PTHLH	Brachydactyly type E	ORPHA:93387	UNKNOWN
PTK7	—		UNKNOWN
PTPN1	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
PTPN11	Leopard syndrome 1	OMIM:151100	MENDELIAN
PTPN11	Metachondromatosis	OMIM:156250	MENDELIAN
PTPN11	Juvenile myelomonocytic leukemia	OMIM:607785	MENDELIAN
PTPN11	Noonan syndrome 1	OMIM:163950	MENDELIAN
PTPN11	Noonan syndrome	ORPHA:648	UNKNOWN
PTPN11	Metachondromatosis	ORPHA:2499	UNKNOWN
PTPN11	Noonan syndrome with multiple lentigines	ORPHA:500	UNKNOWN
PTPN12	Colorectal cancer	OMIM:114500	MENDELIAN
PTPN13	—		UNKNOWN
PTPN14	Choanal atresia and lymphedema	OMIM:613611	MENDELIAN
PTPN2	Oligoarticular juvenile idiopathic arthritis	ORPHA:85410	UNKNOWN
PTPN2	Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	ORPHA:85408	UNKNOWN
PTPN21	—		UNKNOWN
PTPN22	Giant cell arteritis	ORPHA:397	UNKNOWN
PTPN22	Oligoarticular juvenile idiopathic arthritis	ORPHA:85410	UNKNOWN
PTPN22	Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	ORPHA:85408	UNKNOWN
PTPN22	Granulomatosis with polyangiitis	ORPHA:900	UNKNOWN
PTPN22	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
PTPN22	Vogt-Koyanagi-Harada disease	ORPHA:3437	UNKNOWN
PTPN23	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	OMIM:618890	MENDELIAN
PTPN4	—		UNKNOWN
PTPN6	Sweet syndrome	ORPHA:3243	UNKNOWN
PTPRB	—		UNKNOWN
PTPRC	Immunodeficiency 105, severe combined	OMIM:619924	MENDELIAN
PTPRCAP	—		UNKNOWN
PTPRD	—		UNKNOWN
PTPRF	Breasts and/or nipples, aplasia or hypoplasia of, 2	OMIM:616001	MENDELIAN
PTPRJ	Thrombocytopenia 10	OMIM:620484	MENDELIAN
PTPRJ	Colorectal cancer	OMIM:114500	MENDELIAN
PTPRK	—		UNKNOWN
PTPRN2	—		UNKNOWN
PTPRO	Nephrotic syndrome, type 6	OMIM:614196	MENDELIAN
PTPRO	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
PTPRQ	Deafness, autosomal dominant 73	OMIM:617663	MENDELIAN
PTPRQ	Deafness, autosomal recessive 84	OMIM:613391	MENDELIAN
PTPRS	—		UNKNOWN
PTPRT	—		UNKNOWN
PTPRZ1	—		UNKNOWN
PTRF	—		UNKNOWN
PTRH2	Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset	OMIM:616263	MENDELIAN
PTRH2	Infantile multisystem neurologic-endocrine-pancreatic disease	ORPHA:456312	UNKNOWN
PTS	Hyperphenylalaninemia, BH4-deficient, A	OMIM:261640	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
PTS	6-pyruvoyl-tetrahydropterin synthase deficiency	ORPHA:13	UNKNOWN
PUF60	Verheij syndrome	OMIM:615583	MENDELIAN
PUF60	Intellectual disability-cardiac anomalies-short stature-joint laxity syndrome	ORPHA:508498	UNKNOWN
PUF60	8q24.3 microdeletion syndrome	ORPHA:508488	UNKNOWN
PURA	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties	OMIM:616158	MENDELIAN
PURA	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation	ORPHA:438216	UNKNOWN
PURA	Severe neonatal hypotonia-seizures-encephalopathy syndrome due to 5q31.3 microdeletion	ORPHA:314655	UNKNOWN
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1	OMIM:600462	MENDELIAN
PUS1	Mitochondrial myopathy and sideroblastic anemia	ORPHA:2598	UNKNOWN
PVR	—		UNKNOWN
PVRL1	—		UNKNOWN
PVRL3	—		UNKNOWN
PVRL4	—		UNKNOWN
PVT1	—		UNKNOWN
PWRN1	Prader-Willi syndrome	OMIM:176270	MENDELIAN
PWWP2A	—		UNKNOWN
PXDN	Corneal opacification with other ocular anomalies	OMIM:269400	MENDELIAN
PYCR1	Cutis laxa, autosomal recessive, type IIB	OMIM:612940	MENDELIAN
PYCR1	Cutis laxa, autosomal recessive, type IIIB	OMIM:614438	MENDELIAN
PYCR1	Geroderma osteodysplastica	ORPHA:2078	UNKNOWN
PYCR2	Leukodystrophy, hypomyelinating, 10	OMIM:616420	MENDELIAN
PYCR2	PYCR2-related microcephaly-progressive leukoencephalopathy	ORPHA:481152	UNKNOWN
PYCR2	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
PYCRL	—		UNKNOWN
PYGL	Glycogen storage disease VI	OMIM:232700	MENDELIAN
PYGL	Glycogen storage disease due to liver glycogen phosphorylase deficiency	ORPHA:369	UNKNOWN
PYGM	Glycogen storage disease V	OMIM:232600	MENDELIAN
PYGM	Glycogen storage disease due to muscle glycogen phosphorylase deficiency	ORPHA:368	UNKNOWN
PYGO1	—		UNKNOWN
PYY	—		UNKNOWN
PZP	—		UNKNOWN
QARS	—		UNKNOWN
QDPR	Hyperphenylalaninemia, bh4-deficient, C	OMIM:261630	MENDELIAN
QDPR	Dihydropteridine reductase deficiency	ORPHA:226	UNKNOWN
RAB10	—		UNKNOWN
RAB11FIP5	—		UNKNOWN
RAB18	Warburg micro syndrome 3	OMIM:614222	MENDELIAN
RAB18	Micro syndrome	ORPHA:2510	UNKNOWN
RAB23	Carpenter syndrome 1	OMIM:201000	MENDELIAN
RAB23	Carpenter syndrome	ORPHA:65759	UNKNOWN
RAB25	—		UNKNOWN
RAB27A	Griscelli syndrome, type 2	OMIM:607624	MENDELIAN
RAB27A	Griscelli syndrome type 2	ORPHA:79477	UNKNOWN
RAB27B	—		UNKNOWN
RAB28	Cone-Rod dystrophy 18	OMIM:615374	MENDELIAN
RAB28	Cone rod dystrophy	ORPHA:1872	UNKNOWN
RAB2A	—		UNKNOWN
RAB33B	Smith-Mccort dysplasia 2	OMIM:615222	MENDELIAN
RAB34	Orofaciodigital syndrome XX	OMIM:620718	MENDELIAN
RAB35	—		UNKNOWN
RAB39B	Intellectual developmental disorder, X-linked 72	OMIM:300271	MENDELIAN
RAB39B	Waisman syndrome	OMIM:311510	MENDELIAN
RAB39B	Early-onset parkinsonism-intellectual disability syndrome	ORPHA:2379	UNKNOWN
RAB3A	Spinocerebellar ataxia 52	OMIM:621535	MENDELIAN
RAB3GAP1	Warburg micro syndrome 1	OMIM:600118	MENDELIAN
RAB3GAP1	Martsolf syndrome 2	OMIM:619420	MENDELIAN
RAB3GAP1	Micro syndrome	ORPHA:2510	UNKNOWN
RAB3GAP1	Cataract-intellectual disability-hypogonadism syndrome	ORPHA:1387	UNKNOWN
RAB3GAP2	Warburg micro syndrome 2	OMIM:614225	MENDELIAN
RAB3GAP2	Martsolf syndrome 1	OMIM:212720	MENDELIAN
RAB3GAP2	Autosomal recessive spastic paraplegia type 69	ORPHA:401830	UNKNOWN
RAB3GAP2	Micro syndrome	ORPHA:2510	UNKNOWN
RAB3GAP2	Cataract-intellectual disability-hypogonadism syndrome	ORPHA:1387	UNKNOWN
RAB40AL	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
RAB7A	Charcot-Marie-Tooth disease, axonal, type 2B	OMIM:600882	MENDELIAN
RAB7L1	—		UNKNOWN
RABGGTA	—		UNKNOWN
RABL6	—		UNKNOWN
RAC1	Intellectual developmental disorder, autosomal dominant 48	OMIM:617751	MENDELIAN
RAC1	Microcephaly-corporis callosum and cerebellar vermis hypoplasia-facial dysmorphism-intellectual disability syndrome	ORPHA:500159	UNKNOWN
RAC2	Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia	OMIM:618986	MENDELIAN
RAC2	Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia	OMIM:618987	MENDELIAN
RAC2	Immunodeficiency 73A with defective neutrophil chemotaxis and leukocytosis	OMIM:608203	MENDELIAN
RAC2	Infantile LAD-like disease due to RAC2 deficiency	ORPHA:183707	UNKNOWN
RAD1	—		UNKNOWN
RAD17	—		UNKNOWN
RAD18	—		UNKNOWN
RAD21	Mungan syndrome	OMIM:611376	MENDELIAN
RAD21	Cornelia de Lange syndrome 4	OMIM:614701	MENDELIAN
RAD21	Cornelia de Lange syndrome	ORPHA:199	UNKNOWN
RAD21	Trichorhinophalangeal syndrome type 2	ORPHA:502	UNKNOWN
RAD21L	—		UNKNOWN
RAD21L1	—		UNKNOWN
RAD23A	—		UNKNOWN
RAD23B	—		UNKNOWN
RAD50	Nijmegen breakage syndrome-like disorder	OMIM:613078	MENDELIAN
RAD50	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
RAD51	Breast cancer	OMIM:114480	POLYGENIC
RAD51	Mirror movements 2	OMIM:614508	MENDELIAN
RAD51	Mirror movements 1 and/or agenesis of the corpus callosum	OMIM:157600	MENDELIAN
RAD51	Fanconi anemia, complementation group R	OMIM:617244	MENDELIAN
RAD51	Fanconi anemia	ORPHA:84	UNKNOWN
RAD51	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
RAD51	Familial congenital mirror movements	ORPHA:238722	UNKNOWN
RAD51B	—		UNKNOWN
RAD51C	Fanconi anemia, complementation group O	OMIM:613390	MENDELIAN
RAD51C	Familial breast-ovarian cancer-1	OMIM:613399	POLYGENIC
RAD51C	Fanconi anemia	ORPHA:84	UNKNOWN
RAD51C	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
RAD51D	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
RAD52	—		UNKNOWN
RAD54B	Lymphoma, non-hodgkin, familial	OMIM:605027	MENDELIAN
RAD54B	Colorectal cancer	OMIM:114500	MENDELIAN
RAD54L	Lymphoma, non-hodgkin, familial	OMIM:605027	MENDELIAN
RAD54L	Breast cancer	OMIM:114480	POLYGENIC
RAD54L2	—		UNKNOWN
RAD9A	—		UNKNOWN
RAD9B	—		UNKNOWN
RAET1L	—		UNKNOWN
RAF1	Leopard syndrome 2	OMIM:611554	MENDELIAN
RAF1	Noonan syndrome 5	OMIM:611553	MENDELIAN
RAF1	Cardiomyopathy, dilated, 1nn	OMIM:615916	MENDELIAN
RAF1	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
RAF1	Large/giant congenital melanocytic nevus	ORPHA:626	UNKNOWN
RAF1	Noonan syndrome	ORPHA:648	UNKNOWN
RAF1	Noonan syndrome with multiple lentigines	ORPHA:500	UNKNOWN
RAG1	Combined cellular and humoral immune defects with granulomas	OMIM:233650	MENDELIAN
RAG1	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity	OMIM:609889	MENDELIAN
RAG1	Omenn syndrome	OMIM:603554	MENDELIAN
RAG1	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-positive	OMIM:601457	MENDELIAN
RAG1	Omenn syndrome	ORPHA:39041	UNKNOWN
RAG1	T-B-NK+ severe combined immunodeficiency due to complete RAG1/2 deficiency	ORPHA:331206	UNKNOWN
RAG1	Atypical/leaky severe combined immunodeficiency due to partial RAG defect	ORPHA:231154	UNKNOWN
RAG2	Combined cellular and humoral immune defects with granulomas	OMIM:233650	MENDELIAN
RAG2	Omenn syndrome	OMIM:603554	MENDELIAN
RAG2	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-positive	OMIM:601457	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
RAG2	Omenn syndrome	ORPHA:39041	UNKNOWN
RAG2	T-B-NK+ severe combined immunodeficiency due to complete RAG1/2 deficiency	ORPHA:331206	UNKNOWN
RAI1	Potocki-Lupski syndrome	OMIM:610883	MENDELIAN
RAI1	Smith-Magenis syndrome	OMIM:182290	MENDELIAN
RAI1	PMP22-RAI1 contiguous gene duplication syndrome	ORPHA:477817	UNKNOWN
RAI1	Smith-Magenis syndrome	ORPHA:819	UNKNOWN
RAI1	17p11.2 microduplication syndrome	ORPHA:1713	UNKNOWN
RALA	Hiatt-Neu-Cooper neurodevelopmental syndrome	OMIM:619311	MENDELIAN
RALGAPA1	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation	OMIM:618797	MENDELIAN
RALGAPB	—		UNKNOWN
RALGDS	—		UNKNOWN
RANBP2	Encephalopathy, acute, infection-induced, susceptibility to, 3	OMIM:608033	POLYGENIC
RANBP2	Familial acute necrotizing encephalopathy	ORPHA:88619	UNKNOWN
RANBP6	—		UNKNOWN
RANGRF	Brugada syndrome	ORPHA:130	UNKNOWN
RAP1A	—		UNKNOWN
RAP1B	Thrombocytopenia 11 with multiple congenital anomalies and dysmorphic facies	OMIM:620654	MENDELIAN
RAP1GDS1	Alfadhel syndrome	OMIM:620655	MENDELIAN
RAPGEF1	—		UNKNOWN
RAPGEF4	—		UNKNOWN
RAPSN	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	OMIM:616326	MENDELIAN
RAPSN	Fetal akinesia deformation sequence 2	OMIM:618388	MENDELIAN
RAPSN	Fetal akinesia deformation sequence	ORPHA:994	UNKNOWN
RARA	Acute promyelocytic leukemia	OMIM:612376	MENDELIAN
RARA	Acute promyelocytic leukemia	ORPHA:520	UNKNOWN
RARB	Microphthalmia, syndromic 12	OMIM:615524	MENDELIAN
RARS	—		UNKNOWN
RARS2	Pontocerebellar hypoplasia, type 6	OMIM:611523	MENDELIAN
RASA1	Capillary malformation-arteriovenous malformation 1	OMIM:608354	MENDELIAN
RASA1	Basal cell carcinoma, susceptibility to, 1	OMIM:605462	MENDELIAN
RASA1	Parkes Weber syndrome	ORPHA:90307	UNKNOWN
RASA2	Noonan syndrome	ORPHA:648	UNKNOWN
RASGEF1B	—		UNKNOWN
RASGRP1	Immunodeficiency 64	OMIM:618534	MENDELIAN
RASGRP2	Bleeding disorder, platelet-type, 18	OMIM:615888	MENDELIAN
RASSF1	—		UNKNOWN
RASSF5	—		UNKNOWN
RAX	Microphthalmia, isolated 3	OMIM:611038	MENDELIAN
RAX	Isolated complete colobomatous microphthalmia	ORPHA:35612	UNKNOWN
RAX2	Cone-Rod dystrophy 11	OMIM:610381	MENDELIAN
RAX2	Retinitis pigmentosa 95	OMIM:620102	MENDELIAN
RAX2	Macular degeneration, age-related, 6	OMIM:613757	MENDELIAN
RAX2	Cone rod dystrophy	ORPHA:1872	UNKNOWN
RB1	Small cell cancer of the lung	OMIM:182280	MENDELIAN
RB1	Osteosarcoma	OMIM:259500	MENDELIAN
RB1	Bladder cancer	OMIM:109800	MENDELIAN
RB1	Retinoblastoma	OMIM:180200	MENDELIAN
RB1	Monosomy 13q14 syndrome	ORPHA:1587	UNKNOWN
RB1	Osteosarcoma	ORPHA:668	UNKNOWN
RB1CC1	Breast cancer	OMIM:114480	MENDELIAN
RBBP7	Spermatogenic failure, X-linked, 9	OMIM:301137	MENDELIAN
RBBP7	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
RBBP8	Seckel syndrome 2	OMIM:606744	MENDELIAN
RBBP8	Jawad syndrome	OMIM:251255	MENDELIAN
RBBP8	Seckel syndrome	ORPHA:808	UNKNOWN
RBCK1	Polyglucosan body myopathy 1 with or without immunodeficiency	OMIM:615895	MENDELIAN
RBFA	—		UNKNOWN
RBFOX1	—		UNKNOWN
RBFOX3	—		UNKNOWN
RBL1	—		UNKNOWN
RBL2	Brunet-Wagner neurodevelopmental syndrome	OMIM:619690	MENDELIAN
RBM10	TARP syndrome	OMIM:311900	MENDELIAN
RBM10	TARP syndrome	ORPHA:2886	UNKNOWN
RBM20	Cardiomyopathy, dilated, 1DD	OMIM:613172	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
RBM20	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
RBM28	Alopecia, neurologic defects, and endocrinopathy syndrome	OMIM:612079	MENDELIAN
RBM28	ANE syndrome	ORPHA:157954	UNKNOWN
RBM43	—		UNKNOWN
RBM8A	Thrombocytopenia-absent radius syndrome	OMIM:274000	MENDELIAN
RBM8A	Thrombocytopenia-absent radius syndrome	ORPHA:3320	UNKNOWN
RBMXL2	—		UNKNOWN
RBMY1A1	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
RBMY1A1	Chromosome Y microdeletion syndrome	ORPHA:1646	UNKNOWN
RBP3	Retinitis pigmentosa 66	OMIM:615233	MENDELIAN
RBP3	Retinitis pigmentosa	OMIM:268000	MENDELIAN
RBP3	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RBP4	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome	OMIM:615147	MENDELIAN
RBP4	Microphthalmia, isolated, with coloboma 10	OMIM:616428	MENDELIAN
RBPJ	Adams-Oliver syndrome 3	OMIM:614814	MENDELIAN
RBPJ	Adams-Oliver syndrome	ORPHA:974	UNKNOWN
RBX1	—		UNKNOWN
RC3H1	Immune dysregulation and systemic hyperinflammation syndrome	OMIM:618998	MENDELIAN
RCAN1	—		UNKNOWN
RCBTB1	Retinal dystrophy with or without extraocular anomalies	OMIM:617175	MENDELIAN
RCHY1	—		UNKNOWN
RD3	Leber congenital amaurosis 12	OMIM:610612	MENDELIAN
RD3	Leber congenital amaurosis	ORPHA:65	UNKNOWN
RDH11	Retinal dystrophy, juvenile cataracts, and short stature syndrome	OMIM:616108	MENDELIAN
RDH11	Retinitis pigmentosa-juvenile cataract-short stature-intellectual disability syndrome	ORPHA:436245	UNKNOWN
RDH12	Leber congenital amaurosis 13	OMIM:612712	MENDELIAN
RDH12	Leber congenital amaurosis	ORPHA:65	UNKNOWN
RDH12	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RDH5	Fundus albipunctatus	OMIM:136880	MENDELIAN
RDH5	Retinitis punctata albescens	ORPHA:52427	UNKNOWN
RDH8	Stargardt disease 5	OMIM:621259	MENDELIAN
RDM1	—		UNKNOWN
RDX	Deafness, autosomal recessive, 24	OMIM:611022	MENDELIAN
REC114	Oocyte maturation defect 10	OMIM:619176	MENDELIAN
REC8	—		UNKNOWN
RECQL	RECON progeroid syndrome	OMIM:620370	MENDELIAN
RECQL4	Rothmund-Thomson syndrome	OMIM:268400	MENDELIAN
RECQL4	Baller-Gerold syndrome	OMIM:218600	MENDELIAN
RECQL4	RAPADILINO syndrome	OMIM:266280	MENDELIAN
RECQL4	RAPADILINO syndrome	ORPHA:3021	UNKNOWN
RECQL4	Baller-Gerold syndrome	ORPHA:1225	UNKNOWN
RECQL4	Rothmund-Thomson syndrome type 2	ORPHA:221016	UNKNOWN
RECQL5	—		UNKNOWN
REEP1	Spastic paraplegia 31, autosomal dominant	OMIM:610250	MENDELIAN
REEP1	Spinal muscular atrophy, distal, autosomal recessive, 6	OMIM:620011	MENDELIAN
REEP1	Neuronopathy, distal hereditary motor, type VB	OMIM:614751	MENDELIAN
REEP1	Autosomal dominant spastic paraplegia type 31	ORPHA:101011	UNKNOWN
REEP1	Distal hereditary motor neuropathy type 5	ORPHA:139536	UNKNOWN
REEP2	Spastic paraplegia 72, autosomal recessive	OMIM:615625	MENDELIAN
REEP2	Spastic paraplegia 72B, autosomal recessive	OMIM:620606	MENDELIAN
REEP2	Autosomal spastic paraplegia type 72	ORPHA:401849	UNKNOWN
REL	Immunodeficiency 92	OMIM:619652	MENDELIAN
REL	2p15p16.1 microdeletion syndrome	ORPHA:261349	UNKNOWN
RELN	Epilepsy, familial temporal lobe, 7	OMIM:616436	POLYGENIC
RELN	Lissencephaly 2	OMIM:257320	MENDELIAN
RELN	Epilepsy, familial temporal lobe, 1	OMIM:600512	MENDELIAN
RELN	Epilepsy with auditory features	ORPHA:101046	UNKNOWN
RELN	Lissencephaly syndrome, Norman-Roberts type	ORPHA:89844	UNKNOWN
REN	Tubulointerstitial kidney disease, autosomal dominant, 4	OMIM:613092	MENDELIAN
REN	Renal tubular dysgenesis	OMIM:267430	MENDELIAN
REPS2	—		UNKNOWN
RERE	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart	OMIM:616975	MENDELIAN
RERE	RERE-related neurodevelopmental syndrome	ORPHA:494344	UNKNOWN
RERE	1p36 deletion syndrome	ORPHA:1606	UNKNOWN
REST	Fibromatosis, gingival, 5	OMIM:617626	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
REST	Wilms tumor 6	OMIM:616806	POLYGENIC
REST	Deafness, autosomal dominant 27	OMIM:612431	MENDELIAN
REST	Hereditary gingival fibromatosis	ORPHA:2024	UNKNOWN
REST	Nephroblastoma	ORPHA:654	UNKNOWN
RET	Hirschsprung disease, susceptibility to, 1	OMIM:142623	POLYGENIC
RET	Multiple endocrine neoplasia IIA	OMIM:171400	MENDELIAN
RET	Pheochromocytoma	OMIM:171300	MENDELIAN
RET	Multiple endocrine neoplasia, type IIB	OMIM:162300	MENDELIAN
RET	Thyroid carcinoma, familial medullary	OMIM:155240	MENDELIAN
RET	Hirschsprung disease	ORPHA:388	UNKNOWN
RET	Haddad syndrome	ORPHA:99803	UNKNOWN
RET	Renal agenesis, bilateral	ORPHA:1848	UNKNOWN
RET	Sporadic pheochromocytoma/secretory paraganglioma	ORPHA:276621	UNKNOWN
RET	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
RETN	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
REV1	—		UNKNOWN
REV3L	Moebius syndrome	ORPHA:570	UNKNOWN
RFC1	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome	OMIM:614575	MENDELIAN
RFC1	Cerebellar ataxia with neuropathy and bilateral vestibular areflexia syndrome	ORPHA:504476	UNKNOWN
RFC2	Williams syndrome	ORPHA:904	UNKNOWN
RFC3	—		UNKNOWN
RFC4	Morimoto-Ryu-Malicdan neuromuscular syndrome	OMIM:621010	MENDELIAN
RFC5	—		UNKNOWN
RFT1	Congenital disorder of glycosylation, type In	OMIM:612015	MENDELIAN
RFT1	RFT1-CDG	ORPHA:244310	UNKNOWN
RFWD2	—		UNKNOWN
RFX2	—		UNKNOWN
RFX3	—		UNKNOWN
RFX4	—		UNKNOWN
RFX5	MHC class II deficiency 5	OMIM:620818	MENDELIAN
RFX5	MHC class II deficiency 3	OMIM:620816	MENDELIAN
RFX5	Immunodeficiency by defective expression of MHC class II	ORPHA:572	UNKNOWN
RFX6	Mitchell-Riley syndrome	OMIM:615710	MENDELIAN
RFX7	Intellectual developmental disorder, autosomal dominant 71, with behavioral abnormalities	OMIM:620330	MENDELIAN
RFX8	—		UNKNOWN
RFXANK	MHC class II deficiency 2	OMIM:620815	MENDELIAN
RFXANK	Immunodeficiency by defective expression of MHC class II	ORPHA:572	UNKNOWN
RFXAP	MHC class II deficiency 4	OMIM:620817	MENDELIAN
RFXAP	Immunodeficiency by defective expression of MHC class II	ORPHA:572	UNKNOWN
RGL1	—		UNKNOWN
RGL2	—		UNKNOWN
RGMA	—		UNKNOWN
RGR	Retinitis pigmentosa 44	OMIM:613769	MENDELIAN
RGR	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RGS2	—		UNKNOWN
RGS5	Hypertension, essential	OMIM:145500	MENDELIAN
RGS6	—		UNKNOWN
RGS7	—		UNKNOWN
RGS9	Prolonged electroretinal response suppression 1	OMIM:608415	MENDELIAN
RGS9	Bradyopsia	ORPHA:75374	UNKNOWN
RGS9BP	Prolonged electroretinal response suppression 2	OMIM:620344	MENDELIAN
RGS9BP	Bradyopsia	ORPHA:75374	UNKNOWN
RHAG	Overhydrated hereditary stomatocytosis	OMIM:185000	MENDELIAN
RHAG	Anemia, hemolytic, Rh-null, Regulator type	OMIM:268150	MENDELIAN
RHAG	Overhydrated hereditary stomatocytosis	ORPHA:3203	UNKNOWN
RHAG	Rh deficiency syndrome	ORPHA:71275	UNKNOWN
RHBDD2	—		UNKNOWN
RHBDF2	Tylosis with esophageal cancer	OMIM:148500	MENDELIAN
RHBDF2	Palmoplantar keratoderma-esophageal carcinoma syndrome	ORPHA:2198	UNKNOWN
RHCE	Rh-null disease, amorph type	OMIM:617970	MENDELIAN
RHCE	Rh deficiency syndrome	ORPHA:71275	UNKNOWN
RHD	Hemolytic disease of fetus and newborn, RH-induced	OMIM:619462	POLYGENIC
RHD	Rh deficiency syndrome	ORPHA:71275	UNKNOWN
RHEB	—		UNKNOWN
RHO	Night blindness, congenital stationary, autosomal dominant 1	OMIM:610445	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
RHO	Fundus albipunctatus	OMIM:136880	MENDELIAN
RHO	Retinitis pigmentosa 4	OMIM:613731	MENDELIAN
RHO	Retinitis punctata albescens	ORPHA:52427	UNKNOWN
RHO	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RHOA	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic	OMIM:618727	MENDELIAN
RHOB	—		UNKNOWN
RHOG	—		UNKNOWN
RHOH	Epidermodysplasia verruciformis, susceptibility to, 4	OMIM:618307	MENDELIAN
RHOXF2	—		UNKNOWN
RHPN2	—		UNKNOWN
RICTOR	—		UNKNOWN
RIMS1	Cone rod dystrophy	ORPHA:1872	UNKNOWN
RIMS2	Cone-rod synaptic disorder syndrome, congenital nonprogressive	OMIM:618970	MENDELIAN
RIMS3	—		UNKNOWN
RIN2	Macrocephaly, alopecia, cutis laxa, and scoliosis	OMIM:613075	MENDELIAN
RIN2	RIN2 syndrome	ORPHA:217335	UNKNOWN
RINT1	Infantile liver failure syndrome 3	OMIM:618641	MENDELIAN
RIOK2	—		UNKNOWN
RIPK2	—		UNKNOWN
RIPK3	—		UNKNOWN
RIPK4	Popliteal pterygium syndrome, Bartsocas-Papas type 1	OMIM:263650	MENDELIAN
RIPK4	CHAND syndrome	OMIM:214350	MENDELIAN
RIPK4	CHAND syndrome	ORPHA:1401	UNKNOWN
RIPK4	Bartsocas-Papas syndrome	ORPHA:1234	UNKNOWN
RIT1	Noonan syndrome 8	OMIM:615355	MENDELIAN
RIT1	Noonan syndrome	ORPHA:648	UNKNOWN
RIT2	—		UNKNOWN
RLBP1	Fundus albipunctatus	OMIM:136880	MENDELIAN
RLBP1	Newfoundland rod-cone dystrophy	OMIM:607476	MENDELIAN
RLBP1	Bothnia retinal dystrophy	OMIM:607475	MENDELIAN
RLBP1	Retinitis punctata albescens	ORPHA:52427	UNKNOWN
RLBP1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RLBP1	Bothnia retinal dystrophy	ORPHA:85128	UNKNOWN
RLIM	Tonne-Kalscheuer syndrome	OMIM:300978	MENDELIAN
RMDN2	—		UNKNOWN
RMI1	—		UNKNOWN
RMND1	Combined oxidative phosphorylation deficiency 11	OMIM:614922	MENDELIAN
RMRP	Cartilage-hair hypoplasia	OMIM:250250	MENDELIAN
RMRP	Metaphyseal dysplasia without hypotrichosis	OMIM:250460	MENDELIAN
RMRP	Anauxetic dysplasia 1	OMIM:607095	MENDELIAN
RMRP	Cartilage-hair hypoplasia	ORPHA:175	UNKNOWN
RMRP	Omenn syndrome	ORPHA:39041	UNKNOWN
RNASE3	—		UNKNOWN
RNASEH2A	Aicardi-Goutieres syndrome 4	OMIM:610333	MENDELIAN
RNASEH2A	Aicardi-Goutières syndrome	ORPHA:51	UNKNOWN
RNASEH2B	Aicardi-Goutieres syndrome 2	OMIM:610181	MENDELIAN
RNASEH2B	Aicardi-Goutières syndrome	ORPHA:51	UNKNOWN
RNASEH2C	Aicardi-Goutieres syndrome 3	OMIM:610329	MENDELIAN
RNASEH2C	Aicardi-Goutières syndrome	ORPHA:51	UNKNOWN
RNASEL	Prostate cancer, hereditary, 1	OMIM:601518	MENDELIAN
RNASET2	Leukoencephalopathy, cystic, without megalencephaly	OMIM:612951	MENDELIAN
RNF113A	Trichothiodystrophy 5, nonphotosensitive	OMIM:300953	MENDELIAN
RNF113A	Trichothiodystrophy	ORPHA:33364	UNKNOWN
RNF114	—		UNKNOWN
RNF125	Tenorio syndrome	OMIM:616260	MENDELIAN
RNF128	—		UNKNOWN
RNF135	Overgrowth-macrocephaly-facial dysmorphism syndrome	ORPHA:137634	UNKNOWN
RNF139	Renal cell carcinoma, nonpapillary	OMIM:144700	MENDELIAN
RNF168	Riddle syndrome	OMIM:611943	MENDELIAN
RNF168	RIDDLE syndrome	ORPHA:420741	UNKNOWN
RNF17	—		UNKNOWN
RNF170	Spastic paraplegia 85, autosomal recessive	OMIM:619686	MENDELIAN
RNF170	Ataxia, sensory, autosomal dominant	OMIM:608984	MENDELIAN
RNF2	Luo-Schoch-Yamamoto syndrome	OMIM:619460	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
RNF20	—		UNKNOWN
RNF212	Spermatogenic failure 62	OMIM:619673	MENDELIAN
RNF212	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
RNF212B	—		UNKNOWN
RNF213	Moyamoya disease 2, susceptibility to	OMIM:607151	POLYGENIC
RNF213	Moyamoya disease	ORPHA:2573	UNKNOWN
RNF216	Cerebellar ataxia and hypogonadotropic hypogonadism	OMIM:212840	MENDELIAN
RNF216	Cerebellar ataxia-hypogonadism syndrome	ORPHA:1173	UNKNOWN
RNF219	—		UNKNOWN
RNF32	—		UNKNOWN
RNF38	—		UNKNOWN
RNF39	—		UNKNOWN
RNF43	Sessile serrated polyposis cancer syndrome	OMIM:617108	MENDELIAN
RNF43	Serrated polyposis syndrome	ORPHA:157798	UNKNOWN
RNF6	Esophageal cancer, somatic	OMIM:133239	MENDELIAN
RNF6	Squamous cell carcinoma of the esophagus	ORPHA:99977	UNKNOWN
RNF8	—		UNKNOWN
RNLS	—		UNKNOWN
RNMT	—		UNKNOWN
RNU2-2	Developmental and epileptic encephalopathy 119	OMIM:621304	MENDELIAN
RNU4-2	ReNU syndrome	OMIM:620851	MENDELIAN
RNU4-2	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RNU4ATAC	Lowry-Wood syndrome	OMIM:226960	MENDELIAN
RNU4ATAC	Roifman syndrome	OMIM:616651	MENDELIAN
RNU4ATAC	Microcephalic osteodysplastic primordial dwarfism, type I	OMIM:210710	MENDELIAN
RNU4ATAC	Microcephalic osteodysplastic primordial dwarfism types I and III	ORPHA:2636	UNKNOWN
RNU4ATAC	Lowry-Wood syndrome	ORPHA:1824	UNKNOWN
RNU4ATAC	Roifman syndrome	ORPHA:353298	UNKNOWN
RNU5B-1	Neurodevelopmental disorder with seizures and joint laxity	OMIM:621302	MENDELIAN
ROBO1	Neurooculorenal syndrome	OMIM:620305	MENDELIAN
ROBO1	Pituitary hormone deficiency, combined or isolated, 8	OMIM:620303	MENDELIAN
ROBO1	Nystagmus, congenital motor, autosomal recessive	OMIM:257400	MENDELIAN
ROBO1	Pituitary stalk interruption syndrome	ORPHA:95496	UNKNOWN
ROBO2	Vesicoureteral reflux 2	OMIM:610878	MENDELIAN
ROBO3	Gaze palsy, familial horizontal, with progressive scoliosis, 1	OMIM:607313	MENDELIAN
ROBO3	Horizontal gaze palsy with progressive scoliosis	ORPHA:2744	UNKNOWN
ROCK1	—		UNKNOWN
ROCK2	—		UNKNOWN
ROGDI	Kohlschutter-Tonz syndrome	OMIM:226750	MENDELIAN
ROGDI	Amelocerebrohypohidrotic syndrome	ORPHA:1946	UNKNOWN
ROM1	Retinitis pigmentosa 7	OMIM:608133	MENDELIAN
ROM1	Retinitis pigmentosa	OMIM:268000	MENDELIAN
ROM1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
ROPN1L	—		UNKNOWN
ROR2	Robinow syndrome, autosomal recessive	OMIM:268310	MENDELIAN
ROR2	Brachydactyly, type B1	OMIM:113000	MENDELIAN
ROR2	Autosomal recessive Robinow syndrome	ORPHA:1507	UNKNOWN
RORA	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia	OMIM:618060	MENDELIAN
RORB	Epilepsy, idiopathic generalized, susceptibility to, 15	OMIM:618357	POLYGENIC
RORC	Immunodeficiency 42	OMIM:616622	MENDELIAN
ROS1	Cholangiocarcinoma	ORPHA:70567	UNKNOWN
RP1	Retinitis pigmentosa 1	OMIM:180100	MENDELIAN
RP1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RP1L1	Retinitis pigmentosa 88	OMIM:618826	MENDELIAN
RP1L1	Occult macular dystrophy	OMIM:613587	MENDELIAN
RP1L1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RP2	Retinitis pigmentosa 2, X-linked	OMIM:312600	MENDELIAN
RP2	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RP9	Retinitis pigmentosa 9	OMIM:180104	MENDELIAN
RP9	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RPA1	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 6	OMIM:619767	MENDELIAN
RPA2	—		UNKNOWN
RPA3	—		UNKNOWN
RPA4	—		UNKNOWN
RPE65	Leber congenital amaurosis, type II	OMIM:204100	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
RPE65	Retinitis pigmentosa 87 with choroidal involvement	OMIM:618697	MENDELIAN
RPE65	Retinitis pigmentosa 20	OMIM:613794	MENDELIAN
RPE65	Leber congenital amaurosis	ORPHA:65	UNKNOWN
RPE65	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RPE65	Severe early-childhood-onset retinal dystrophy	ORPHA:364055	UNKNOWN
RPGR	Retinitis pigmentosa 3	OMIM:300029	MENDELIAN
RPGR	Retinitis pigmentosa, X-linked, and sinorespiratory infections, withor without deafness	OMIM:300455	MENDELIAN
RPGR	Macular degeneration, X-linked atrophic	OMIM:300834	MENDELIAN
RPGR	Cone-rod dystrophy, X-linked, 1	OMIM:304020	MENDELIAN
RPGR	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
RPGR	Cone rod dystrophy	ORPHA:1872	UNKNOWN
RPGR	Retinitis pigmentosa	ORPHA:791	UNKNOWN
RPGR	Achromatopsia	ORPHA:49382	UNKNOWN
RPGRIP1	Cone-rod dystrophy 13	OMIM:608194	MENDELIAN
RPGRIP1	Leber congenital amaurosis 6	OMIM:613826	MENDELIAN
RPGRIP1	Leber congenital amaurosis	ORPHA:65	UNKNOWN
RPGRIP1	Cone rod dystrophy	ORPHA:1872	UNKNOWN
RPGRIP1	Meckel syndrome	ORPHA:564	UNKNOWN
RPGRIP1L	Meckel syndrome, type 5	OMIM:611561	MENDELIAN
RPGRIP1L	Joubert syndrome 7	OMIM:611560	MENDELIAN
RPGRIP1L	COACH syndrome 3	OMIM:619113	MENDELIAN
RPGRIP1L	Joubert syndrome with renal defect	ORPHA:220497	UNKNOWN
RPGRIP1L	Joubert syndrome with hepatic defect	ORPHA:1454	UNKNOWN
RPGRIP1L	Meckel syndrome	ORPHA:564	UNKNOWN
RPH3AL	—		UNKNOWN
RPIA	Ribose 5-phosphate isomerase deficiency	OMIM:608611	MENDELIAN
RPL10	Intellectual developmental disorder, X-linked syndromic 35	OMIM:300998	MENDELIAN
RPL10	X-linked microcephaly-growth retardation-prognathism-cryptorchidism syndrome	ORPHA:435938	UNKNOWN
RPL10	X-linked intellectual disability-cerebellar hypoplasia-spondylo-epiphyseal dysplasia syndrome	ORPHA:459070	UNKNOWN
RPL11	Diamond-Blackfan anemia 7	OMIM:612562	MENDELIAN
RPL11	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPL15	Diamond-Blackfan anemia 12	OMIM:615550	MENDELIAN
RPL15	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPL21	Hypotrichosis 12	OMIM:615885	MENDELIAN
RPL21	Hypotrichosis simplex	ORPHA:55654	UNKNOWN
RPL24	—		UNKNOWN
RPL26	Diamond-Blackfan anemia 11	OMIM:614900	MENDELIAN
RPL26	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPL27	Diamond-Blackfan anemia 16	OMIM:617408	MENDELIAN
RPL27	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPL35A	Diamond-Blackfan anemia 5	OMIM:612528	MENDELIAN
RPL35A	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPL36	—		UNKNOWN
RPL38	—		UNKNOWN
RPL5	Diamond-Blackfan anemia 6	OMIM:612561	MENDELIAN
RPL5	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPL6	—		UNKNOWN
RPN2	—		UNKNOWN
RPS10	Diamond-Blackfan anemia 9	OMIM:613308	MENDELIAN
RPS10	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPS14	Chromosome 5q deletion syndrome	OMIM:153550	MENDELIAN
RPS14	Myelodysplastic syndrome associated with isolated del(5q) chromosome abnormality	ORPHA:86841	UNKNOWN
RPS17	Diamond-Blackfan anemia 4	OMIM:612527	MENDELIAN
RPS17	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPS19	Diamond-Blackfan anemia 1	OMIM:105650	MENDELIAN
RPS19	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPS24	Diamond-Blackfan anemia 3	OMIM:610629	MENDELIAN
RPS24	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPS26	Diamond-Blackfan anemia 10	OMIM:613309	MENDELIAN
RPS26	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPS27	Diamond-Blackfan anemia 17	OMIM:617409	MENDELIAN
RPS27	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPS27A	—		UNKNOWN
RPS28	Diamond-Blackfan anemia 15 with mandibulofacial dysostosis	OMIM:606164	MENDELIAN
RPS28	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
RPS29	Diamond-Blackfan anemia 13	OMIM:615909	MENDELIAN
RPS29	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPS3	—		UNKNOWN
RPS4Y2	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
RPS5	—		UNKNOWN
RPS6KA3	Coffin-Lowry syndrome	OMIM:303600	MENDELIAN
RPS6KA3	Intellectual developmental disorder, X-linked 19	OMIM:300844	MENDELIAN
RPS6KA3	Coffin-Lowry syndrome	ORPHA:192	UNKNOWN
RPS6KA3	Symptomatic form of Coffin-Lowry syndrome in female carriers	ORPHA:276630	UNKNOWN
RPS6KA4	—		UNKNOWN
RPS6KB1	—		UNKNOWN
RPS6KB2	—		UNKNOWN
RPS6KL1	—		UNKNOWN
RPS7	Diamond-Blackfan anemia 8	OMIM:612563	MENDELIAN
RPS7	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
RPSA	Asplenia, isolated congenital	OMIM:271400	MENDELIAN
RPSAP52	—		UNKNOWN
RPTOR	—		UNKNOWN
RRAS	Noonan syndrome	ORPHA:648	UNKNOWN
RRAS2	Noonan syndrome 12	OMIM:618624	MENDELIAN
RRAS2	Noonan syndrome	ORPHA:648	UNKNOWN
RREB1	22q11.2 deletion syndrome	ORPHA:567	UNKNOWN
RRH	—		UNKNOWN
RRM1	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6	OMIM:620647	MENDELIAN
RRM1	Autosomal recessive progressive external ophthalmoplegia	ORPHA:254886	UNKNOWN
RRM2	—		UNKNOWN
RRM2B	Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction	OMIM:268315	MENDELIAN
RRM2B	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5	OMIM:613077	MENDELIAN
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	OMIM:612075	MENDELIAN
RRM2B	Mitochondrial neurogastrointestinal encephalomyopathy	ORPHA:298	UNKNOWN
RRM2B	Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy	ORPHA:329336	UNKNOWN
RRM2B	Autosomal dominant progressive external ophthalmoplegia	ORPHA:254892	UNKNOWN
RRM2B	Kearns-Sayre syndrome	ORPHA:480	UNKNOWN
RRP1B	—		UNKNOWN
RS1	Retinoschisis 1, X-linked, juvenile	OMIM:312700	MENDELIAN
RS1	X-linked retinoschisis	ORPHA:792	UNKNOWN
RSC1A1	—		UNKNOWN
RSPH1	Ciliary dyskinesia, primary, 24	OMIM:615481	MENDELIAN
RSPH1	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
RSPH4A	Ciliary dyskinesia, primary, 11	OMIM:612649	MENDELIAN
RSPH4A	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
RSPH9	Ciliary dyskinesia, primary, 12	OMIM:612650	MENDELIAN
RSPH9	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
RSP01	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and 46,xx sex reversal	OMIM:610644	MENDELIAN
RSP01	Palmoplantar keratoderma-XX sex reversal-predisposition to squamous cell carcinoma syndrome	ORPHA:85112	UNKNOWN
RSP04	Anonychia congenita	OMIM:206800	MENDELIAN
RSRC1	Intellectual developmental disorder, autosomal recessive 70	OMIM:618402	MENDELIAN
RTEL1	Dyskeratosis congenita, autosomal recessive 5	OMIM:615190	MENDELIAN
RTEL1	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3	OMIM:616373	MENDELIAN
RTEL1	Idiopathic pulmonary fibrosis	ORPHA:2032	UNKNOWN
RTEL1	Hoyeraal-Hreidarsson syndrome	ORPHA:3322	UNKNOWN
RTEL1	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
RTF1	—		UNKNOWN
RTN2	Spastic paraplegia 12, autosomal dominant	OMIM:604805	MENDELIAN
RTN2	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity	OMIM:620854	MENDELIAN
RTN2	Autosomal dominant spastic paraplegia type 12	ORPHA:100993	UNKNOWN
RTN3	—		UNKNOWN
RTN4R	Schizophrenia	OMIM:181500	POLYGENIC
RTN4RL1	—		UNKNOWN
RTTN	Microcephaly, short stature, and polymicrogyria with or without seizures	OMIM:614833	MENDELIAN
RTTN	Microcephalic cortical malformations-short stature due to RTTN deficiency	ORPHA:468631	UNKNOWN
RUNX1	Platelet disorder, familial, with associated myeloid malignancy	OMIM:601399	MENDELIAN
RUNX1	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
RUNX1	Aggressive systemic mastocytosis	ORPHA:98850	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
RUNX1	Chronic myeloid leukemia	ORPHA:521	UNKNOWN
RUNX2	Metaphyseal dysplasia with maxillary hypoplasia and brachydactyly	OMIM:156510	MENDELIAN
RUNX2	Cleidocranial dysplasia	OMIM:119600	MENDELIAN
RUNX2	Cleidocranial dysplasia	ORPHA:1452	UNKNOWN
RUNX3	—		UNKNOWN
RUVBL1	—		UNKNOWN
RXFP2	—		UNKNOWN
RXRA	—		UNKNOWN
RXRG	—		UNKNOWN
RYK	—		UNKNOWN
RYR1	Minicore myopathy with external ophthalmoplegia	OMIM:255320	MENDELIAN
RYR1	King-Denborough syndrome	OMIM:619542	MENDELIAN
RYR1	Malignant hyperthermia, susceptibility to, 1	OMIM:145600	POLYGENIC
RYR1	Central core disease	OMIM:117000	MENDELIAN
RYR1	Benign Samaritan congenital myopathy	ORPHA:324581	UNKNOWN
RYR1	Moderate multiminicore disease with hand involvement	ORPHA:178145	UNKNOWN
RYR1	Autosomal recessive centronuclear myopathy	ORPHA:169186	UNKNOWN
RYR1	Autosomal dominant centronuclear myopathy	ORPHA:169189	UNKNOWN
RYR1	Exercise-induced malignant hyperthermia	ORPHA:466650	UNKNOWN
RYR1	Congenital multicore myopathy with external ophthalmoplegia	ORPHA:98905	UNKNOWN
RYR1	Congenital myopathy with myasthenic-like onset	ORPHA:424107	UNKNOWN
RYR1	Central core disease	ORPHA:597	UNKNOWN
RYR1	Malignant hyperthermia of anesthesia	ORPHA:423	UNKNOWN
RYR2	Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome	OMIM:115000	MENDELIAN
RYR2	Ventricular tachycardia, catecholaminergic polymorphic, 1, with or without atrial dysfunction and/or dilated cardiomyopathy	OMIM:604772	MENDELIAN
RYR3	Congenital myopathy 20	OMIM:620310	MENDELIAN
S100A14	—		UNKNOWN
S100B	—		UNKNOWN
S1PR1	—		UNKNOWN
S1PR3	—		UNKNOWN
SAA1	AA amyloidosis	ORPHA:85445	UNKNOWN
SAA2	—		UNKNOWN
SACS	Spastic ataxia, Charlevoix-Saguenay type	OMIM:270550	MENDELIAN
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	ORPHA:98	UNKNOWN
SAE1	—		UNKNOWN
SAG	Retinitis pigmentosa 96, autosomal dominant	OMIM:620228	MENDELIAN
SAG	Oguchi disease 1	OMIM:258100	MENDELIAN
SAG	Retinitis pigmentosa 47	OMIM:613758	MENDELIAN
SAG	Oguchi disease	ORPHA:75382	UNKNOWN
SAG	Retinitis pigmentosa	ORPHA:791	UNKNOWN
SAGE1	—		UNKNOWN
SALL1	Townes-Brocks syndrome 1	OMIM:107480	MENDELIAN
SALL1	Townes-Brocks syndrome	ORPHA:857	UNKNOWN
SALL2	Coloboma, ocular, autosomal recessive	OMIM:216820	MENDELIAN
SALL4	IVIC syndrome	OMIM:147750	MENDELIAN
SALL4	Duane-Radial ray syndrome	OMIM:607323	MENDELIAN
SALL4	Duane retraction syndrome	ORPHA:233	UNKNOWN
SALL4	Acro-renal-ocular syndrome	ORPHA:959	UNKNOWN
SALL4	IVIC syndrome	ORPHA:2307	UNKNOWN
SAMD4A	—		UNKNOWN
SAMD9	Tumoral calcinosis, normophosphatemic, familial	OMIM:610455	MENDELIAN
SAMD9	Monosomy 7 myelodysplasia and leukemia syndrome 2	OMIM:619041	MENDELIAN
SAMD9	Mirage syndrome	OMIM:617053	MENDELIAN
SAMHD1	Aicardi-Goutieres syndrome 5	OMIM:612952	MENDELIAN
SAMHD1	Chilblain lupus 2	OMIM:614415	MENDELIAN
SAMHD1	Aicardi-GoutiÃ`res syndrome	ORPHA:51	UNKNOWN
SAR1B	Chylomicron retention disease	OMIM:246700	MENDELIAN
SAR1B	Chylomicron retention disease	ORPHA:71	UNKNOWN
SARDH	Sarcosinemia	OMIM:268900	MENDELIAN
SARDH	Sarcosinemia	ORPHA:3129	UNKNOWN
SARS	—		UNKNOWN
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis syndrome	OMIM:613845	MENDELIAN
SART1	—		UNKNOWN
SART3	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
SASH1	Dyschromatosis universalis hereditaria	OMIM:127500	MENDELIAN
SASH1	Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma	OMIM:618373	MENDELIAN
SASS6	Microcephaly 14, primary, autosomal recessive	OMIM:616402	MENDELIAN
SASS6	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
SAT1	Keratosis follicularis spinulosa decalvans	ORPHA:2340	UNKNOWN
SAT1	Pediatric systemic lupus erythematosus	ORPHA:93552	UNKNOWN
SATB1	Developmental delay with dysmorphic facies and dental anomalies	OMIM:619228	MENDELIAN
SATB1	Kohlschutter-Tonz syndrome-like	OMIM:619229	MENDELIAN
SATB2	Glass syndrome	OMIM:612313	MENDELIAN
SATB2	SATB2-associated syndrome due to a pathogenic variant	ORPHA:576283	UNKNOWN
SATB2	SATB2-associated syndrome due to a chromosomal rearrangement	ORPHA:251028	UNKNOWN
SATB2	2q32q33 deletion syndrome	ORPHA:251019	UNKNOWN
SATL1	—		UNKNOWN
SBDS	Aplastic anemia	OMIM:609135	POLYGENIC
SBDS	Shwachman-Diamond syndrome 1	OMIM:260400	MENDELIAN
SBDS	Idiopathic aplastic anemia	ORPHA:88	UNKNOWN
SBDS	Shwachman-Diamond syndrome	ORPHA:811	UNKNOWN
SBF1	Charcot-Marie-Tooth disease, type 4B3	OMIM:615284	MENDELIAN
SBF2	Charcot-Marie-Tooth disease, type 4B2	OMIM:604563	MENDELIAN
SBF2	Charcot-Marie-Tooth disease type 4B2	ORPHA:99956	UNKNOWN
SBNO1	—		UNKNOWN
SC5DL	—		UNKNOWN
SCAF4	Fliedner-Zweier syndrome	OMIM:620511	MENDELIAN
SCAP	—		UNKNOWN
SCAPER	Intellectual developmental disorder and retinitis pigmentosa	OMIM:618195	MENDELIAN
SCAPER	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
SCAPER	Retinitis pigmentosa	ORPHA:791	UNKNOWN
SCARB2	Epilepsy, progressive myoclonic, 4, with or without renal failure	OMIM:254900	MENDELIAN
SCARB2	Progressive myoclonic epilepsy type 1	ORPHA:308	UNKNOWN
SCARB2	Gaucher disease type 1	ORPHA:77259	UNKNOWN
SCARF2	Van den Ende-Gupta syndrome	OMIM:600920	MENDELIAN
SCARF2	Van den Ende-Gupta syndrome	ORPHA:2460	UNKNOWN
SCG2	—		UNKNOWN
SCG3	—		UNKNOWN
SCG5	—		UNKNOWN
SCGB1A1	—		UNKNOWN
SCGB1D2	—		UNKNOWN
SCGB3A2	Asthma, susceptibility to	OMIM:600807	POLYGENIC
SCLT1	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
SCML1	—		UNKNOWN
SCML2	—		UNKNOWN
SCN10A	Episodic pain syndrome, familial, 2	OMIM:615551	MENDELIAN
SCN10A	Brugada syndrome	ORPHA:130	UNKNOWN
SCN10A	Primary erythromelalgia	ORPHA:90026	UNKNOWN
SCN10A	Paroxysmal extreme pain disorder	ORPHA:46348	UNKNOWN
SCN11A	Neuropathy, hereditary sensory and autonomic, type VII	OMIM:615548	MENDELIAN
SCN11A	Episodic pain syndrome, familial, 3	OMIM:615552	MENDELIAN
SCN11A	Primary erythromelalgia	ORPHA:90026	UNKNOWN
SCN11A	Paroxysmal extreme pain disorder	ORPHA:46348	UNKNOWN
SCN1A	Generalized epilepsy with febrile seizures plus, type 2	OMIM:604403	MENDELIAN
SCN1A	Epileptic encephalopathy, early infantile, 6 (Dravet syndrome)	OMIM:607208	MENDELIAN
SCN1A	Migraine, familial hemiplegic, 3	OMIM:609634	MENDELIAN
SCN1A	Developmental and epileptic encephalopathy 6B, non-Dravet	OMIM:619317	MENDELIAN
SCN1A	Genetic epilepsy with febrile seizure plus	ORPHA:36387	UNKNOWN
SCN1A	Lennox-Gastaut syndrome	ORPHA:2382	UNKNOWN
SCN1A	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
SCN1A	Epilepsy with myoclonic-atonic seizures	ORPHA:1942	UNKNOWN
SCN1A	Epilepsy of infancy with migrating focal seizures	ORPHA:293181	UNKNOWN
SCN1A	Familial or sporadic hemiplegic migraine	ORPHA:569	UNKNOWN
SCN1A	Dravet syndrome	ORPHA:33069	UNKNOWN
SCN1B	Brugada syndrome 5	OMIM:612838	MENDELIAN
SCN1B	Generalized epilepsy with febrile seizures plus, type 1	OMIM:604233	MENDELIAN
SCN1B	Atrial fibrillation, familial, 13	OMIM:615377	MENDELIAN
SCN1B	Epileptic encephalopathy, early infantile, 52	OMIM:617350	MENDELIAN
SCN1B	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
SCN1B	Genetic epilepsy with febrile seizure plus	ORPHA:36387	UNKNOWN
SCN1B	Brugada syndrome	ORPHA:130	UNKNOWN
SCN1B	Hereditary progressive cardiac conduction defect	ORPHA:871	UNKNOWN
SCN1B	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
SCN1B	Dravet syndrome	ORPHA:33069	UNKNOWN
SCN2A	Episodic ataxia, type 9	OMIM:618924	MENDELIAN
SCN2A	Epileptic encephalopathy, early infantile, 11	OMIM:613721	MENDELIAN
SCN2A	Seizures, benign familial infantile, 3	OMIM:607745	MENDELIAN
SCN2A	Self-limited infantile epilepsy	ORPHA:306	UNKNOWN
SCN2A	Genetic epilepsy with febrile seizure plus	ORPHA:36387	UNKNOWN
SCN2A	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
SCN2A	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
SCN2A	Epilepsy of infancy with migrating focal seizures	ORPHA:293181	UNKNOWN
SCN2A	Self-limited neonatal/infantile epilepsy	ORPHA:140927	UNKNOWN
SCN2A	Alternating hemiplegia of childhood	ORPHA:2131	UNKNOWN
SCN2A	Dravet syndrome	ORPHA:33069	UNKNOWN
SCN2B	Atrial fibrillation, familial, 14	OMIM:615378	MENDELIAN
SCN2B	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
SCN2B	Brugada syndrome	ORPHA:130	UNKNOWN
SCN3A	Epilepsy, familial focal, with variable foci 4	OMIM:617935	MENDELIAN
SCN3A	Developmental and epileptic encephalopathy 62	OMIM:617938	MENDELIAN
SCN3A	Familial focal epilepsy with variable foci	ORPHA:98820	UNKNOWN
SCN3A	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
SCN3B	Brugada syndrome 7	OMIM:613120	MENDELIAN
SCN3B	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
SCN3B	Brugada syndrome	ORPHA:130	UNKNOWN
SCN4A	Myotonia, potassium-aggravated	OMIM:608390	MENDELIAN
SCN4A	Myasthenic syndrome, congenital, 16	OMIM:614198	MENDELIAN
SCN4A	Hyperkalemic periodic paralysis	OMIM:170500	MENDELIAN
SCN4A	Hypokalemic periodic paralysis, type 1	OMIM:170400	MENDELIAN
SCN4A	Hypokalemic periodic paralysis, type 2	OMIM:613345	MENDELIAN
SCN4A	Paramyotonia congenita	OMIM:168300	MENDELIAN
SCN4A	Congenital myopathy 22A, classic	OMIM:620351	MENDELIAN
SCN4A	Congenital myopathy 22B, severe fetal	OMIM:620369	MENDELIAN
SCN4A	Paramyotonia congenita of Von Eulenburg	ORPHA:684	UNKNOWN
SCN4A	Hyperkalemic periodic paralysis	ORPHA:682	UNKNOWN
SCN4A	Hypokalemic periodic paralysis	ORPHA:681	UNKNOWN
SCN4A	Myotonia permanens	ORPHA:99735	UNKNOWN
SCN4A	Acetazolamide-responsive myotonia	ORPHA:99736	UNKNOWN
SCN4A	Myotonia fluctuans	ORPHA:99734	UNKNOWN
SCN4B	Long QT syndrome 10	OMIM:611819	MENDELIAN
SCN4B	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
SCN5A	Progressive familial heart block, type IA	OMIM:113900	MENDELIAN
SCN5A	Sudden infant death syndrome	OMIM:272120	POLYGENIC
SCN5A	Sick sinus syndrome 1	OMIM:608567	MENDELIAN
SCN5A	Atrial fibrillation, familial, 10	OMIM:614022	MENDELIAN
SCN5A	Long QT syndrome 3	OMIM:603830	MENDELIAN
SCN5A	Ventricular fibrillation, familial, 1	OMIM:603829	MENDELIAN
SCN5A	Cardiomyopathy, dilated, 1E	OMIM:601154	MENDELIAN
SCN5A	Brugada syndrome 1	OMIM:601144	MENDELIAN
SCN5A	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
SCN5A	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
SCN5A	Brugada syndrome	ORPHA:130	UNKNOWN
SCN5A	Hereditary progressive cardiac conduction defect	ORPHA:871	UNKNOWN
SCN5A	Isolated atrial standstill	ORPHA:1344	UNKNOWN
SCN7A	—		UNKNOWN
SCN8A	Cognitive impairment with or without cerebellar ataxia	OMIM:614306	MENDELIAN
SCN8A	Myoclonus, familial, 2	OMIM:618364	MENDELIAN
SCN8A	Seizures, benign familial infantile, 5	OMIM:617080	MENDELIAN
SCN8A	Epileptic encephalopathy, early infantile, 13	OMIM:614558	MENDELIAN
SCN8A	Self-limited infantile epilepsy	ORPHA:306	UNKNOWN
SCN8A	Myoclonic epilepsy of infancy	ORPHA:86909	UNKNOWN
SCN8A	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
SCN9A	Neuropathy, hereditary sensory and autonomic, type II	OMIM:201300	MENDELIAN
SCN9A	Indifference to pain, congenital, autosomal recessive	OMIM:243000	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
SCN9A	Erythermalgia, primary	OMIM:133020	MENDELIAN
SCN9A	Paroxysmal extreme pain disorder	OMIM:167400	MENDELIAN
SCN9A	Genetic epilepsy with febrile seizure plus	ORPHA:36387	UNKNOWN
SCN9A	Hereditary sensory and autonomic neuropathy type 2	ORPHA:970	UNKNOWN
SCN9A	Primary erythromelalgia	ORPHA:90026	UNKNOWN
SCN9A	Paroxysmal extreme pain disorder	ORPHA:46348	UNKNOWN
SCN9A	Dravet syndrome	ORPHA:33069	UNKNOWN
SCNN1A	Bronchiectasis with or without elevated sweat chloride 2	OMIM:613021	MENDELIAN
SCNN1A	Liddle syndrome 3	OMIM:618126	MENDELIAN
SCNN1A	Pseudohypoaldosteronism, type I, autosomal recessive	OMIM:264350	MENDELIAN
SCNN1A	Generalized pseudohypoaldosteronism type 1	ORPHA:171876	UNKNOWN
SCNN1A	Idiopathic bronchiectasis	ORPHA:60033	UNKNOWN
SCNN1A	Brugada syndrome	ORPHA:130	UNKNOWN
SCNN1A	Liddle syndrome	ORPHA:526	UNKNOWN
SCNN1B	Liddle syndrome 1	OMIM:177200	MENDELIAN
SCNN1B	Pseudohypoaldosteronism, type IB2, autosomal recessive	OMIM:620125	MENDELIAN
SCNN1B	Bronchiectasis with or without elevated sweat chloride 1	OMIM:211400	MENDELIAN
SCNN1B	Generalized pseudohypoaldosteronism type 1	ORPHA:171876	UNKNOWN
SCNN1B	Idiopathic bronchiectasis	ORPHA:60033	UNKNOWN
SCNN1B	Liddle syndrome	ORPHA:526	UNKNOWN
SCNN1D	—		UNKNOWN
SCNN1G	Bronchiectasis with or without elevated sweat chloride 3	OMIM:613071	MENDELIAN
SCNN1G	Pseudohypoaldosteronism, type IB3, autosomal recessive	OMIM:620126	MENDELIAN
SCNN1G	Liddle syndrome 2	OMIM:618114	MENDELIAN
SCNN1G	Generalized pseudohypoaldosteronism type 1	ORPHA:171876	UNKNOWN
SCNN1G	Idiopathic bronchiectasis	ORPHA:60033	UNKNOWN
SCNN1G	Liddle syndrome	ORPHA:526	UNKNOWN
SCO1	Mitochondrial complex IV deficiency, nuclear type 4	OMIM:619048	MENDELIAN
SCO1	Mitochondrial complex IV deficiency, nuclear type 1	OMIM:220110	MENDELIAN
SCO2	Mitochondrial complex IV deficiency, nuclear type 2	OMIM:604377	MENDELIAN
SCO2	Myopia 6	OMIM:608908	MENDELIAN
SCO2	Mitochondrial complex IV deficiency, nuclear type 1	OMIM:220110	MENDELIAN
SCO2	Autosomal recessive axonal Charcot-Marie-Tooth disease due to copper metabolism defect	ORPHA:521411	UNKNOWN
SCP2	Leukoencephalopathy with dystonia and motor neuropathy	OMIM:613724	MENDELIAN
SCRIB	—		UNKNOWN
SCT	—		UNKNOWN
SCUBE2	—		UNKNOWN
SDC3	Obesity	OMIM:601665	POLYGENIC
SDCCAG8	Bardet-Biedl syndrome 16	OMIM:615993	MENDELIAN
SDCCAG8	Senior-Loken syndrome 7	OMIM:613615	MENDELIAN
SDCCAG8	Senior-Loken syndrome	ORPHA:3156	UNKNOWN
SDCCAG8	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
SDHA	Neurodegeneration with ataxia and late-onset optic atrophy	OMIM:619259	MENDELIAN
SDHA	Parangangliomas 5	OMIM:614165	MENDELIAN
SDHA	Mitochondrial complex II deficiency	OMIM:252011	MENDELIAN
SDHA	Cardiomyopathy, dilated, 1GG	OMIM:613642	MENDELIAN
SDHA	Carney-Stratakis syndrome	ORPHA:97286	UNKNOWN
SDHA	Isolated succinate-CoQ reductase deficiency	ORPHA:3208	UNKNOWN
SDHA	Gastrointestinal stromal tumor	ORPHA:44890	UNKNOWN
SDHA	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
SDHA	Carney triad	ORPHA:139411	UNKNOWN
SDHA	Hereditary pheochromocytoma-paranganglioma	ORPHA:29072	UNKNOWN
SDHAF1	Mitochondrial complex II deficiency, nuclear type 2	OMIM:619166	MENDELIAN
SDHAF1	Isolated succinate-CoQ reductase deficiency	ORPHA:3208	UNKNOWN
SDHAF2	Parangangliomas 2	OMIM:601650	MENDELIAN
SDHAF2	Hereditary pheochromocytoma-paranganglioma	ORPHA:29072	UNKNOWN
SDHB	Carney-Stratakis syndrome	OMIM:606864	MENDELIAN
SDHB	Mitochondrial complex II deficiency, nuclear type 4	OMIM:619224	MENDELIAN
SDHB	Parangangliomas 4	OMIM:115310	MENDELIAN
SDHB	Gastrointestinal stromal tumor	OMIM:606764	MENDELIAN
SDHB	Carney-Stratakis syndrome	ORPHA:97286	UNKNOWN
SDHB	Isolated succinate-CoQ reductase deficiency	ORPHA:3208	UNKNOWN
SDHB	Gastrointestinal stromal tumor	ORPHA:44890	UNKNOWN
SDHB	Carney triad	ORPHA:139411	UNKNOWN
SDHB	Sporadic pheochromocytoma/secretory paraganglioma	ORPHA:276621	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
SDHB	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
SDHC	Carney-Stratakis syndrome	OMIM:606864	MENDELIAN
SDHC	Gastrointestinal stromal tumor	OMIM:606764	MENDELIAN
SDHC	Paragangliomas 3	OMIM:605373	MENDELIAN
SDHC	Carney-Stratakis syndrome	ORPHA:97286	UNKNOWN
SDHC	Gastrointestinal stromal tumor	ORPHA:44890	UNKNOWN
SDHC	Carney triad	ORPHA:139411	UNKNOWN
SDHC	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
SDHD	Carney-Stratakis syndrome	OMIM:606864	MENDELIAN
SDHD	Mitochondrial complex II deficiency, nuclear type 3	OMIM:619167	MENDELIAN
SDHD	Paragangliomas 1	OMIM:168000	MENDELIAN
SDHD	Carney-Stratakis syndrome	ORPHA:97286	UNKNOWN
SDHD	Isolated succinate-CoQ reductase deficiency	ORPHA:3208	UNKNOWN
SDHD	Carcinoid syndrome	ORPHA:100093	UNKNOWN
SDHD	Sporadic pheochromocytoma/secretory paraganglioma	ORPHA:276621	UNKNOWN
SDHD	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
SDR39U1	—		UNKNOWN
SEC23A	Cranio-lenticulosutural dysplasia	OMIM:607812	MENDELIAN
SEC23A	Cranio-lenticulosutural dysplasia	ORPHA:50814	UNKNOWN
SEC23B	Anemia, dyserythropoietic congenital, type II	OMIM:224100	MENDELIAN
SEC23B	Cowden syndrome 7	OMIM:616858	MENDELIAN
SEC24D	Cole-Carpenter syndrome 2	OMIM:616294	MENDELIAN
SEC24D	Cole-Carpenter syndrome	ORPHA:2050	UNKNOWN
SEC63	Polycystic liver disease 2	OMIM:617004	MENDELIAN
SEC63	Isolated polycystic liver disease	ORPHA:2924	UNKNOWN
SECISBP2	Thyroid hormone metabolism, abnormal	OMIM:609698	MENDELIAN
SECISBP2	Short stature-delayed bone age due to thyroid hormone metabolism deficiency	ORPHA:171706	UNKNOWN
SEL1L	Neurodevelopmental disorder with hypotonia, poor growth, dysmorphic facies, and agammaglobulinemia	OMIM:621068	MENDELIAN
SEL1L	Neurodevelopmental disorder with poor growth, absent speech, progressive ataxia, and dysmorphic facies	OMIM:621067	MENDELIAN
SELE	—		UNKNOWN
SELL	—		UNKNOWN
SELP	—		UNKNOWN
SELPLG	—		UNKNOWN
SEMA3A	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
SEMA3A	Hypogonadotropic hypogonadism 16 with or without anosmia	OMIM:614897	POLYGENIC
SEMA3A	Brugada syndrome	ORPHA:130	UNKNOWN
SEMA3A	Kallmann syndrome	ORPHA:478	UNKNOWN
SEMA3D	Hirschsprung disease	ORPHA:388	UNKNOWN
SEMA3E	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
SEMA3E	CHARGE syndrome	ORPHA:138	UNKNOWN
SEMA4A	Retinitis pigmentosa 35	OMIM:610282	MENDELIAN
SEMA4A	Cone-Rod dystrophy 10	OMIM:610283	MENDELIAN
SEMA4A	Cone rod dystrophy	ORPHA:1872	UNKNOWN
SEMA4A	Retinitis pigmentosa	ORPHA:791	UNKNOWN
SEMA4A	Familial colorectal cancer Type X	ORPHA:440437	UNKNOWN
SEMA4C	—		UNKNOWN
SEMA4D	Primary sclerosing cholangitis	ORPHA:171	UNKNOWN
SEMA4G	—		UNKNOWN
SEMA5A	Monosomy 5p syndrome	ORPHA:281	UNKNOWN
SEMA6D	—		UNKNOWN
SEMA7A	Cholestasis, progressive familial intrahepatic, 11	OMIM:619874	MENDELIAN
SEMG1	—		UNKNOWN
SEP15	—		UNKNOWN
SEPN1	—		UNKNOWN
SEPP1	—		UNKNOWN
SEPSECS	Pontocerebellar hypoplasia, type 2D	OMIM:613811	MENDELIAN
SEPSECS	Pontocerebellar hypoplasia type 2	ORPHA:2524	UNKNOWN
SEPT12	—		UNKNOWN
SEPT14	—		UNKNOWN
SEPT5	—		UNKNOWN
SEPT7	—		UNKNOWN
SEPT9	—		UNKNOWN
SERAC1	3-Methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	OMIM:614739	MENDELIAN
SERAC1	MEGDEL syndrome	ORPHA:352328	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
SERHL2	—		UNKNOWN
SERPINA1	Alpha-1-Antitrypsin deficiency	OMIM:613490	MENDELIAN
SERPINA1	Alpha-1-antitrypsin deficiency	ORPHA:60	UNKNOWN
SERPINA1	Cystic fibrosis	ORPHA:586	UNKNOWN
SERPINA10	—		UNKNOWN
SERPINA3	—		UNKNOWN
SERPINA6	Corticosteroid-binding globulin deficiency	OMIM:611489	MENDELIAN
SERPINB11	—		UNKNOWN
SERPINB3	—		UNKNOWN
SERPINB4	—		UNKNOWN
SERPINB5	—		UNKNOWN
SERPINB6	Deafness, autosomal recessive 91	OMIM:613453	MENDELIAN
SERPINB7	Palmoplantar keratoderma, Nagashima type	OMIM:615598	MENDELIAN
SERPINB7	Palmoplantar keratoderma, Nagashima type	ORPHA:140966	UNKNOWN
SERPINC1	Antithrombin III deficiency	OMIM:613118	MENDELIAN
SERPINC1	Hereditary thrombophilia due to congenital antithrombin deficiency	ORPHA:82	UNKNOWN
SERPIND1	Heparin cofactor II deficiency	OMIM:612356	MENDELIAN
SERPINE1	Plasminogen activator inhibitor-1 deficiency	OMIM:613329	MENDELIAN
SERPINE1	Congenital plasminogen activator inhibitor type 1 deficiency	ORPHA:465	UNKNOWN
SERPINF1	Osteogenesis imperfecta, type VI	OMIM:613982	MENDELIAN
SERPINF2	Plasmin inhibitor deficiency	OMIM:262850	MENDELIAN
SERPINF2	Congenital alpha2-antiplasmin deficiency	ORPHA:79	UNKNOWN
SERPING1	Angioedema, hereditary, 1	OMIM:106100	MENDELIAN
SERPING1	Complement component 4, partial deficiency of	OMIM:120790	MENDELIAN
SERPING1	Hereditary angioedema type 1	ORPHA:100050	UNKNOWN
SERPINH1	Preterm premature rupture of the membranes	OMIM:610504	POLYGENIC
SERPINH1	Osteogenesis imperfecta, type X	OMIM:613848	MENDELIAN
SERPINI1	Encephalopathy, familial, with neuroserpin inclusion bodies	OMIM:604218	MENDELIAN
SERPINI2	—		UNKNOWN
SERTAD1	—		UNKNOWN
SESN2	—		UNKNOWN
SETBP1	Schinzel-Giedion midface-retraction syndrome	OMIM:269150	MENDELIAN
SETBP1	Intellectual developmental disorder, autosomal dominant 29	OMIM:616078	MENDELIAN
SETBP1	Schinzel-Giedion syndrome	ORPHA:798	UNKNOWN
SETD1A	Epilepsy, early-onset, with or without developmental delay	OMIM:618832	MENDELIAN
SETD1A	Neurodevelopmental disorder with speech impairment and dysmorphic facies	OMIM:619056	MENDELIAN
SETD1B	Intellectual developmental disorder with seizures and language delay	OMIM:619000	MENDELIAN
SETD2	Intellectual developmental disorder, autosomal dominant 70	OMIM:620157	MENDELIAN
SETD2	Rabin-Pappas syndrome	OMIM:620155	MENDELIAN
SETD2	Luscan-Lumish syndrome	OMIM:616831	MENDELIAN
SETD5	Intellectual developmental disorder, autosomal dominant 23	OMIM:615761	MENDELIAN
SETD5	Distal deletion 3p syndrome	ORPHA:1620	UNKNOWN
SETD8	—		UNKNOWN
SETDB1	—		UNKNOWN
SETDB2	—		UNKNOWN
SETX	Amyotrophic lateral sclerosis 4, juvenile	OMIM:602433	MENDELIAN
SETX	Spinocerebellar ataxia, autosomal recessive 1	OMIM:606002	MENDELIAN
SETX	Spinocerebellar ataxia with axonal neuropathy type 2	ORPHA:64753	UNKNOWN
SETX	Amyotrophic lateral sclerosis type 4	ORPHA:357043	UNKNOWN
SEZ6	—		UNKNOWN
SEZ6L	—		UNKNOWN
SEZ6L2	—		UNKNOWN
SF1	—		UNKNOWN
SF3B1	Myelodysplastic syndrome	OMIM:614286	MENDELIAN
SF3B1	Acquired idiopathic sideroblastic anemia	ORPHA:75564	UNKNOWN
SF3B4	Acrofacial dysostosis 1, Nager type	OMIM:154400	MENDELIAN
SF3B4	Nager syndrome	ORPHA:245	UNKNOWN
SF3B4	Acrofacial dysostosis, RodrÃ-guez type	ORPHA:1788	UNKNOWN
SFRP1	—		UNKNOWN
SFTPA1	Interstitial lung disease 1	OMIM:619611	MENDELIAN
SFTPA1	Idiopathic pulmonary fibrosis	ORPHA:2032	UNKNOWN
SFTPA2	Pulmonary fibrosis, idiopathic	OMIM:178500	MENDELIAN
SFTPA2	Idiopathic pulmonary fibrosis	ORPHA:2032	UNKNOWN
SFTPB	Surfactant metabolism dysfunction, pulmonary, 1	OMIM:265120	MENDELIAN
SFTPB	Pediatric acute respiratory distress syndrome	ORPHA:685082	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
SFTPB	Neonatal acute respiratory distress syndrome due to SP-B deficiency	ORPHA:217563	UNKNOWN
SFTPC	Pulmonary fibrosis, idiopathic	OMIM:178500	MENDELIAN
SFTPC	Surfactant metabolism dysfunction, pulmonary, 2	OMIM:610913	MENDELIAN
SFTPC	Pediatric acute respiratory distress syndrome	ORPHA:685082	UNKNOWN
SFTPC	Idiopathic pulmonary fibrosis	ORPHA:2032	UNKNOWN
SFTPD	—		UNKNOWN
SFXN4	Combined oxidative phosphorylation deficiency 18	OMIM:615578	MENDELIAN
SGCA	Muscular dystrophy, limb-girdle, type 2D	OMIM:608099	MENDELIAN
SGCA	Alpha-sarcoglycan-related limb-girdle muscular dystrophy R3	ORPHA:62	UNKNOWN
SGCB	Muscular dystrophy, limb-girdle, type 2E	OMIM:604286	MENDELIAN
SGCB	Beta-sarcoglycan-related limb-girdle muscular dystrophy R4	ORPHA:119	UNKNOWN
SGCD	Muscular dystrophy, limb-girdle, type 2F	OMIM:601287	MENDELIAN
SGCD	Cardiomyopathy, dilated, 1L	OMIM:606685	MENDELIAN
SGCD	Delta-sarcoglycan-related limb-girdle muscular dystrophy R6	ORPHA:219	UNKNOWN
SGCD	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
SGCE	Dystonia-11, myoclonic	OMIM:159900	MENDELIAN
SGCE	Myoclonus-dystonia syndrome	ORPHA:36899	UNKNOWN
SGCG	Muscular dystrophy, limb-girdle, type 2C	OMIM:253700	MENDELIAN
SGCG	Gamma-sarcoglycan-related limb-girdle muscular dystrophy R5	ORPHA:353	UNKNOWN
SGK1	—		UNKNOWN
SGK110	—		UNKNOWN
SGK223	—		UNKNOWN
SGOL1	—		UNKNOWN
SGSH	Mucopolysaccharidosis type IIIA	OMIM:252900	MENDELIAN
SGSM3	Intellectual developmental disorder, autosomal recessive 84	OMIM:620401	MENDELIAN
SH2B1	Severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency	ORPHA:329249	UNKNOWN
SH2B1	Distal 16p11.2 microdeletion syndrome	ORPHA:261222	UNKNOWN
SH2B1	Proximal 16p11.2 microdeletion syndrome	ORPHA:261197	UNKNOWN
SH2B3	Erythrocytosis, familial, 1	OMIM:133100	MENDELIAN
SH2B3	Myelofibrosis with myeloid metaplasia, somatic	OMIM:254450	MENDELIAN
SH2B3	Thrombocythemia 1	OMIM:187950	MENDELIAN
SH2B3	Essential thrombocythemia	ORPHA:3318	UNKNOWN
SH2D1A	Lymphoproliferative syndrome, X-linked, 1	OMIM:308240	MENDELIAN
SH3BP2	Cherubism	OMIM:118400	MENDELIAN
SH3BP2	Cherubism	ORPHA:184	UNKNOWN
SH3GL1	Leukemia, acute myeloid	OMIM:601626	MENDELIAN
SH3PXD2B	Frank-ter Haar syndrome	OMIM:249420	MENDELIAN
SH3PXD2B	Frank-Ter Haar syndrome	ORPHA:137834	UNKNOWN
SH3TC2	Mononeuropathy of the median nerve, mild	OMIM:613353	MENDELIAN
SH3TC2	Charcot-Marie-Tooth disease, type 4C	OMIM:601596	MENDELIAN
SH3TC2	Charcot-Marie-Tooth disease type 4C	ORPHA:99949	UNKNOWN
SHANK1	—		UNKNOWN
SHANK3	Phelan-Mcdermid syndrome	OMIM:606232	MENDELIAN
SHANK3	Schizophrenia 15	OMIM:613950	POLYGENIC
SHARPIN	Autoinflammation with episodic fever and immune dysregulation	OMIM:620795	MENDELIAN
SHBG	—		UNKNOWN
SHFM1	—		UNKNOWN
SHH	Microphthalmia, isolated, with coloboma 5	OMIM:611638	MENDELIAN
SHH	Solitary median maxillary central incisor	OMIM:147250	MENDELIAN
SHH	Holoprosencephaly 3	OMIM:142945	MENDELIAN
SHH	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
SHH	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
SHH	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
SHH	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
SHH	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
SHH	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
SHH	Tibial hemimelia-polysyndactyly-triphalangeal thumb syndrome	ORPHA:988	UNKNOWN
SHH	Isolated radial hemimelia	ORPHA:93321	UNKNOWN
SHH	Syndactyly type 4	ORPHA:93405	UNKNOWN
SHMT1	—		UNKNOWN
SHOC1	Spermatogenic failure 75	OMIM:619949	MENDELIAN
SHOC1	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
SHOC2	Noonan syndrome-like with loose anagen hair 1	OMIM:607721	MENDELIAN
SHOC2	Noonan syndrome-like disorder with loose anagen hair	ORPHA:2701	UNKNOWN
SHOX	Langer mesomelic dysplasia	OMIM:249700	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
SHOX	Leri-Weill dyschondrosteosis	OMIM:127300	MENDELIAN
SHOX	Short stature, idiopathic familial	OMIM:300582	MENDELIAN
SHOX	LÃ©ri-Weill dyschondrosteosis	ORPHA:240	UNKNOWN
SHOX	Langer mesomelic dysplasia	ORPHA:2632	UNKNOWN
SHOX	SHOX-related short stature	ORPHA:314795	UNKNOWN
SHOX2	—		UNKNOWN
SHQ1	Dystonia 35, childhood-onset	OMIM:619921	MENDELIAN
SHQ1	Neurodevelopmental disorder with dystonia and seizures	OMIM:619922	MENDELIAN
SHQ1	Early-onset generalized limb-onset dystonia	ORPHA:256	UNKNOWN
SHROOM3	—		UNKNOWN
SHROOM4	X-linked intellectual disability, Stocco Dos Santos type	ORPHA:85288	UNKNOWN
SI	Sucrase-isomaltase deficiency, congenital	OMIM:222900	MENDELIAN
SI	Congenital sucrase-isomaltase deficiency	ORPHA:35122	UNKNOWN
SIGLEC12	—		UNKNOWN
SIGLEC14	—		UNKNOWN
SIGLEC16	—		UNKNOWN
SIGMAR1	Spinal muscular atrophy, distal, autosomal recessive, 2	OMIM:605726	MENDELIAN
SIGMAR1	Amyotrophic lateral sclerosis 16, juvenile	OMIM:614373	MENDELIAN
SIGMAR1	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
SIK1	Epileptic encephalopathy, early infantile, 30	OMIM:616341	MENDELIAN
SIK1	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
SIK1	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
SIK3	Spondyloepimetaphyseal dysplasia, Krakow type	OMIM:618162	MENDELIAN
SIL1	Marinesco-Sjogren syndrome	OMIM:248800	MENDELIAN
SIL1	Marinesco-SjÃ¶gren syndrome	ORPHA:559	UNKNOWN
SIM1	6q16 microdeletion syndrome	ORPHA:171829	UNKNOWN
SIM1	SIM1-related Prader-Willi-like syndrome	ORPHA:398079	UNKNOWN
SIM1	Obesity due to SIM1 deficiency	ORPHA:369873	UNKNOWN
SIM2	—		UNKNOWN
SIN3A	Witteveen-Kolk syndrome	OMIM:613406	MENDELIAN
SIN3A	SIN3-related intellectual disability syndrome due to a point mutation	ORPHA:500166	UNKNOWN
SIN3A	15q24 microdeletion syndrome	ORPHA:94065	UNKNOWN
SIN3B	SIN3-related intellectual disability syndrome due to a point mutation	ORPHA:500166	UNKNOWN
SIPA1	—		UNKNOWN
SIPA1L1	—		UNKNOWN
SIRT1	—		UNKNOWN
SIRT3	—		UNKNOWN
SIRT5	—		UNKNOWN
SIX1	Branchiootic syndrome 3	OMIM:608389	MENDELIAN
SIX1	Branchiootorenal syndrome 1	OMIM:113650	MENDELIAN
SIX1	Deafness, autosomal dominant 23	OMIM:605192	MENDELIAN
SIX1	Branchiootic syndrome	ORPHA:52429	UNKNOWN
SIX1	BOR syndrome	ORPHA:107	UNKNOWN
SIX2	SIX2-related frontonasal dysplasia	ORPHA:488437	UNKNOWN
SIX3	Holoprosencephaly 2	OMIM:157170	MENDELIAN
SIX3	Schizencephaly	OMIM:269160	MENDELIAN
SIX3	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
SIX3	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
SIX3	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
SIX3	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
SIX3	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
SIX3	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
SIX5	Branchiootorenal syndrome 2	OMIM:610896	MENDELIAN
SIX5	BOR syndrome	ORPHA:107	UNKNOWN
SIX6	Optic disc anomalies with retinal and/or macular dystrophy	OMIM:212550	MENDELIAN
SIX6	Microphthalmia, syndromic 3	OMIM:206900	MENDELIAN
SIX6	Isolated complete colobomatous microphthalmia	ORPHA:35612	UNKNOWN
SKI	Shprintzen-Goldberg craniosynostosis syndrome	OMIM:182212	MENDELIAN
SKI	Shprintzen-Goldberg syndrome	ORPHA:2462	UNKNOWN
SKI	1p36 deletion syndrome	ORPHA:1606	UNKNOWN
SKIV2L	—		UNKNOWN
SLA2	—		UNKNOWN
SLBP	—		UNKNOWN
SLC10A1	Hypercholanemia, familial 2	OMIM:619256	MENDELIAN
SLC10A2	Bile acid malabsorption, primary	OMIM:613291	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
SLC11A1	Cystic fibrosis	ORPHA:586	UNKNOWN
SLC11A2	Anemia, hypochromic microcytic, with iron overload 1	OMIM:206100	MENDELIAN
SLC12A1	Bartter syndrome, type 1, antenatal	OMIM:601678	MENDELIAN
SLC12A2	Delpire-Mcneill syndrome	OMIM:619083	MENDELIAN
SLC12A2	Kilquist syndrome	OMIM:619080	MENDELIAN
SLC12A2	Deafness, autosomal dominant 78	OMIM:619081	MENDELIAN
SLC12A3	Gitelman syndrome	OMIM:263800	MENDELIAN
SLC12A3	Gitelman syndrome	ORPHA:358	UNKNOWN
SLC12A5	Epilepsy, idiopathic generalized, susceptibility to, 14	OMIM:616685	POLYGENIC
SLC12A5	Epileptic encephalopathy, early infantile, 34	OMIM:616645	MENDELIAN
SLC12A5	Epilepsy of infancy with migrating focal seizures	ORPHA:293181	UNKNOWN
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	OMIM:218000	MENDELIAN
SLC12A6	Charcot-Marie-Tooth disease, axonal, type 2II	OMIM:620068	MENDELIAN
SLC12A6	Corpus callosum agenesis-neuronopathy syndrome	ORPHA:1496	UNKNOWN
SLC12A7	—		UNKNOWN
SLC13A2	—		UNKNOWN
SLC13A5	Epileptic encephalopathy, early infantile, 25, with amelogenesis imperfecta	OMIM:615905	MENDELIAN
SLC13A5	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
SLC13A5	Amelocerebrohypohidrotic syndrome	ORPHA:1946	UNKNOWN
SLC14A2	—		UNKNOWN
SLC15A1	—		UNKNOWN
SLC16A1	Hyperinsulinemic hypoglycemia, familial, 7	OMIM:610021	MENDELIAN
SLC16A1	Erythrocyte lactate transporter defect	OMIM:245340	MENDELIAN
SLC16A1	Monocarboxylate transporter 1 deficiency	OMIM:616095	MENDELIAN
SLC16A12	Cataract, juvenile, with microcornea and glucosuria	OMIM:612018	MENDELIAN
SLC16A2	Allan-Herndon-Dudley syndrome	OMIM:300523	MENDELIAN
SLC16A2	Allan-Herndon-Dudley syndrome	ORPHA:59	UNKNOWN
SLC16A3	—		UNKNOWN
SLC16A7	—		UNKNOWN
SLC17A1	—		UNKNOWN
SLC17A3	Gout susceptibility 4	OMIM:612671	POLYGENIC
SLC17A5	Salla disease	OMIM:604369	MENDELIAN
SLC17A5	Infantile sialic acid storage disease	OMIM:269920	MENDELIAN
SLC17A8	Deafness, autosomal dominant 25	OMIM:605583	MENDELIAN
SLC17A9	Porokeratosis 8, disseminated superficial Actinic type	OMIM:616063	MENDELIAN
SLC17A9	Disseminated superficial actinic porokeratosis	ORPHA:79152	UNKNOWN
SLC18A1	—		UNKNOWN
SLC19A1	Immunodeficiency 114, folate-responsive	OMIM:620603	MENDELIAN
SLC19A1	Megaloblastic anemia, folate-responsive	OMIM:601775	MENDELIAN
SLC19A2	Thiamine-Responsive megaloblastic anemia syndrome	OMIM:249270	MENDELIAN
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	ORPHA:49827	UNKNOWN
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	OMIM:607483	MENDELIAN
SLC19A3	Infantile spasms-psychomotor retardation-progressive brain atrophy-basal ganglia disease syndrome	ORPHA:263410	UNKNOWN
SLC1A1	Dicarboxylicamino aciduria	OMIM:222730	MENDELIAN
SLC1A1	Dicarboxylic aminoaciduria	ORPHA:2195	UNKNOWN
SLC1A2	Epileptic encephalopathy, early infantile, 41	OMIM:617105	MENDELIAN
SLC1A2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
SLC1A3	Episodic ataxia, type 6	OMIM:612656	MENDELIAN
SLC1A3	Episodic ataxia type 6	ORPHA:209967	UNKNOWN
SLC1A3	Alternating hemiplegia of childhood	ORPHA:2131	UNKNOWN
SLC1A5	—		UNKNOWN
SLC20A2	Basal ganglia calcification, idiopathic, 1	OMIM:213600	MENDELIAN
SLC20A2	Bilateral striopallidodentate calcinosis	ORPHA:1980	UNKNOWN
SLC22A1	—		UNKNOWN
SLC22A11	—		UNKNOWN
SLC22A12	Hypouricemia, renal, 1	OMIM:220150	MENDELIAN
SLC22A12	Hereditary renal hypouricemia	ORPHA:94088	UNKNOWN
SLC22A14	—		UNKNOWN
SLC22A18	—		UNKNOWN
SLC22A2	—		UNKNOWN
SLC22A25	—		UNKNOWN
SLC22A3	—		UNKNOWN
SLC22A4	Rheumatoid arthritis	OMIM:180300	POLYGENIC
SLC22A5	Carnitine deficiency, systemic primary	OMIM:212140	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
SLC22A5	Systemic primary carnitine deficiency	ORPHA:158	UNKNOWN
SLC22A6	—		UNKNOWN
SLC22A9	—		UNKNOWN
SLC23A1	—		UNKNOWN
SLC24A1	Night blindness, congenital stationary, type 1D	OMIM:613830	MENDELIAN
SLC24A2	—		UNKNOWN
SLC24A4	Amelogenesis imperfecta, type IIA5	OMIM:615887	MENDELIAN
SLC24A5	Albinism, oculocutaneous, type VI	OMIM:113750	MENDELIAN
SLC24A5	Oculocutaneous albinism type 6	ORPHA:370097	UNKNOWN
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria	OMIM:615182	MENDELIAN
SLC25A1	Myasthenic syndrome, congenital, 23, presynaptic	OMIM:618197	MENDELIAN
SLC25A12	Epileptic encephalopathy, early infantile, 39	OMIM:612949	MENDELIAN
SLC25A13	Citrullinemia, type II, neonatal-onset	OMIM:605814	MENDELIAN
SLC25A13	Citrullinemia, type II, adult-onset	OMIM:603471	MENDELIAN
SLC25A13	Neonatal intrahepatic cholestasis due to citrin deficiency	ORPHA:247598	UNKNOWN
SLC25A13	Citrullinemia type II	ORPHA:247585	UNKNOWN
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	OMIM:238970	MENDELIAN
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	ORPHA:415	UNKNOWN
SLC25A19	Microcephaly, Amish type	OMIM:607196	MENDELIAN
SLC25A19	Thiamine metabolism dysfunction syndrome 4 (bilateral striatal degenerationand progressive polyneuropathy type)	OMIM:613710	MENDELIAN
SLC25A19	Amish lethal microcephaly	ORPHA:99742	UNKNOWN
SLC25A20	Carnitine-acylcarnitine translocase deficiency	OMIM:212138	MENDELIAN
SLC25A20	Carnitine-acylcarnitine translocase deficiency	ORPHA:159	UNKNOWN
SLC25A21	Mitochondrial DNA depletion syndrome 18	OMIM:618811	MENDELIAN
SLC25A22	Epileptic encephalopathy, early infantile, 3	OMIM:609304	MENDELIAN
SLC25A22	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
SLC25A22	Epilepsy of infancy with migrating focal seizures	ORPHA:293181	UNKNOWN
SLC25A3	Mitochondrial phosphate carrier deficiency	OMIM:610773	MENDELIAN
SLC25A3	Cardiomyopathy-hypotonia-lactic acidosis syndrome	ORPHA:91130	UNKNOWN
SLC25A38	Anemia, sideroblastic, 2, pyridoxine-refractory	OMIM:205950	MENDELIAN
SLC25A39	—		UNKNOWN
SLC25A4	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2	OMIM:609283	MENDELIAN
SLC25A4	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type)	OMIM:615418	MENDELIAN
SLC25A4	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type), autosomal dominant	OMIM:617184	MENDELIAN
SLC25A4	Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome	ORPHA:1369	UNKNOWN
SLC25A4	Autosomal dominant progressive external ophthalmoplegia	ORPHA:254892	UNKNOWN
SLC25A43	—		UNKNOWN
SLC25A46	Neuropathy, hereditary motor and sensory, type VIB	OMIM:616505	MENDELIAN
SLC25A46	Pontocerebellar hypoplasia, type 1E	OMIM:619303	MENDELIAN
SLC25A46	Pontocerebellar hypoplasia type 1	ORPHA:2254	UNKNOWN
SLC25A5	—		UNKNOWN
SLC26A1	Nephrolithiasis, calcium oxalate	OMIM:167030	MENDELIAN
SLC26A1	Hypersulfaturia	OMIM:620372	MENDELIAN
SLC26A10	—		UNKNOWN
SLC26A2	Achondrogenesis, type IB	OMIM:600972	MENDELIAN
SLC26A2	Epiphyseal dysplasia, multiple, 4	OMIM:226900	MENDELIAN
SLC26A2	Atelosteogenesis, type II	OMIM:256050	MENDELIAN
SLC26A2	Diastrophic dysplasia	OMIM:222600	MENDELIAN
SLC26A2	Atelosteogenesis type II	ORPHA:56304	UNKNOWN
SLC26A2	Achondrogenesis type 1B	ORPHA:93298	UNKNOWN
SLC26A2	Diastrophic dysplasia	ORPHA:628	UNKNOWN
SLC26A2	Multiple epiphyseal dysplasia type 4	ORPHA:93307	UNKNOWN
SLC26A3	Diarrhea 1, secretory chloride, congenital	OMIM:214700	MENDELIAN
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	OMIM:600791	MENDELIAN
SLC26A4	Pendred syndrome	OMIM:274600	MENDELIAN
SLC26A4	Thyroid hypoplasia	ORPHA:95720	UNKNOWN
SLC26A4	Athyreosis	ORPHA:95713	UNKNOWN
SLC26A4	Pendred syndrome	ORPHA:705	UNKNOWN
SLC26A5	Deafness, autosomal recessive 61	OMIM:613865	MENDELIAN
SLC26A6	—		UNKNOWN
SLC26A8	Spermatogenic failure 3	OMIM:606766	MENDELIAN
SLC26A9	Cystic fibrosis	ORPHA:586	UNKNOWN
SLC27A1	—		UNKNOWN
SLC27A4	Ichthyosis prematurity syndrome	OMIM:608649	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
SLC27A4	Ichthyosis-prematurity syndrome	ORPHA:88621	UNKNOWN
SLC27A5	—		UNKNOWN
SLC28A1	Uridine-cytidineuria	OMIM:618477	MENDELIAN
SLC28A2	—		UNKNOWN
SLC28A3	—		UNKNOWN
SLC29A1	—		UNKNOWN
SLC29A2	—		UNKNOWN
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome	OMIM:602782	MENDELIAN
SLC29A3	H syndrome	ORPHA:168569	UNKNOWN
SLC29A3	Dysosteosclerosis	ORPHA:1782	UNKNOWN
SLC29A4	—		UNKNOWN
SLC2A1	Dystonia 9	OMIM:601042	MENDELIAN
SLC2A1	Glut1 deficiency syndrome 2	OMIM:612126	MENDELIAN
SLC2A1	Stomatin-deficient cryohydrocytosis with neurologic defects	OMIM:608885	MENDELIAN
SLC2A1	GLUT1 deficiency syndrome 1, infantile onset, severe	OMIM:606777	MENDELIAN
SLC2A1	Epilepsy, idiopathic generalized, susceptibility to, 12	OMIM:614847	POLYGENIC
SLC2A1	Paroxysmal exertion-induced dyskinesia	ORPHA:98811	UNKNOWN
SLC2A1	Epilepsy with myoclonic-atonic seizures	ORPHA:1942	UNKNOWN
SLC2A1	Hereditary cryohydrocytosis with reduced stomatin	ORPHA:168577	UNKNOWN
SLC2A1	Alternating hemiplegia of childhood	ORPHA:2131	UNKNOWN
SLC2A1	Classic glucose transporter type 1 deficiency syndrome	ORPHA:71277	UNKNOWN
SLC2A1	Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity	ORPHA:53583	UNKNOWN
SLC2A10	Arterial tortuosity syndrome	OMIM:208050	MENDELIAN
SLC2A10	Arterial tortuosity syndrome	ORPHA:3342	UNKNOWN
SLC2A2	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
SLC2A2	Fanconi-Bickel syndrome	OMIM:227810	MENDELIAN
SLC2A2	Fanconi-Bickel syndrome	ORPHA:2088	UNKNOWN
SLC2A4	—		UNKNOWN
SLC2A9	Hypouricemia, renal, 2	OMIM:612076	MENDELIAN
SLC2A9	Hereditary renal hypouricemia	ORPHA:94088	UNKNOWN
SLC30A10	Hypermanganesemia with dystonia 1	OMIM:613280	MENDELIAN
SLC30A10	Cirrhosis-dystonia-polycythemia-hypermanganesemia syndrome	ORPHA:309854	UNKNOWN
SLC30A2	Zinc deficiency, transient neonatal	OMIM:608118	MENDELIAN
SLC30A5	—		UNKNOWN
SLC30A8	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
SLC31A1	Neurodegeneration and seizures due to copper transport defect	OMIM:620306	MENDELIAN
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration	OMIM:614482	MENDELIAN
SLC33A1	Spastic paraplegia 42, autosomal dominant	OMIM:612539	MENDELIAN
SLC33A1	Autosomal dominant spastic paraplegia type 42	ORPHA:171863	UNKNOWN
SLC34A1	Hypercalcemia, infantile 2	OMIM:616963	MENDELIAN
SLC34A1	Fanconi renotubular syndrome 2	OMIM:613388	MENDELIAN
SLC34A1	Nephrolithiasis/osteoporosis, hypophosphatemic, 1	OMIM:612286	MENDELIAN
SLC34A1	Primary Fanconi renotubular syndrome	ORPHA:3337	UNKNOWN
SLC34A1	Hereditary hypophosphatemic rickets with hypercalciuria	ORPHA:157215	UNKNOWN
SLC34A2	Pulmonary alveolar microlithiasis	OMIM:265100	MENDELIAN
SLC34A2	Pulmonary alveolar microlithiasis	ORPHA:60025	UNKNOWN
SLC34A3	Hypophosphatemic rickets with hypercalciuria, hereditary	OMIM:241530	MENDELIAN
SLC34A3	Hereditary hypophosphatemic rickets with hypercalciuria	ORPHA:157215	UNKNOWN
SLC35A1	Congenital disorder of glycosylation, type II f	OMIM:603585	MENDELIAN
SLC35A1	SLC35A1-CDG	ORPHA:238459	UNKNOWN
SLC35A2	Congenital disorder of glycosylation, type II m	OMIM:300896	MENDELIAN
SLC35A2	SLC35A2-CDG	ORPHA:356961	UNKNOWN
SLC35A3	Arthrogryposis, impaired intellectual development, and seizures	OMIM:615553	MENDELIAN
SLC35A3	Autism spectrum disorder-epilepsy-arthrogryposis syndrome	ORPHA:370943	UNKNOWN
SLC35C1	Congenital disorder of glycosylation, type II c	OMIM:266265	MENDELIAN
SLC35C1	Leukocyte adhesion deficiency type II	ORPHA:99843	UNKNOWN
SLC35D1	Schneckenbecken dysplasia	OMIM:269250	MENDELIAN
SLC35D1	Schneckenbecken dysplasia	ORPHA:3144	UNKNOWN
SLC35D3	—		UNKNOWN
SLC35F3	—		UNKNOWN
SLC35G2	—		UNKNOWN
SLC36A2	Iminoglycinuria	OMIM:242600	MENDELIAN
SLC36A2	Glycinuria with or without oxalate urolithiasis	OMIM:138500	MENDELIAN
SLC36A2	Iminoglycinuria	ORPHA:42062	UNKNOWN
SLC37A4	Congenital disorder of glycosylation, type II w	OMIM:619525	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
SLC37A4	Glycogen storage disease Ib	OMIM:232220	MENDELIAN
SLC37A4	Glycogen storage disease Ic	OMIM:232240	MENDELIAN
SLC37A4	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib	ORPHA:79259	UNKNOWN
SLC38A8	Foveal hypoplasia 2	OMIM:609218	MENDELIAN
SLC39A12	—		UNKNOWN
SLC39A13	Ehlers-Danlos syndrome, spondylodysplastic type, 3	OMIM:612350	MENDELIAN
SLC39A13	SLC39A13-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:157965	UNKNOWN
SLC39A4	Acrodermatitis enteropathica, Zinc-Deficiency type	OMIM:201100	MENDELIAN
SLC39A4	Acrodermatitis enteropathica	ORPHA:37	UNKNOWN
SLC39A5	Myopia 24, autosomal dominant	OMIM:615946	MENDELIAN
SLC3A1	Cystinuria	OMIM:220100	MENDELIAN
SLC3A1	Hypotonia-cystinuria syndrome	ORPHA:163690	UNKNOWN
SLC3A1	2p21 microdeletion syndrome	ORPHA:163693	UNKNOWN
SLC40A1	Hemochromatosis, type 4	OMIM:606069	MENDELIAN
SLC41A1	Nephronophthisis-like nephropathy 2	OMIM:619468	MENDELIAN
SLC44A2	—		UNKNOWN
SLC45A1	Intellectual developmental disorder with neuropsychiatric features	OMIM:617532	MENDELIAN
SLC45A2	Albinism, oculocutaneous, type IV	OMIM:606574	MENDELIAN
SLC45A2	Oculocutaneous albinism type 4	ORPHA:79435	UNKNOWN
SLC46A1	Folate malabsorption, hereditary	OMIM:229050	MENDELIAN
SLC46A1	Hereditary folate malabsorption	ORPHA:90045	UNKNOWN
SLC47A1	—		UNKNOWN
SLC47A2	—		UNKNOWN
SLC4A1	Renal tubular acidosis, distal, autosomal dominant	OMIM:179800	MENDELIAN
SLC4A1	Renal tubular acidosis, distal, with hemolytic anemia	OMIM:611590	MENDELIAN
SLC4A1	Spherocytosis, type 4	OMIM:612653	MENDELIAN
SLC4A1	Ovalocytosis, hereditary hemolytic	OMIM:166900	MENDELIAN
SLC4A1	Cryohydrocytosis	OMIM:185020	MENDELIAN
SLC4A1	Dehydrated hereditary stomatocytosis	ORPHA:3202	UNKNOWN
SLC4A1	Southeast Asian ovalocytosis	ORPHA:98868	UNKNOWN
SLC4A1	Hereditary spherocytosis	ORPHA:822	UNKNOWN
SLC4A10	Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities	OMIM:620746	MENDELIAN
SLC4A11	Corneal endothelial dystrophy and perceptive deafness	OMIM:217400	MENDELIAN
SLC4A11	Corneal endothelial dystrophy, autosomal recessive	OMIM:217700	MENDELIAN
SLC4A11	Corneal dystrophy, Fuchs endothelial, 4	OMIM:613268	MENDELIAN
SLC4A11	Corneal dystrophy-perceptive deafness syndrome	ORPHA:1490	UNKNOWN
SLC4A11	Congenital hereditary endothelial dystrophy type II	ORPHA:293603	UNKNOWN
SLC4A11	Fuchs endothelial corneal dystrophy	ORPHA:98974	UNKNOWN
SLC4A2	Osteopetrosis, autosomal recessive 9	OMIM:620366	MENDELIAN
SLC4A3	Short QT syndrome 7	OMIM:620231	MENDELIAN
SLC4A3	Congenital short QT syndrome	ORPHA:51083	UNKNOWN
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities and mentalretardation	OMIM:604278	MENDELIAN
SLC4A5	—		UNKNOWN
SLC4A7	—		UNKNOWN
SLC52A1	Riboflavin deficiency	OMIM:615026	MENDELIAN
SLC52A2	Brown-Vialetto-Van laere syndrome 2	OMIM:614707	MENDELIAN
SLC52A3	Brown-Vialetto-Van Laere syndrome 1	OMIM:211530	MENDELIAN
SLC52A3	Bulbar palsy, progressive, of childhood	OMIM:211500	MENDELIAN
SLC5A1	Glucose/galactose malabsorption	OMIM:606824	MENDELIAN
SLC5A1	Glucose-galactose malabsorption	ORPHA:35710	UNKNOWN
SLC5A11	—		UNKNOWN
SLC5A2	Renal glucosuria	OMIM:233100	MENDELIAN
SLC5A2	Familial renal glucosuria	ORPHA:69076	UNKNOWN
SLC5A5	Thyroid hormonogenesis, genetic defect in, 1	OMIM:274400	MENDELIAN
SLC5A5	Familial thyroid dys hormonogenesis	ORPHA:95716	UNKNOWN
SLC5A7	Neuronopathy, distal hereditary motor, type VIIA	OMIM:158580	MENDELIAN
SLC5A7	Myasthenic syndrome, congenital, 20, presynaptic	OMIM:617143	MENDELIAN
SLC6A1	Myoclonic-atonic epilepsy	OMIM:616421	MENDELIAN
SLC6A1	Epilepsy with myoclonic-atonic seizures	ORPHA:1942	UNKNOWN
SLC6A1	Proximal 3p25.3 microdeletion syndrome	ORPHA:435638	UNKNOWN
SLC6A11	Proximal 3p25.3 microdeletion syndrome	ORPHA:435638	UNKNOWN
SLC6A12	—		UNKNOWN
SLC6A13	—		UNKNOWN
SLC6A14	Cystic fibrosis	ORPHA:586	UNKNOWN
SLC6A17	Intellectual developmental disorder, autosomal recessive 48	OMIM:616269	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
SLC6A17	Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome	ORPHA:457212	UNKNOWN
SLC6A18	Iminoglycinuria	ORPHA:42062	UNKNOWN
SLC6A19	Hartnup disorder	OMIM:234500	MENDELIAN
SLC6A19	Hartnup disease	ORPHA:2116	UNKNOWN
SLC6A19	Iminoglycinuria	ORPHA:42062	UNKNOWN
SLC6A2	Orthostatic intolerance	OMIM:604715	MENDELIAN
SLC6A20	Iminoglycinuria	ORPHA:42062	UNKNOWN
SLC6A3	Parkinsonism-Dystonia, infantile, 1	OMIM:613135	MENDELIAN
SLC6A3	Infantile dystonia-parkinsonism	ORPHA:238455	UNKNOWN
SLC6A4	Obsessive-Compulsive disorder 1	OMIM:164230	POLYGENIC
SLC6A5	Hyperekplexia 3	OMIM:614618	MENDELIAN
SLC6A5	Hereditary hyperekplexia	ORPHA:3197	UNKNOWN
SLC6A6	Hypotaurinemic retinal degeneration and cardiomyopathy	OMIM:145350	MENDELIAN
SLC6A8	Cerebral creatine deficiency syndrome 1	OMIM:300352	MENDELIAN
SLC6A8	X-linked creatine transporter deficiency	ORPHA:52503	UNKNOWN
SLC6A9	Scoliosis, isolated, susceptibility to, 6	OMIM:621428	POLYGENIC
SLC6A9	Glycine encephalopathy with normal serum glycine	OMIM:617301	MENDELIAN
SLC7A1	—		UNKNOWN
SLC7A10	—		UNKNOWN
SLC7A11	—		UNKNOWN
SLC7A14	Retinitis pigmentosa 68	OMIM:615725	MENDELIAN
SLC7A14	Retinitis pigmentosa	ORPHA:791	UNKNOWN
SLC7A2	—		UNKNOWN
SLC7A5	—		UNKNOWN
SLC7A7	Lysinuric protein intolerance	OMIM:222700	MENDELIAN
SLC7A7	Lysinuric protein intolerance	ORPHA:470	UNKNOWN
SLC7A9	Cystinuria	OMIM:220100	MENDELIAN
SLC8A1	—		UNKNOWN
SLC9A1	Lichtenstein-Knorr syndrome	OMIM:616291	MENDELIAN
SLC9A3	Diarrhea 8, secretory sodium, congenital	OMIM:616868	MENDELIAN
SLC9A3	Cystic fibrosis	ORPHA:586	UNKNOWN
SLC9A3R1	—		UNKNOWN
SLC9A6	Neurodegenerative disorder, X-linked, female-restricted, with parkinsonism and cognitive impairment	OMIM:301142	MENDELIAN
SLC9A6	Intellectual developmental disorder, X-linked syndromic, Christianson type	OMIM:300243	MENDELIAN
SLC9A6	Christianson syndrome	ORPHA:85278	UNKNOWN
SLCO1A2	—		UNKNOWN
SLCO1B1	Hyperbilirubinemia, Rotor type, digenic	OMIM:237450	MENDELIAN
SLCO1B1	Rotor syndrome	ORPHA:3111	UNKNOWN
SLCO1B3	Hyperbilirubinemia, Rotor type, digenic	OMIM:237450	MENDELIAN
SLCO1B3	Rotor syndrome	ORPHA:3111	UNKNOWN
SLCO1C1	—		UNKNOWN
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal dominant	OMIM:167100	MENDELIAN
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive 2	OMIM:614441	MENDELIAN
SLCO2A1	Pachydermoperiostosis	ORPHA:2796	UNKNOWN
SLCO2B1	—		UNKNOWN
SLCO5A1	—		UNKNOWN
SLFN5	—		UNKNOWN
SLIT1	—		UNKNOWN
SLIT2	—		UNKNOWN
SLIT3	—		UNKNOWN
SLITRK1	Trichotillomania	OMIM:613229	MENDELIAN
SLITRK1	Gilles de la tourette syndrome	OMIM:137580	MENDELIAN
SLITRK2	Intellectual developmental disorder, X-linked 111	OMIM:301107	MENDELIAN
SLITRK5	—		UNKNOWN
SLITRK6	Deafness and myopia	OMIM:221200	MENDELIAN
SLK	—		UNKNOWN
SLMAP	Brugada syndrome	ORPHA:130	UNKNOWN
SLTM	—		UNKNOWN
SLURP1	Meleda disease	OMIM:248300	MENDELIAN
SLURP1	Mal de Meleda	ORPHA:87503	UNKNOWN
SLX1A	—		UNKNOWN
SLX4	Fanconi anemia, complementation group P	OMIM:613951	MENDELIAN
SLX4	Fanconi anemia	ORPHA:84	UNKNOWN
SMAD1	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
SMAD2	Loeys-Dietz syndrome 6	OMIM:619656	MENDELIAN
SMAD2	Congenital heart defects, multiple types, 8, with or without heterotaxy	OMIM:619657	MENDELIAN
SMAD2	Loeys-Dietz syndrome	ORPHA:60030	UNKNOWN
SMAD2	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
SMAD3	Loeys-Dietz syndrome 3	OMIM:613795	MENDELIAN
SMAD3	Loeys-Dietz syndrome	ORPHA:60030	UNKNOWN
SMAD3	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
SMAD3	Aneurysm-osteoarthritis syndrome	ORPHA:284984	UNKNOWN
SMAD4	Juvenile polyposis syndrome	OMIM:174900	MENDELIAN
SMAD4	Myhre syndrome	OMIM:139210	MENDELIAN
SMAD4	Pancreatic cancer	OMIM:260350	MENDELIAN
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	OMIM:175050	MENDELIAN
SMAD4	Generalized juvenile polyposis/juvenile polyposis coli	ORPHA:329971	UNKNOWN
SMAD4	Myhre syndrome	ORPHA:2588	UNKNOWN
SMAD4	Familial pancreatic carcinoma	ORPHA:1333	UNKNOWN
SMAD4	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
SMAD4	Hereditary hemorrhagic telangiectasia	ORPHA:774	UNKNOWN
SMAD5	—		UNKNOWN
SMAD6	Craniosynostosis 7	OMIM:617439	POLYGENIC
SMAD6	Radioulnar synostosis	OMIM:179300	POLYGENIC
SMAD6	Aortic valve disease 2	OMIM:614823	MENDELIAN
SMAD6	Familial bicuspid aortic valve	ORPHA:402075	UNKNOWN
SMAD7	Colorectal cancer, susceptibility to, 3	OMIM:612229	POLYGENIC
SMAD9	Pulmonary hypertension, primary, 2	OMIM:615342	MENDELIAN
SMARCA1	—		UNKNOWN
SMARCA2	Nicolaides-Baraitser syndrome	OMIM:601358	MENDELIAN
SMARCA2	Blepharophimosis-impaired intellectual development syndrome	OMIM:619293	MENDELIAN
SMARCA2	Nicolaides-Baraitser syndrome	ORPHA:3051	UNKNOWN
SMARCA4	Coffin-Siris syndrome 4	OMIM:614609	MENDELIAN
SMARCA4	Rhabdoid tumor predisposition syndrome 2	OMIM:613325	POLYGENIC
SMARCA4	Otosclerosis 12	OMIM:620792	MENDELIAN
SMARCA4	Coffin-Siris syndrome	ORPHA:1465	UNKNOWN
SMARCAD1	Fingerprints, absence of	OMIM:136000	MENDELIAN
SMARCAD1	Huriez syndrome	OMIM:181600	MENDELIAN
SMARCAD1	Basan syndrome	OMIM:129200	MENDELIAN
SMARCAD1	Huriez syndrome	ORPHA:384	UNKNOWN
SMARCAD1	Absence of fingerprints-congenital milia syndrome	ORPHA:1658	UNKNOWN
SMARCAL1	Immunoosseous dysplasia, Schimke type	OMIM:242900	MENDELIAN
SMARCAL1	Schimke immuno-osseous dysplasia	ORPHA:1830	UNKNOWN
SMARCB1	Rhabdoid tumor predisposition syndrome 1	OMIM:609322	MENDELIAN
SMARCB1	Schwannomatosis	OMIM:162091	POLYGENIC
SMARCB1	Coffin-Siris syndrome 3	OMIM:614608	MENDELIAN
SMARCB1	Coffin-Siris syndrome	ORPHA:1465	UNKNOWN
SMARCB1	Atypical teratoid rhabdoid tumor	ORPHA:99966	UNKNOWN
SMARCB1	Full schwannomatosis	ORPHA:93921	UNKNOWN
SMARCB1	Meningioma	ORPHA:2495	UNKNOWN
SMARCC1	Hydrocephalus, congenital, 5, susceptibility to	OMIM:620241	POLYGENIC
SMARCC2	Coffin-Siris syndrome 8	OMIM:618362	MENDELIAN
SMARCC2	Coffin-Siris syndrome	ORPHA:1465	UNKNOWN
SMARCD1	Coffin-Siris syndrome 11	OMIM:618779	MENDELIAN
SMARCD1	Coffin-Siris syndrome	ORPHA:1465	UNKNOWN
SMARCE1	Meningioma, familial, susceptibility to	OMIM:607174	POLYGENIC
SMARCE1	Coffin-Siris syndrome 5	OMIM:616938	MENDELIAN
SMARCE1	Coffin-Siris syndrome	ORPHA:1465	UNKNOWN
SMARCE1	Meningioma	ORPHA:2495	UNKNOWN
SMC1A	Developmental and epileptic encephalopathy 85, with or without midline brain defects	OMIM:301044	MENDELIAN
SMC1A	Cornelia de Lange syndrome 2	OMIM:300590	MENDELIAN
SMC1A	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
SMC1A	Cornelia de Lange syndrome	ORPHA:199	UNKNOWN
SMC1A	Atypical Rett syndrome	ORPHA:3095	UNKNOWN
SMC1B	—		UNKNOWN
SMC3	Cornelia de Lange syndrome 3	OMIM:610759	MENDELIAN
SMC3	Cornelia de Lange syndrome	ORPHA:199	UNKNOWN
SMC5	Atelis syndrome 2	OMIM:620185	MENDELIAN
SMC6	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
SMCHD1	Bosma arhinia microphthalmia syndrome	OMIM:603457	MENDELIAN
SMCHD1	Facioscapulohumeral muscular dystrophy 2	OMIM:158901	MENDELIAN
SMCHD1	Facioscapulohumeral dystrophy	ORPHA:269	UNKNOWN
SMCHD1	Hyposmia-nasal and ocular hypoplasia-hypogonadotropic hypogonadism syndrome	ORPHA:2250	UNKNOWN
SMEK1	—		UNKNOWN
SMEK2	—		UNKNOWN
SMG1	—		UNKNOWN
SMG6	—		UNKNOWN
SMIM3	—		UNKNOWN
SMN1	Spinal muscular atrophy, type III	OMIM:253400	MENDELIAN
SMN1	Spinal muscular atrophy, type I	OMIM:253300	MENDELIAN
SMN1	Spinal muscular atrophy, type IV	OMIM:271150	MENDELIAN
SMN1	Spinal muscular atrophy, type II	OMIM:253550	MENDELIAN
SMN2	Spinal muscular atrophy, type III	OMIM:253400	POLYGENIC
SMNDC1	—		UNKNOWN
SMO	Pallister-Hall-like syndrome	OMIM:241800	MENDELIAN
SMO	Basal cell carcinoma, susceptibility to, 1	OMIM:605462	MENDELIAN
SMO	Curry-Jones syndrome	OMIM:601707	MENDELIAN
SMO	Hirschsprung disease	ORPHA:388	UNKNOWN
SMO	Curry-Jones syndrome	ORPHA:1553	UNKNOWN
SMO	Meningioma	ORPHA:2495	UNKNOWN
SMOC1	Microphthalmia with limb anomalies	OMIM:206920	MENDELIAN
SMOC1	Microphthalmia with limb anomalies	ORPHA:1106	UNKNOWN
SMOC2	Dentin dysplasia, type I, with microdontia and misshapen teeth	OMIM:125400	MENDELIAN
SMOX	—		UNKNOWN
SMPD1	Niemann-pick disease, type B	OMIM:607616	MENDELIAN
SMPD1	Niemann-Pick disease, type A	OMIM:257200	MENDELIAN
SMPD1	Chronic visceral acid sphingomyelinase deficiency	ORPHA:77293	UNKNOWN
SMPD1	Infantile neurovisceral acid sphingomyelinase deficiency	ORPHA:77292	UNKNOWN
SMPD1	Chronic neurovisceral acid sphingomyelinase deficiency	ORPHA:618891	UNKNOWN
SMPD3	—		UNKNOWN
SMPX	Deafness, X-linked 4	OMIM:300066	MENDELIAN
SMPX	Myopathy, distal, 7, adult-onset, X-linked	OMIM:301075	MENDELIAN
SMS	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type	OMIM:309583	MENDELIAN
SMS	X-linked intellectual disability, Snyder type	ORPHA:3063	UNKNOWN
SMUG1	—		UNKNOWN
SMYD3	—		UNKNOWN
SNAI2	Piebaldism	ORPHA:2884	UNKNOWN
SNAI2	Waardenburg syndrome type 2	ORPHA:895	UNKNOWN
SNAP25	Myasthenic syndrome, congenital, 18	OMIM:616330	MENDELIAN
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	OMIM:609528	MENDELIAN
SNAP29	CEDNIK syndrome	ORPHA:66631	UNKNOWN
SNAPC4	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction	OMIM:620515	MENDELIAN
SNAPC5	—		UNKNOWN
SNCA	Parkinson disease 4	OMIM:605543	MENDELIAN
SNCA	Dementia, lewy body	OMIM:127750	MENDELIAN
SNCA	Parkinson disease 1, autosomal dominant	OMIM:168601	MENDELIAN
SNCA	Young-onset Parkinson disease	ORPHA:2828	UNKNOWN
SNCA	Parkinsonian-pyramidal syndrome	ORPHA:171695	UNKNOWN
SNCA	Hereditary late-onset Parkinson disease	ORPHA:411602	UNKNOWN
SNCAIP	Parkinson disease, late-onset	OMIM:168600	MENDELIAN
SNCB	Dementia, lewy body	OMIM:127750	MENDELIAN
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism	OMIM:614501	MENDELIAN
SNORD115-1	Prader-Willi syndrome	OMIM:176270	MENDELIAN
SNORD116-1	Prader-Willi syndrome	OMIM:176270	MENDELIAN
SNORD116-10	—		UNKNOWN
SNORD50A	—		UNKNOWN
SNRK	—		UNKNOWN
SNRNP200	Retinitis pigmentosa 33	OMIM:610359	MENDELIAN
SNRNP200	Retinitis pigmentosa	ORPHA:791	UNKNOWN
SNRPB	Cerebrocostomandibular syndrome	OMIM:117650	MENDELIAN
SNRPB	Cerebrocostomandibular syndrome	ORPHA:1393	UNKNOWN
SNRPE	Hypotrichosis 11	OMIM:615059	MENDELIAN
SNRPE	Hypotrichosis simplex	ORPHA:55654	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
SNRPN	Autism susceptibility 1	OMIM:209850	MENDELIAN
SNRPN	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2	ORPHA:177904	UNKNOWN
SNRPN	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1	ORPHA:177901	UNKNOWN
SNRPN	Prader-Willi syndrome due to imprinting mutation	ORPHA:177910	UNKNOWN
SNRPN	Prader-Willi syndrome due to translocation	ORPHA:177907	UNKNOWN
SNRPN	Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15	ORPHA:98754	UNKNOWN
SNRPN	Angelman syndrome due to imprinting defect in 15q11-q13	ORPHA:411515	UNKNOWN
SNTA1	Long QT syndrome 12	OMIM:612955	MENDELIAN
SNTB1	—		UNKNOWN
SNTG1	—		UNKNOWN
SNTG2	—		UNKNOWN
SNURF	—		UNKNOWN
SNX1	—		UNKNOWN
SNX10	Osteopetrosis, autosomal recessive 8	OMIM:615085	MENDELIAN
SNX10	Autosomal recessive malignant osteopetrosis	ORPHA:667	UNKNOWN
SNX14	Spinocerebellar ataxia, autosomal recessive 20	OMIM:616354	MENDELIAN
SNX14	Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome	ORPHA:397709	UNKNOWN
SNX19	—		UNKNOWN
SNX3	—		UNKNOWN
SNX5	—		UNKNOWN
SNX8	—		UNKNOWN
SOAT1	—		UNKNOWN
SOBP	Impaired intellectual development, anterior maxillary protrusion, and strabismus	OMIM:613671	MENDELIAN
SOCS1	Autoinflammatory syndrome, familial, with or without immunodeficiency	OMIM:619375	MENDELIAN
SOCS3	—		UNKNOWN
SOD1	Amyotrophic lateral sclerosis 1	OMIM:105400	MENDELIAN
SOD1	Spastic tetraplegia and axial hypotonia, progressive	OMIM:618598	MENDELIAN
SOD1	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
SOD3	—		UNKNOWN
SOGA3	—		UNKNOWN
SOHLH1	Ovarian dysgenesis 5	OMIM:617690	MENDELIAN
SOHLH1	Spermatogenic failure 32	OMIM:618115	MENDELIAN
SOHLH1	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
SON	Zttk syndrome	OMIM:617140	MENDELIAN
SON	ZTTK syndrome	ORPHA:500150	UNKNOWN
SORBS1	—		UNKNOWN
SORCS1	—		UNKNOWN
SORD	Sorbitol dehydrogenase deficiency with peripheral neuropathy	OMIM:618912	MENDELIAN
SORD	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
SORL1	Early-onset autosomal dominant Alzheimer disease	ORPHA:1020	UNKNOWN
SOS1	Fibromatosis, gingival, 1	OMIM:135300	MENDELIAN
SOS1	Noonan syndrome 4	OMIM:610733	MENDELIAN
SOS1	Hereditary gingival fibromatosis	ORPHA:2024	UNKNOWN
SOS1	Noonan syndrome	ORPHA:648	UNKNOWN
SOS2	Noonan syndrome 9	OMIM:616559	MENDELIAN
SOS2	Noonan syndrome	ORPHA:648	UNKNOWN
SOST	van Buchem disease	OMIM:239100	MENDELIAN
SOST	Craniodiaphyseal dysplasia, autosomal dominant	OMIM:122860	MENDELIAN
SOST	Sclerosteosis 1	OMIM:269500	MENDELIAN
SOST	Sclerosteosis	ORPHA:3152	UNKNOWN
SOST	Craniodiaphyseal dysplasia	ORPHA:1513	UNKNOWN
SOST	Hyperostosis corticalis generalisata	ORPHA:3416	UNKNOWN
SOX10	Waardenburg syndrome, type 2E	OMIM:611584	MENDELIAN
SOX10	Peripheral demyelinating neuropathy, central dysmyelination, Waardenburg syndrome, and Hirschsprung disease	OMIM:609136	MENDELIAN
SOX10	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
SOX10	Waardenburg syndrome, type 4C	OMIM:613266	MENDELIAN
SOX10	Waardenburg-Shah syndrome	ORPHA:897	UNKNOWN
SOX10	Waardenburg syndrome type 2	ORPHA:895	UNKNOWN
SOX10	Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease	ORPHA:163746	UNKNOWN
SOX10	Kallmann syndrome	ORPHA:478	UNKNOWN
SOX11	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism	OMIM:615866	MENDELIAN
SOX11	Coffin-Siris syndrome	ORPHA:1465	UNKNOWN
SOX17	Pulmonary hypertension, primary, 7	OMIM:621248	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
SOX17	Vesicoureteral reflux 3	OMIM:613674	MENDELIAN
SOX18	Hypotrichosis-Lymphedema-Telangiectasia-Renal defect syndrome	OMIM:137940	MENDELIAN
SOX18	Hypotrichosis-lymphedema-telangiectasia syndrome	OMIM:607823	MENDELIAN
SOX18	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome	ORPHA:69735	UNKNOWN
SOX2	Microphthalmia, syndromic 3	OMIM:206900	MENDELIAN
SOX2	Septo-optic dysplasia spectrum	ORPHA:3157	UNKNOWN
SOX2	Anophthalmia/microphthalmia-esophageal atresia syndrome	ORPHA:77298	UNKNOWN
SOX2	Isolated complete colobomatous microphthalmia	ORPHA:35612	UNKNOWN
SOX3	Panhypopituitarism, X-linked	OMIM:312000	MENDELIAN
SOX3	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency	OMIM:300123	MENDELIAN
SOX3	46,XX testicular difference of sex development	ORPHA:393	UNKNOWN
SOX3	Non-acquired panhypopituitarism	ORPHA:90695	UNKNOWN
SOX3	Septo-optic dysplasia spectrum	ORPHA:3157	UNKNOWN
SOX3	X-linked intellectual disability with isolated growth hormone deficiency	ORPHA:67045	UNKNOWN
SOX4	Coffin-Siris syndrome 10	OMIM:618506	MENDELIAN
SOX4	Coffin-Siris syndrome	ORPHA:1465	UNKNOWN
SOX5	Lamb-Shaffer syndrome	OMIM:616803	MENDELIAN
SOX5	Developmental and speech delay due to SOX5 deficiency	ORPHA:313892	UNKNOWN
SOX5	Large/giant congenital melanocytic nevus	ORPHA:626	UNKNOWN
SOX6	Tolchin-Le Caignec syndrome	OMIM:618971	MENDELIAN
SOX7	—		UNKNOWN
SOX8	—		UNKNOWN
SOX9	46,XY sex reversal 10	OMIM:616425	MENDELIAN
SOX9	Campomelic dysplasia	OMIM:114290	MENDELIAN
SOX9	46,XX sex reversal 2	OMIM:278850	MENDELIAN
SOX9	46,XX testicular difference of sex development	ORPHA:393	UNKNOWN
SOX9	46,XY partial gonadal dysgenesis	ORPHA:251510	UNKNOWN
SOX9	46,XY complete gonadal dysgenesis	ORPHA:242	UNKNOWN
SOX9	Campomelic dysplasia	ORPHA:140	UNKNOWN
SOX9	Isolated Pierre Robin sequence	ORPHA:718	UNKNOWN
SOX9	46,XX ovotesticular difference of sex development	ORPHA:2138	UNKNOWN
SP100	—		UNKNOWN
SP110	Hepatic venoocclusive disease with immunodeficiency	OMIM:235550	MENDELIAN
SP110	Hepatic veno-occlusive disease-immunodeficiency syndrome	ORPHA:79124	UNKNOWN
SP7	Osteogenesis imperfecta, type XII	OMIM:613849	MENDELIAN
SP7	Craniodiaphyseal dysplasia	ORPHA:1513	UNKNOWN
SP8	—		UNKNOWN
SPAG1	Ciliary dyskinesia, primary, 28	OMIM:615505	MENDELIAN
SPAG1	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
SPAG16	—		UNKNOWN
SPAG17	Spermatogenic failure 55	OMIM:619380	MENDELIAN
SPAG17	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
SPANXN1	—		UNKNOWN
SPANXN5	—		UNKNOWN
SPAST	Spastic paraplegia 4, autosomal dominant	OMIM:182601	MENDELIAN
SPAST	Autosomal dominant spastic paraplegia type 4	ORPHA:100985	UNKNOWN
SPATA13	—		UNKNOWN
SPATA16	Spermatogenic failure 6	OMIM:102530	MENDELIAN
SPATA17	—		UNKNOWN
SPATA20	—		UNKNOWN
SPATA21	—		UNKNOWN
SPATA22	Premature ovarian failure 25	OMIM:621002	MENDELIAN
SPATA22	Spermatogenic failure 96	OMIM:621001	MENDELIAN
SPATA31C1	—		UNKNOWN
SPATA31D1	—		UNKNOWN
SPATA32	—		UNKNOWN
SPATA5L1	—		UNKNOWN
SPATA7	Leber congenital amaurosis-3 (LCA3)/Retinitis pigmentosa, juvenile, autosomal recessive	OMIM:604232	MENDELIAN
SPATA7	Leber congenital amaurosis	ORPHA:65	UNKNOWN
SPATA7	Retinitis pigmentosa	ORPHA:791	UNKNOWN
SPATA7	Severe early-childhood-onset retinal dystrophy	ORPHA:364055	UNKNOWN
SPDL1	—		UNKNOWN
SPECC1	—		UNKNOWN
SPECC1L	Teebi hypertelorism syndrome 1	OMIM:145420	MENDELIAN
SPECC1L	Facial clefting, oblique, 1	OMIM:600251	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
SPECC1L	SPECC1L-related hypertelorism syndrome	ORPHA:1519	UNKNOWN
SPEF2	Spermatogenic failure 43	OMIM:618751	MENDELIAN
SPEF2	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
SPEG	Centronuclear myopathy 5	OMIM:615959	MENDELIAN
SPEG	Autosomal recessive centronuclear myopathy	ORPHA:169186	UNKNOWN
SPEN	Radio-Tartaglia syndrome	OMIM:619312	MENDELIAN
SPEN	1p36 deletion syndrome	ORPHA:1606	UNKNOWN
SPERT	—		UNKNOWN
SPG11	Spastic paraplegia 11, autosomal recessive	OMIM:604360	MENDELIAN
SPG11	Amyotrophic lateral sclerosis 5, juvenile	OMIM:602099	MENDELIAN
SPG11	Charcot-Marie-Tooth disease, axonal, type 2X	OMIM:616668	MENDELIAN
SPG11	Autosomal recessive spastic paraplegia type 11	ORPHA:2822	UNKNOWN
SPG11	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
SPG20	—		UNKNOWN
SPG21	Mast syndrome	OMIM:248900	MENDELIAN
SPG21	Autosomal recessive spastic paraplegia type 21	ORPHA:101001	UNKNOWN
SPG7	Spastic paraplegia 7, autosomal recessive	OMIM:607259	MENDELIAN
SPG7	Spastic paraplegia type 7	ORPHA:99013	UNKNOWN
SPG7	Primary lateral sclerosis	ORPHA:35689	UNKNOWN
SPGF2	—		UNKNOWN
SPI1	Agammaglobulinemia 10, autosomal dominant	OMIM:619707	MENDELIAN
SPI1	Autosomal non-syndromic agammaglobulinemia	ORPHA:33110	UNKNOWN
SPIDR	Ovarian dysgenesis 9	OMIM:619665	MENDELIAN
SPIDR	46,XX gonadal dysgenesis	ORPHA:243	UNKNOWN
SPINK1	Pancreatitis, hereditary	OMIM:167800	MENDELIAN
SPINK1	Tropical calcific pancreatitis	OMIM:608189	MENDELIAN
SPINK1	Tropical pancreatitis	ORPHA:103918	UNKNOWN
SPINK2	Spermatogenic failure 29	OMIM:618091	MENDELIAN
SPINK2	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
SPINK5	Netherton syndrome	OMIM:256500	MENDELIAN
SPINK5	Netherton syndrome	ORPHA:634	UNKNOWN
SPINT2	Diarrhea 3, secretory sodium, congenital, with or without other congenital anomalies	OMIM:270420	MENDELIAN
SPO11	—		UNKNOWN
SPOCD1	—		UNKNOWN
SPOCK1	—		UNKNOWN
SPOP	Nabais Sa-de Vries syndrome, type 1	OMIM:618828	MENDELIAN
SPOP	Nabais Sa-de vries syndrome, type 2	OMIM:618829	MENDELIAN
SPP1	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
SPP1	Pediatric systemic lupus erythematosus	ORPHA:93552	UNKNOWN
SPP2	—		UNKNOWN
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	OMIM:612716	MENDELIAN
SPR	Dopa-responsive dystonia due to sepiapterin reductase deficiency	ORPHA:70594	UNKNOWN
SPRED1	Legius syndrome	OMIM:611431	MENDELIAN
SPRED1	Legius syndrome	ORPHA:137605	UNKNOWN
SPRED2	Noonan syndrome 14	OMIM:619745	MENDELIAN
SPRED2	Noonan syndrome	ORPHA:648	UNKNOWN
SPRN	—		UNKNOWN
SPRR3	—		UNKNOWN
SPRTN	Ruijs-Aalfs syndrome	OMIM:616200	MENDELIAN
SPRY2	Iga nephropathy, susceptibility to, 3	OMIM:616818	POLYGENIC
SPRY3	—		UNKNOWN
SPRY4	Hypogonadotropic hypogonadism 17 with or without anosmia	OMIM:615266	MENDELIAN
SPRY4	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
SPRY4	Kallmann syndrome	ORPHA:478	UNKNOWN
SPRY4	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
SPSB1	—		UNKNOWN
SPTA1	Spherocytosis, autosomal recessive	OMIM:270970	MENDELIAN
SPTA1	Elliptocytosis 2	OMIM:130600	MENDELIAN
SPTA1	Pyropoikilocytosis, hereditary	OMIM:266140	MENDELIAN
SPTA1	Hereditary elliptocytosis	ORPHA:288	UNKNOWN
SPTA1	Hereditary spherocytosis	ORPHA:822	UNKNOWN
SPTAN1	Epileptic encephalopathy, early infantile, 5	OMIM:613477	MENDELIAN
SPTAN1	Neuronopathy, distal hereditary motor, autosomal dominant 11	OMIM:620528	MENDELIAN
SPTAN1	Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia	OMIM:620538	MENDELIAN
SPTAN1	Developmental delay with or without epilepsy	OMIM:620540	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
SPTAN1	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
SPTB	Elliptocytosis-3	OMIM:617948	MENDELIAN
SPTB	Spherocytosis, type 2	OMIM:616649	MENDELIAN
SPTB	Hereditary elliptocytosis	ORPHA:288	UNKNOWN
SPTB	Hereditary spherocytosis	ORPHA:822	UNKNOWN
SPTBN1	Developmental delay, impaired speech, and behavioral abnormalities	OMIM:619475	MENDELIAN
SPTBN2	Spinocerebellar ataxia, autosomal recessive 14	OMIM:615386	MENDELIAN
SPTBN2	Spinocerebellar ataxia 5	OMIM:600224	MENDELIAN
SPTBN2	Spectrin-associated autosomal recessive cerebellar ataxia	ORPHA:352403	UNKNOWN
SPTBN2	Spinocerebellar ataxia type 5	ORPHA:98766	UNKNOWN
SPTBN5	—		UNKNOWN
SPTLC1	Amyotrophic lateral sclerosis 27, juvenile	OMIM:620285	MENDELIAN
SPTLC1	Neuropathy, hereditary sensory and autonomic, type IA	OMIM:162400	MENDELIAN
SPTLC1	Hereditary sensory and autonomic neuropathy type 1	ORPHA:36386	UNKNOWN
SPTLC1	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
SPTLC2	Neuropathy, hereditary sensory and autonomic, type IC	OMIM:613640	MENDELIAN
SPTLC2	Hereditary sensory and autonomic neuropathy type 1	ORPHA:36386	UNKNOWN
SPTLC2	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
SQSTM1	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3	OMIM:616437	MENDELIAN
SQSTM1	Paget disease of bone 3	OMIM:167250	MENDELIAN
SQSTM1	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	OMIM:617145	MENDELIAN
SQSTM1	Myopathy, distal, with rimmed vacuoles	OMIM:617158	MENDELIAN
SQSTM1	Behavioral variant of frontotemporal dementia	ORPHA:275864	UNKNOWN
SQSTM1	Frontotemporal dementia with motor neuron disease	ORPHA:275872	UNKNOWN
SQSTM1	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
SQSTM1	Distal myopathy, Welander type	ORPHA:603	UNKNOWN
SRC	Thrombocytopenia 6	OMIM:616937	MENDELIAN
SRC	Colorectal cancer	OMIM:114500	MENDELIAN
SRCAP	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities	OMIM:619595	MENDELIAN
SRCAP	Floating-Harbor syndrome	OMIM:136140	MENDELIAN
SRCAP	Floating-Harbor syndrome	ORPHA:2044	UNKNOWN
SRD5A2	Pseudovaginal perineoscrotal hypospadias	OMIM:264600	MENDELIAN
SRD5A2	46,XY difference of sex development due to 5-alpha-reductase 2 deficiency	ORPHA:753	UNKNOWN
SRD5A3	Kahrizi syndrome	OMIM:612713	MENDELIAN
SRD5A3	Congenital disorder of glycosylation, type IQ	OMIM:612379	MENDELIAN
SRD5A3	SRD5A3-CDG	ORPHA:324737	UNKNOWN
SREBF1	Mucoepithelial dysplasia, hereditary	OMIM:158310	MENDELIAN
SREBF1	Ichthyosis, follicular, with atrichia and photophobia syndrome 2	OMIM:619016	MENDELIAN
SREBF1	Hirschsprung disease	ORPHA:388	UNKNOWN
SREBF2	—		UNKNOWN
SREK1	—		UNKNOWN
SRGAP2	—		UNKNOWN
SRI	—		UNKNOWN
SRP72	Bone marrow failure syndrome 1	OMIM:614675	MENDELIAN
SRPRA	—		UNKNOWN
SRPX	—		UNKNOWN
SRPX2	Rolandic epilepsy, impaired intellectual development, and speech dyspraxia	OMIM:300643	MENDELIAN
SRPX2	Bilateral perisylvian polymicrogyria	ORPHA:98889	UNKNOWN
SRPX2	Rolandic epilepsy-speech dyspraxia syndrome	ORPHA:163721	UNKNOWN
SRPX2	Self-limited epilepsy with centrotemporal spikes	ORPHA:1945	UNKNOWN
SRR	—		UNKNOWN
SRRM2	Intellectual developmental disorder, autosomal dominant 72	OMIM:620439	MENDELIAN
SRSF1	Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities	OMIM:620489	MENDELIAN
SRSF2	Aggressive systemic mastocytosis	ORPHA:98850	UNKNOWN
SRSF2	Systemic mastocytosis with associated hematologic neoplasm	ORPHA:98849	UNKNOWN
SRY	46XY sex reversal 1	OMIM:400044	MENDELIAN
SRY	46XX sex reversal 1	OMIM:400045	MENDELIAN
SRY	46,XX testicular difference of sex development	ORPHA:393	UNKNOWN
SRY	46,XY partial gonadal dysgenesis	ORPHA:251510	UNKNOWN
SRY	46,XY complete gonadal dysgenesis	ORPHA:242	UNKNOWN
SRY	46,XX ovotesticular difference of sex development	ORPHA:2138	UNKNOWN
SRY	45,X/46,XY mixed gonadal dysgenesis	ORPHA:1772	UNKNOWN
SS18L1	—		UNKNOWN
SSBP1	Optic atrophy with negative electroretinograms	OMIM:165510	MENDELIAN
SSH1	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
SSPN	—		UNKNOWN
SSR4	Congenital disorder of glycosylation, type Iy	OMIM:300934	MENDELIAN
SSR4	SSR4-CDG	ORPHA:370927	UNKNOWN
SST	—		UNKNOWN
SSTR2	—		UNKNOWN
SSTR5	—		UNKNOWN
SSX1	Spermatogenic failure, X-linked, 5	OMIM:301099	MENDELIAN
SSX1	Sarcoma, synovial	OMIM:300813	MENDELIAN
SSX7	—		UNKNOWN
ST14	Ichthyosis, congenital, autosomal recessive 11	OMIM:602400	MENDELIAN
ST14	Ichthyosis-hypotrichosis syndrome	ORPHA:91132	UNKNOWN
ST3GAL1	—		UNKNOWN
ST3GAL2	—		UNKNOWN
ST3GAL3	Developmental and epileptic encephalopathy 15	OMIM:615006	MENDELIAN
ST3GAL3	Intellectual developmental disorder, autosomal recessive 12	OMIM:611090	MENDELIAN
ST3GAL4	—		UNKNOWN
ST3GAL5	Salt and pepper developmental regression syndrome	OMIM:609056	MENDELIAN
ST3GAL6	—		UNKNOWN
ST5	—		UNKNOWN
ST6GAL1	—		UNKNOWN
ST6GAL2	—		UNKNOWN
ST6GALNAC1	—		UNKNOWN
ST6GALNAC2	—		UNKNOWN
ST6GALNAC3	—		UNKNOWN
ST6GALNAC4	—		UNKNOWN
ST6GALNAC5	—		UNKNOWN
ST6GALNAC6	—		UNKNOWN
ST7	—		UNKNOWN
ST8SIA1	—		UNKNOWN
ST8SIA2	—		UNKNOWN
ST8SIA3	—		UNKNOWN
ST8SIA4	—		UNKNOWN
ST8SIA5	—		UNKNOWN
ST8SIA6	—		UNKNOWN
STAC3	Congenital myopathy 13	OMIM:255995	MENDELIAN
STAC3	Native American myopathy	ORPHA:168572	UNKNOWN
STAG1	Intellectual developmental disorder, autosomal dominant 47	OMIM:617635	MENDELIAN
STAG1	STAG1-related intellectual disability-facial dysmorphism-gastroesophageal reflux syndrome	ORPHA:502434	UNKNOWN
STAG2	Holoprosencephaly 13, X-linked	OMIM:301043	MENDELIAN
STAG2	Neurodevelopmental disorder, X-linked, with craniofacial abnormalities	OMIM:301022	MENDELIAN
STAG2	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
STAG2	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
STAG2	Xq25 microduplication syndrome	ORPHA:521258	UNKNOWN
STAG3	Spermatogenic failure 61	OMIM:619672	MENDELIAN
STAG3	Premature ovarian failure 8	OMIM:615723	MENDELIAN
STAG3	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
STAMPB	Microcephaly-Capillary malformation syndrome	OMIM:614261	MENDELIAN
STAR	Lipoid congenital adrenal hyperplasia	OMIM:201710	MENDELIAN
STAR	Familial glucocorticoid deficiency	ORPHA:361	UNKNOWN
STARD6	—		UNKNOWN
STARD9	—		UNKNOWN
STAT1	Immunodeficiency 31C	OMIM:614162	MENDELIAN
STAT1	Mycobacterial and viral infections, susceptibility to, autosomal recessive	OMIM:613796	MENDELIAN
STAT1	Immunodeficiency 31A	OMIM:614892	MENDELIAN
STAT1	STAT1-related autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome	ORPHA:391487	UNKNOWN
STAT2	Pseudo-Torch syndrome 3	OMIM:618886	MENDELIAN
STAT2	Immunodeficiency 44	OMIM:616636	MENDELIAN
STAT3	Autoimmune disease, multisystem, infantile-onset, 1	OMIM:615952	MENDELIAN
STAT3	Hyper-IgE recurrent infection syndrome	OMIM:147060	MENDELIAN
STAT3	Isolated permanent neonatal diabetes mellitus	ORPHA:99885	UNKNOWN
STAT3	Autosomal dominant hyper-IgE syndrome due to STAT3 deficiency	ORPHA:2314	UNKNOWN
STAT3	Acute promyelocytic leukemia	ORPHA:520	UNKNOWN
STAT4	Disabling pansclerotic morphea of childhood	OMIM:620443	MENDELIAN
STAT4	Behçet disease	ORPHA:117	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
STAT4	Oligoarticular juvenile idiopathic arthritis	ORPHA:85410	UNKNOWN
STAT4	Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	ORPHA:85408	UNKNOWN
STAT4	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
STAT4	Pediatric systemic lupus erythematosus	ORPHA:93552	UNKNOWN
STAT5A	—		UNKNOWN
STAT5B	Growth hormone insensitivity with immune dysregulation 2, autosomal dominant	OMIM:618985	MENDELIAN
STAT5B	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive	OMIM:245590	MENDELIAN
STAT5B	Laron syndrome with immunodeficiency	ORPHA:220465	UNKNOWN
STAT5B	Acute promyelocytic leukemia	ORPHA:520	UNKNOWN
STAT6	Hyper-IgE syndrome 6, autosomal dominant, with recurrent infections	OMIM:620532	MENDELIAN
STAT6	Solitary fibrous tumor	ORPHA:2126	UNKNOWN
STBD1	—		UNKNOWN
STEAP3	Anemia, hypochromic microcytic, with iron overload 2	OMIM:615234	MENDELIAN
STEAP3	Severe congenital hypochromic anemia with ringed sideroblasts	ORPHA:300298	UNKNOWN
STEAP4	—		UNKNOWN
STH	—		UNKNOWN
STIL	Microcephaly 7, primary, autosomal recessive	OMIM:612703	MENDELIAN
STIL	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
STIL	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
STIL	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
STIL	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
STIL	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
STIL	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
STIM1	Immunodeficiency 10	OMIM:612783	MENDELIAN
STIM1	Myopathy, tubular aggregate, 1	OMIM:160565	MENDELIAN
STIM1	Stormorken syndrome	OMIM:185070	MENDELIAN
STIM1	Stormorken-Sjaastad-Langslet syndrome	ORPHA:3204	UNKNOWN
STIM1	Tubular aggregate myopathy	ORPHA:2593	UNKNOWN
STIM2	—		UNKNOWN
STK10	—		UNKNOWN
STK11	Testicular tumor, somatic	OMIM:273300	MENDELIAN
STK11	Pancreatic cancer	OMIM:260350	MENDELIAN
STK11	Peutz-Jeghers syndrome	OMIM:175200	MENDELIAN
STK11	Melanoma, cutaneous malignant	OMIM:155600	MENDELIAN
STK11	Peutz-Jeghers syndrome	ORPHA:2869	UNKNOWN
STK11IP	—		UNKNOWN
STK19	—		UNKNOWN
STK3	—		UNKNOWN
STK32A	—		UNKNOWN
STK33	Spermatogenic failure 93	OMIM:620849	MENDELIAN
STK35	—		UNKNOWN
STK36	Ciliary dyskinesia, primary, 46	OMIM:619436	MENDELIAN
STK36	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
STK39	—		UNKNOWN
STK4	Immunodeficiency-110 with lymphoproliferation	OMIM:614868	MENDELIAN
STK40	—		UNKNOWN
STMN1	—		UNKNOWN
STOM	—		UNKNOWN
STOX1	Preeclampsia/eclampsia 4	OMIM:609404	MENDELIAN
STOX1	Preeclampsia	ORPHA:275555	UNKNOWN
STPG1	—		UNKNOWN
STRA13	—		UNKNOWN
STRA6	Microphthalmia, syndromic 9	OMIM:601186	MENDELIAN
STRA6	Matthew-Wood syndrome	ORPHA:2470	UNKNOWN
STRA8	—		UNKNOWN
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy	OMIM:611087	MENDELIAN
STRADA	Polyhydramnios-megalencephaly-symptomatic epilepsy syndrome	ORPHA:500533	UNKNOWN
STRC	Spermatogenic failure 7	OMIM:612997	MENDELIAN
STRC	Deafness, autosomal recessive 16	OMIM:603720	MENDELIAN
STRC	Deafness, sensorineural, and male infertility	OMIM:611102	MENDELIAN
STRC	Deafness-infertility syndrome	ORPHA:94064	UNKNOWN
STS	Ichthyosis, X-linked	OMIM:308100	MENDELIAN
STS	Syndromic recessive X-linked ichthyosis	ORPHA:281090	UNKNOWN
STS	Recessive X-linked ichthyosis	ORPHA:461	UNKNOWN
STT3A	Congenital disorder of glycosylation, type lw	OMIM:615596	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
STT3A	Congenital disorder of glycosylation, type Iw, autosomal dominant	OMIM:619714	MENDELIAN
STT3A	STT3A-CDG	ORPHA:370921	UNKNOWN
STT3B	Congenital disorder of glycosylation, type Ix	OMIM:615597	MENDELIAN
STT3B	STT3B-CDG	ORPHA:370924	UNKNOWN
STUB1	Spinocerebellar ataxia 48	OMIM:618093	MENDELIAN
STUB1	Spinocerebellar ataxia, autosomal recessive 16	OMIM:615768	MENDELIAN
STUB1	Autosomal recessive cerebellar ataxia due to STUB1 deficiency	ORPHA:412057	UNKNOWN
STX11	Hemophagocytic lymphohistiocytosis, familial, 4	OMIM:603552	MENDELIAN
STX11	Familial hemophagocytic lymphohistiocytosis	ORPHA:540	UNKNOWN
STX16	Pseudohypoparathyroidism, type IB	OMIM:603233	MENDELIAN
STX16	Pseudohypoparathyroidism type 1B	ORPHA:94089	UNKNOWN
STX1A	Williams syndrome	ORPHA:904	UNKNOWN
STX1A	Cystic fibrosis	ORPHA:586	UNKNOWN
STX1B	Generalized epilepsy with febrile seizures plus, type 9	OMIM:616172	MENDELIAN
STX1B	Genetic epilepsy with febrile seizure plus	ORPHA:36387	UNKNOWN
STX2	—		UNKNOWN
STXBP1	Developmental and epileptic encephalopathy 4	OMIM:612164	MENDELIAN
STXBP1	STXBP1-related developmental and epileptic encephalopathy	ORPHA:599373	UNKNOWN
STXBP1	9q33.3q34.11 microdeletion syndrome	ORPHA:495818	UNKNOWN
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5	OMIM:613101	MENDELIAN
STXBP2	Familial hemophagocytic lymphohistiocytosis	ORPHA:540	UNKNOWN
STXBP5	—		UNKNOWN
STXBP5L	—		UNKNOWN
STYXL1	—		UNKNOWN
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	OMIM:612073	MENDELIAN
SUCLA2	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	ORPHA:1933	UNKNOWN
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	OMIM:245400	MENDELIAN
SUCLG1	Fatal infantile lactic acidosis with methylmalonic aciduria	ORPHA:17	UNKNOWN
SUCLG2	—		UNKNOWN
SUCO	—		UNKNOWN
SUFU	Joubert syndrome 32	OMIM:617757	MENDELIAN
SUFU	Meningioma, familial, susceptibility to	OMIM:607174	POLYGENIC
SUFU	Basal cell nevus syndrome 2	OMIM:620343	MENDELIAN
SUFU	Medulloblastoma	OMIM:155255	POLYGENIC
SUFU	Gorlin syndrome	ORPHA:377	UNKNOWN
SUFU	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
SUFU	Meningioma	ORPHA:2495	UNKNOWN
SUFU	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
SULF1	—		UNKNOWN
SULT1A1	—		UNKNOWN
SULT1A3	—		UNKNOWN
SULT1C2	—		UNKNOWN
SULT1C3	—		UNKNOWN
SULT1E1	—		UNKNOWN
SULT2A1	—		UNKNOWN
SULT2B1	Ichthyosis, congenital, autosomal recessive 14	OMIM:617571	MENDELIAN
SULT2B1	Lamellar ichthyosis	ORPHA:313	UNKNOWN
SULT2B1	Congenital ichthyosiform erythroderma	ORPHA:79394	UNKNOWN
SULT4A1	—		UNKNOWN
SUMF1	Multiple sulfatase deficiency	OMIM:272200	MENDELIAN
SUMF1	Multiple sulfatase deficiency	ORPHA:585	UNKNOWN
SUMO1	Orofacial cleft 10	OMIM:613705	MENDELIAN
SUMO1	Oligodontia	ORPHA:99798	UNKNOWN
SUN1	—		UNKNOWN
SUN2	—		UNKNOWN
SUN5	Spermatogenic failure 16	OMIM:617187	MENDELIAN
SUN5	Male infertility due to acephalic spermatozoa	ORPHA:529970	UNKNOWN
SUOX	Sulfite oxidase deficiency	OMIM:272300	MENDELIAN
SUPT16H	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum	OMIM:619480	MENDELIAN
SUPT16H	14q11.2 microduplication syndrome	ORPHA:261229	UNKNOWN
SUPT3H	—		UNKNOWN
SUPT5H	—		UNKNOWN
SURF1	Charcot-Marie-Tooth disease, type 4K	OMIM:616684	MENDELIAN
SURF1	Mitochondrial complex IV deficiency, nuclear type 1	OMIM:220110	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
SUV420H1	—		UNKNOWN
SUZ12	Imagawa-Matsumoto syndrome	OMIM:618786	MENDELIAN
SUZ12	Weaver syndrome	ORPHA:3447	UNKNOWN
SV2B	—		UNKNOWN
SVIL	Myofibrillar myopathy 10	OMIM:619040	MENDELIAN
SYCE1	Premature ovarian failure 12	OMIM:616947	MENDELIAN
SYCE1	Spermatogenic failure 15	OMIM:616950	MENDELIAN
SYCE1	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
SYCE2	—		UNKNOWN
SYCP1	—		UNKNOWN
SYCP2	Spermatogenic failure 1	OMIM:258150	MENDELIAN
SYCP3	Spermatogenic failure 4	OMIM:270960	MENDELIAN
SYCP3	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
SYK	Immunodeficiency 82 with systemic inflammation	OMIM:619381	MENDELIAN
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	OMIM:300491	MENDELIAN
SYN1	Intellectual developmental disorder, X-linked 50	OMIM:300115	MENDELIAN
SYN1	X-linked epilepsy-learning disabilities-behavior disorders syndrome	ORPHA:85294	UNKNOWN
SYN2	Schizophrenia	OMIM:181500	POLYGENIC
SYN3	—		UNKNOWN
SYNE1	Emery-Dreifuss muscular dystrophy 4, autosomal dominant	OMIM:612998	MENDELIAN
SYNE1	Spinocerebellar ataxia, autosomal recessive 8	OMIM:610743	MENDELIAN
SYNE1	Arthrogryposis multiplex congenita 3, myogenic type	OMIM:618484	MENDELIAN
SYNE1	Autosomal recessive ataxia, Beauce type	ORPHA:88644	UNKNOWN
SYNE1	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
SYNE1	Autosomal dominant Emery-Dreifuss muscular dystrophy	ORPHA:98853	UNKNOWN
SYNE1	Autosomal recessive myogenic arthrogryposis multiplex congenita	ORPHA:319332	UNKNOWN
SYNE2	Emery-Dreifuss muscular dystrophy 5, autosomal dominant	OMIM:612999	MENDELIAN
SYNE2	Autosomal dominant Emery-Dreifuss muscular dystrophy	ORPHA:98853	UNKNOWN
SYNE4	Deafness, autosomal recessive 76	OMIM:615540	MENDELIAN
SYNGAP1	Intellectual developmental disorder, autosomal dominant 5	OMIM:612621	MENDELIAN
SYNGAP1	SYNGAP1-related developmental and epileptic encephalopathy	ORPHA:544254	UNKNOWN
SYNGAP1	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
SYNGAP1	Epilepsy with myoclonic-atonic seizures	ORPHA:1942	UNKNOWN
SYNGR1	—		UNKNOWN
SYNJ1	Parkinson disease 20, early-onset	OMIM:615530	MENDELIAN
SYNJ1	Epileptic encephalopathy, early infantile, 53	OMIM:617389	MENDELIAN
SYNJ1	Young-onset Parkinson disease	ORPHA:2828	UNKNOWN
SYNJ1	Atypical juvenile parkinsonism	ORPHA:391411	UNKNOWN
SYNJ1	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
SYNM	—		UNKNOWN
SYNPO	—		UNKNOWN
SYNPR	—		UNKNOWN
SYP	Intellectual developmental disorder, X-linked 96	OMIM:300802	MENDELIAN
SYT1	Baker-Gordon syndrome	OMIM:618218	MENDELIAN
SYT1	Infantile hypotonia-oculomotor anomalies-hyperkinetic movements-developmental delay syndrome	ORPHA:522077	UNKNOWN
SYT11	—		UNKNOWN
SYT14	Spinocerebellar ataxia, autosomal recessive 11	OMIM:614229	MENDELIAN
SYT14	Autosomal recessive cerebellar ataxia-psychomotor delay syndrome	ORPHA:284271	UNKNOWN
SYT17	—		UNKNOWN
SYT2	Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive	OMIM:619461	MENDELIAN
SYT2	Myasthenic syndrome, congenital, 7, presynaptic	OMIM:616040	MENDELIAN
SYTL3	—		UNKNOWN
SYTL5	—		UNKNOWN
SZT2	Developmental and epileptic encephalopathy 18	OMIM:615476	MENDELIAN
SZT2	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
T	—		UNKNOWN
TAAR1	—		UNKNOWN
TAAR2	—		UNKNOWN
TAAR6	—		UNKNOWN
TAAR9	—		UNKNOWN
TAB2	Congenital heart defects, multiple types, 2	OMIM:614980	MENDELIAN
TAC3	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
TAC3	Hypogonadotropic hypogonadism 10 with or without anosmia	OMIM:614839	MENDELIAN
TAC3	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TACO1	Mitochondrial complex IV deficiency, nuclear type 8	OMIM:619052	MENDELIAN
TACO1	Mitochondrial complex IV deficiency, nuclear type 1	OMIM:220110	MENDELIAN
TACR1	—		UNKNOWN
TACR3	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
TACR3	Hypogonadotropic hypogonadism 11 with or without anosmia	OMIM:614840	MENDELIAN
TACR3	Kallmann syndrome	ORPHA:478	UNKNOWN
TACR3	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
TACSTD2	Corneal dystrophy, gelatinous drop-like	OMIM:204870	MENDELIAN
TACSTD2	Gelatinous drop-like corneal dystrophy	ORPHA:98957	UNKNOWN
TADA2A	—		UNKNOWN
TAF1	Intellectual developmental disorder, X-linked syndromic 33	OMIM:300966	MENDELIAN
TAF1	Dystonia 3, torsion, X-linked	OMIM:314250	MENDELIAN
TAF1	X-linked intellectual disability-global development delay-facial dysmorphism-sacral caudal remnant syndrome	ORPHA:480907	UNKNOWN
TAF1	X-linked dystonia-parkinsonism	ORPHA:53351	UNKNOWN
TAF15	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
TAF1C	—		UNKNOWN
TAF1L	—		UNKNOWN
TAF2	Neurodevelopmental disorder with feeding difficulties, thin corpus callosum, and foot deformity	OMIM:615599	MENDELIAN
TAF2	Microcephaly-thin corpus callosum-intellectual disability syndrome	ORPHA:397951	UNKNOWN
TAF4	Intellectual developmental disorder, autosomal dominant 73	OMIM:620450	MENDELIAN
TAF4B	Spermatogenic failure 13	OMIM:615841	MENDELIAN
TAF4B	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
TAF6	Alazami-Yuan syndrome	OMIM:617126	MENDELIAN
TAF7L	—		UNKNOWN
TAF9B	—		UNKNOWN
TAL1	Leukemia, acute lymphocytic, susceptibility to, 1	OMIM:613065	MENDELIAN
TAL2	Leukemia, acute lymphocytic, susceptibility to, 1	OMIM:613065	MENDELIAN
TALDO1	Transaldolase deficiency	OMIM:606003	MENDELIAN
TALDO1	Transaldolase deficiency	ORPHA:101028	UNKNOWN
TANC1	—		UNKNOWN
TANC2	Intellectual developmental disorder with autistic features and language delay, with or without seizures	OMIM:618906	MENDELIAN
TAOK1	Developmental delay with or without intellectual impairment or behavioral abnormalities	OMIM:619575	MENDELIAN
TAP1	Bare lymphocyte syndrome, type I	OMIM:604571	MENDELIAN
TAP2	MHC class I deficiency 2	OMIM:620813	MENDELIAN
TAPBP	MHC class I deficiency 3	OMIM:620814	MENDELIAN
TARDBP	Amyotrophic lateral sclerosis 10 with or without frontotemporal dementia	OMIM:612069	MENDELIAN
TARDBP	Frontotemporal dementia with motor neuron disease	ORPHA:275872	UNKNOWN
TARDBP	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
TARDBP	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
TARS2	Combined oxidative phosphorylation deficiency 21	OMIM:615918	MENDELIAN
TAS1R1	—		UNKNOWN
TAS1R2	—		UNKNOWN
TAS1R3	—		UNKNOWN
TAS2R16	Alcohol dependence	OMIM:103780	POLYGENIC
TAS2R19	—		UNKNOWN
TAS2R3	—		UNKNOWN
TAS2R43	—		UNKNOWN
TAS2R46	—		UNKNOWN
TAS2R50	—		UNKNOWN
TAS2R9	—		UNKNOWN
TAT	Tyrosine transaminase deficiency	OMIM:276600	MENDELIAN
TAT	Tyrosinemia type 2	ORPHA:28378	UNKNOWN
TAZ	—		UNKNOWN
TBC1D1	—		UNKNOWN
TBC1D20	Warburg micro syndrome 4	OMIM:615663	MENDELIAN
TBC1D20	Micro syndrome	ORPHA:2510	UNKNOWN
TBC1D23	Pontocerebellar hypoplasia, type 11	OMIM:617695	MENDELIAN
TBC1D24	Epilepsy, rolandic, with paroxysmal exercise-induced dystonia and writer's cramp	OMIM:608105	MENDELIAN
TBC1D24	Doors syndrome	OMIM:220500	MENDELIAN
TBC1D24	Developmental and epileptic encephalopathy 16	OMIM:615338	MENDELIAN
TBC1D24	Myoclonic epilepsy, familial infantile	OMIM:605021	MENDELIAN
TBC1D24	Deafness, autosomal recessive 86	OMIM:614617	MENDELIAN
TBC1D24	Deafness, autosomal dominant 65	OMIM:616044	MENDELIAN
TBC1D24	DOORS syndrome	ORPHA:79500	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TBC1D24	Progressive myoclonic epilepsy with dystonia	ORPHA:352596	UNKNOWN
TBC1D24	Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome	ORPHA:163727	UNKNOWN
TBC1D24	Myoclonic epilepsy of infancy	ORPHA:86909	UNKNOWN
TBC1D24	Epilepsy of infancy with migrating focal seizures	ORPHA:293181	UNKNOWN
TBC1D25	—		UNKNOWN
TBC1D30	—		UNKNOWN
TBC1D7	Macrocephaly/megalencephaly syndrome, autosomal recessive	OMIM:248000	MENDELIAN
TBC1D7	Isolated megalencephaly	ORPHA:2477	UNKNOWN
TBC1D9	—		UNKNOWN
TBCE	Kenny-caffey syndrome, type 1	OMIM:244460	MENDELIAN
TBCE	Hypoparathyroidism-retardation-dysmorphism syndrome	OMIM:241410	MENDELIAN
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy	OMIM:617207	MENDELIAN
TBCE	Sanjad-Sakati syndrome	ORPHA:2323	UNKNOWN
TBCE	Autosomal recessive Kenny-Caffey syndrome	ORPHA:93324	UNKNOWN
TBCE	Early-onset progressive encephalopathy-spastic ataxia-distal spinal muscular atrophy syndrome	ORPHA:496756	UNKNOWN
TBCEL	—		UNKNOWN
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	OMIM:616900	MENDELIAN
TBCK	TBCK-related encephalopathy-severe hypotonia-craniofacial dysmorphism syndrome	ORPHA:488632	UNKNOWN
TBK1	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4	OMIM:616439	MENDELIAN
TBK1	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8	OMIM:617900	POLYGENIC
TBK1	Autoinflammation with arthritis and vasculitis	OMIM:620880	MENDELIAN
TBK1	Frontotemporal dementia with motor neuron disease	ORPHA:275872	UNKNOWN
TBK1	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
TBK1	Herpes simplex virus encephalitis	ORPHA:1930	UNKNOWN
TBL1X	Hypothyroidism, congenital, nongoitrous, 8	OMIM:301033	MENDELIAN
TBL1XR1	Pierpont syndrome	OMIM:602342	MENDELIAN
TBL1XR1	Intellectual developmental disorder, autosomal dominant 41	OMIM:616944	MENDELIAN
TBL1XR1	Pierpont syndrome	ORPHA:487825	UNKNOWN
TBL1XR1	Acute promyelocytic leukemia	ORPHA:520	UNKNOWN
TBL1Y	Deafness, Y-linked 2	OMIM:400047	MENDELIAN
TBP	Spinocerebellar ataxia 17	OMIM:607136	MENDELIAN
TBP	Parkinson disease, late-onset	OMIM:168600	POLYGENIC
TBP	Spinocerebellar ataxia type 17	ORPHA:98759	UNKNOWN
TBR1	Intellectual developmental disorder with autism and speech delay	OMIM:606053	MENDELIAN
TBR1	Developmental delay-language impairment-dopa responsive dystonia-parkinsonism syndrome due to 2q24 microdeletion	ORPHA:1617	UNKNOWN
TBX1	Conotruncal heart malformations	OMIM:217095	MENDELIAN
TBX1	Digeorge syndrome	OMIM:188400	MENDELIAN
TBX1	Tetralogy of Fallot	OMIM:187500	MENDELIAN
TBX1	Velocardiofacial syndrome	OMIM:192430	MENDELIAN
TBX1	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
TBX1	22q11.2 deletion syndrome	ORPHA:567	UNKNOWN
TBX1	22q11.2 duplication syndrome	ORPHA:1727	UNKNOWN
TBX10	—		UNKNOWN
TBX15	Cousin syndrome	OMIM:260660	MENDELIAN
TBX15	Pelviscapular dysplasia	ORPHA:93333	UNKNOWN
TBX18	Congenital anomalies of kidney and urinary tract 2	OMIM:143400	MENDELIAN
TBX19	Acth deficiency, isolated	OMIM:201400	MENDELIAN
TBX19	Congenital isolated ACTH deficiency	ORPHA:199296	UNKNOWN
TBX2	Vertebral anomalies and variable endocrine and T-cell dysfunction	OMIM:618223	MENDELIAN
TBX20	Atrial septal defect 4	OMIM:611363	MENDELIAN
TBX20	Atrial septal defect, ostium secundum type	ORPHA:99103	UNKNOWN
TBX21	Immunodeficiency 88	OMIM:619630	MENDELIAN
TBX21	Asthma, nasal polyps, and aspirin intolerance	OMIM:208550	MENDELIAN
TBX22	Cleft palate, X-linked	OMIM:303400	MENDELIAN
TBX22	Charge-Like syndrome, X-linked	OMIM:302905	MENDELIAN
TBX22	Abruzzo-Erickson syndrome	ORPHA:921	UNKNOWN
TBX3	Ulnar-Mammary syndrome	OMIM:181450	MENDELIAN
TBX3	Ulnar-mammary syndrome	ORPHA:3138	UNKNOWN
TBX4	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension	OMIM:147891	MENDELIAN
TBX4	Amelia, autosomal recessive	OMIM:601360	MENDELIAN
TBX4	Familial clubfoot due to 17q23.1q23.2 microduplication	ORPHA:238578	UNKNOWN
TBX4	Coxopodopatellar syndrome	ORPHA:1509	UNKNOWN
TBX4	Tetraamelia-multiple malformations syndrome	ORPHA:3301	UNKNOWN
TBX4	17q23.1q23.2 microdeletion syndrome	ORPHA:261279	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TBX5	Holt-Oram syndrome	OMIM:142900	MENDELIAN
TBX5	Holt-Oram syndrome	ORPHA:392	UNKNOWN
TBX6	Spondylocostal dysostosis 5	OMIM:122600	MENDELIAN
TBX6	Autosomal recessive spondylocostal dysostosis	ORPHA:2311	UNKNOWN
TBX6	Autosomal dominant spondylocostal dysostosis	ORPHA:1797	UNKNOWN
TBXA2R	Bleeding disorder, platelet-type, 13, susceptibility to	OMIM:614009	POLYGENIC
TBXAS1	Ghosal hematodiaphyseal syndrome	OMIM:231095	MENDELIAN
TBXAS1	Ghosal hematodiaphyseal dysplasia	ORPHA:1802	UNKNOWN
TCAP	Cardiomyopathy, familial hypertrophic, 25	OMIM:607487	MENDELIAN
TCAP	Muscular dystrophy, limb-girdle, type 2G	OMIM:601954	MENDELIAN
TCAP	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
TCEAL1	Neurodevelopmental disorder with gait disturbance, dysmorphic facies and behavioral abnormalities, X-linked	OMIM:301094	MENDELIAN
TCEB1	—		UNKNOWN
TCEB2	—		UNKNOWN
TCEB3	—		UNKNOWN
TCEB3B	—		UNKNOWN
TCEB3C	—		UNKNOWN
TCEB3CL2	—		UNKNOWN
TCF12	Craniosynostosis 3	OMIM:615314	MENDELIAN
TCF12	Hypogonadotropic hypogonadism 26 with or without anosmia	OMIM:619718	MENDELIAN
TCF12	Non-syndromic bicoronal craniosynostosis	ORPHA:35099	UNKNOWN
TCF20	Developmental delay with variable intellectual impairment and behavioral abnormalities	OMIM:618430	MENDELIAN
TCF21	—		UNKNOWN
TCF3	Agammaglobulinemia 8, autosomal dominant	OMIM:616941	MENDELIAN
TCF3	Agammaglobulinemia 8B, autosomal recessive	OMIM:619824	MENDELIAN
TCF3	Autosomal non-syndromic agammaglobulinemia	ORPHA:33110	UNKNOWN
TCF4	Corneal dystrophy, fuchs endothelial, 3	OMIM:613267	MENDELIAN
TCF4	Pitt-Hopkins syndrome	OMIM:610954	MENDELIAN
TCF4	Pitt-Hopkins syndrome	ORPHA:2896	UNKNOWN
TCF4	Primary sclerosing cholangitis	ORPHA:171	UNKNOWN
TCF4	Fuchs endothelial corneal dystrophy	ORPHA:98974	UNKNOWN
TCF7	—		UNKNOWN
TCF7L1	—		UNKNOWN
TCF7L2	Diabetes mellitus, noninsulin-dependent	OMIM:125853	POLYGENIC
TCIRG1	Osteopetrosis, autosomal recessive 1	OMIM:259700	MENDELIAN
TCIRG1	Intermediate osteopetrosis	ORPHA:210110	UNKNOWN
TCIRG1	Autosomal recessive malignant osteopetrosis	ORPHA:667	UNKNOWN
TCIRG1	Autosomal dominant severe congenital neutropenia	ORPHA:486	UNKNOWN
TCIRG1	Dysosteosclerosis	ORPHA:1782	UNKNOWN
TCL1B	—		UNKNOWN
TCL4	—		UNKNOWN
TCN2	Transcobalamin II deficiency	OMIM:275350	MENDELIAN
TCN2	Transcobalamin II deficiency	ORPHA:859	UNKNOWN
TCOF1	Treacher Collins syndrome 1	OMIM:154500	MENDELIAN
TCOF1	Treacher-Collins syndrome	ORPHA:861	UNKNOWN
TCP1	Intellectual developmental disorder with polymicrogyria and seizures	OMIM:621021	MENDELIAN
TCTE1	—		UNKNOWN
TCTE3	—		UNKNOWN
TCTN1	Joubert syndrome 13	OMIM:614173	MENDELIAN
TCTN1	Meckel syndrome	ORPHA:564	UNKNOWN
TCTN1	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
TCTN2	Joubert syndrome 24	OMIM:616654	MENDELIAN
TCTN2	Meckel syndrome, type 8	OMIM:613885	MENDELIAN
TCTN2	Meckel syndrome	ORPHA:564	UNKNOWN
TCTN2	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
TCTN3	Joubert syndrome 18	OMIM:614815	MENDELIAN
TCTN3	Orofaciodigital syndrome IV	OMIM:258860	MENDELIAN
TCTN3	Orofaciodigital syndrome type 4	ORPHA:2753	UNKNOWN
TCTN3	Orofaciodigital syndrome type 6	ORPHA:2754	UNKNOWN
TCTN3	Meckel syndrome	ORPHA:564	UNKNOWN
TCTN3	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
TDG	—		UNKNOWN
TDGF1	—		UNKNOWN
TDO2	Hypertryptophanemia	OMIM:600627	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
TDO2	Hypertryptophanemia	ORPHA:2224	UNKNOWN
TDP1	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1	OMIM:607250	MENDELIAN
TDP1	Spinocerebellar ataxia with axonal neuropathy type 1	ORPHA:94124	UNKNOWN
TDP2	Spinocerebellar ataxia, autosomal recessive 23	OMIM:616949	MENDELIAN
TDP2	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to TUD deficiency	ORPHA:404493	UNKNOWN
TDRD1	—		UNKNOWN
TDRD12	—		UNKNOWN
TDRD5	—		UNKNOWN
TDRD7	Cataract, autosomal recessive congenital 4	OMIM:613887	MENDELIAN
TDRD9	Spermatogenic failure 30	OMIM:618110	MENDELIAN
TDRD9	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
TDRKH	—		UNKNOWN
TEAD1	Sveinsson chorioretinal atrophy	OMIM:108985	MENDELIAN
TEC	—		UNKNOWN
TECPR2	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay	OMIM:615031	MENDELIAN
TECPR2	Hereditary sensory and autonomic neuropathy due to TECPR2 mutation	ORPHA:320385	UNKNOWN
TECR	Intellectual developmental disorder, autosomal recessive 14	OMIM:614020	MENDELIAN
TECTA	Deafness, autosomal recessive 21	OMIM:603629	MENDELIAN
TECTA	Deafness, autosomal dominant nonsyndromic sensorineural 12	OMIM:601543	MENDELIAN
TEK	Glaucoma 3, primary infantile, B	OMIM:600975	MENDELIAN
TEK	Venous malformations, multiple cutaneous and mucosal	OMIM:600195	MENDELIAN
TEK	Glaucoma 3, primary congenital, E	OMIM:617272	MENDELIAN
TEK	Blue rubber bleb nevus syndrome	ORPHA:1059	UNKNOWN
TEK	Congenital glaucoma	ORPHA:98976	UNKNOWN
TEKT2	—		UNKNOWN
TELO2	You-Hoover-Fong syndrome	OMIM:616954	MENDELIAN
TELO2	TELO2-related intellectual disability-neurodevelopmental disorder	ORPHA:488642	UNKNOWN
TENM2	—		UNKNOWN
TENM3	Microphthalmia, isolated, with coloboma 9	OMIM:615145	MENDELIAN
TENM4	Tremor, hereditary essential, 5	OMIM:616736	MENDELIAN
TEP1	—		UNKNOWN
TERB1	Spermatogenic failure 60	OMIM:619646	MENDELIAN
TERB1	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
TERB2	Spermatogenic failure 59	OMIM:619645	MENDELIAN
TERB2	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
TERC	Pulmonary fibrosis, idiopathic	OMIM:178500	MENDELIAN
TERC	Dyskeratosis congenita, autosomal dominant, 1	OMIM:127550	MENDELIAN
TERC	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 2	OMIM:614743	MENDELIAN
TERC	Idiopathic aplastic anemia	ORPHA:88	UNKNOWN
TERC	Idiopathic pulmonary fibrosis	ORPHA:2032	UNKNOWN
TERC	Hoyeraal-Hreidarsson syndrome	ORPHA:3322	UNKNOWN
TERC	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
TERT	Pulmonary fibrosis, idiopathic	OMIM:178500	MENDELIAN
TERT	Melanoma, cutaneous malignant, susceptibility to, 9	OMIM:615134	POLYGENIC
TERT	Aplastic anemia	OMIM:609135	MENDELIAN
TERT	Dyskeratosis congenita, autosomal dominant 2	OMIM:613989	MENDELIAN
TERT	Dyskeratosis congenita, autosomal dominant, 1	OMIM:127550	MENDELIAN
TERT	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1	OMIM:614742	MENDELIAN
TERT	Leukemia, acute myeloid	OMIM:601626	POLYGENIC
TERT	Idiopathic aplastic anemia	ORPHA:88	UNKNOWN
TERT	Adrenocortical carcinoma	ORPHA:1501	UNKNOWN
TERT	Idiopathic pulmonary fibrosis	ORPHA:2032	UNKNOWN
TERT	Hoyeraal-Hreidarsson syndrome	ORPHA:3322	UNKNOWN
TERT	Familial melanoma	ORPHA:618	UNKNOWN
TERT	Meningioma	ORPHA:2495	UNKNOWN
TERT	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
TET1	—		UNKNOWN
TET2	Myelodysplastic syndrome	OMIM:614286	MENDELIAN
TET2	Immunodeficiency 75	OMIM:619126	MENDELIAN
TET2	Essential thrombocythemia	ORPHA:3318	UNKNOWN
TET2	Aggressive systemic mastocytosis	ORPHA:98850	UNKNOWN
TET2	Systemic mastocytosis with associated hematologic neoplasm	ORPHA:98849	UNKNOWN
TET2	Myelodysplastic neoplasm with low blasts	ORPHA:98826	UNKNOWN
TET2	Primary myelofibrosis	ORPHA:824	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TET2	Acquired idiopathic sideroblastic anemia	ORPHA:75564	UNKNOWN
TET3	Beck-Fahrner syndrome	OMIM:618798	MENDELIAN
TEX11	Spermatogenic failure, X-linked 2	OMIM:309120	MENDELIAN
TEX11	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
TEX13B	—		UNKNOWN
TEX14	Spermatogenic failure 23	OMIM:617707	MENDELIAN
TEX14	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
TEX15	Spermatogenic failure 25	OMIM:617960	MENDELIAN
TEX15	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
TEX28	—		UNKNOWN
TF	Atransferrinemia	OMIM:209300	MENDELIAN
TF	Congenital atransferrinemia	ORPHA:1195	UNKNOWN
TFAM	Mitochondrial DNA depletion syndrome 15 (hepatocerebral type)	OMIM:617156	MENDELIAN
TFAP2A	Branchiooculofacial syndrome	OMIM:113620	MENDELIAN
TFAP2A	Branchio-oculo-facial syndrome	ORPHA:1297	UNKNOWN
TFAP2B	Char syndrome	OMIM:169100	MENDELIAN
TFAP2B	Patent ductus arteriosus 2	OMIM:617035	MENDELIAN
TFAP2B	Char syndrome	ORPHA:46627	UNKNOWN
TFCP2	—		UNKNOWN
TFDP1	—		UNKNOWN
TFE3	Intellectual developmental disorder, X-linked, syndromic, with pigmentary mosaicism and coarse facies	OMIM:301066	MENDELIAN
TFE3	Renal cell carcinoma, xp11-associated	OMIM:300854	MENDELIAN
TFF1	—		UNKNOWN
TFG	Neuropathy, hereditary motor and sensory, Okinawa type	OMIM:604484	MENDELIAN
TFG	Spastic paraplegia 57, autosomal recessive	OMIM:615658	MENDELIAN
TFG	Hereditary motor and sensory neuropathy, Okinawa type	ORPHA:90117	UNKNOWN
TFG	Autosomal recessive spastic paraplegia type 57	ORPHA:431329	UNKNOWN
TFPI	—		UNKNOWN
TFR2	Hemochromatosis, type 3	OMIM:604250	MENDELIAN
TFR2	TFR2-related hemochromatosis	ORPHA:225123	UNKNOWN
TFRC	Immunodeficiency 46	OMIM:616740	MENDELIAN
TG	Thyroid dysmorphogenesis 3	OMIM:274700	MENDELIAN
TG	Autoimmune thyroid disease, susceptibility to, 3	OMIM:608175	POLYGENIC
TG	Familial thyroid dysmorphogenesis	ORPHA:95716	UNKNOWN
TGDS	Catel-Manzke syndrome	OMIM:616145	MENDELIAN
TGDS	Pierre-Robin sequence-Manzke dysostosis-clinodactyly of the index finger syndrome	ORPHA:1388	UNKNOWN
TGFB1	Cystic fibrosis	OMIM:219700	POLYGENIC
TGFB1	Inflammatory bowel disease, immunodeficiency, and encephalopathy	OMIM:618213	MENDELIAN
TGFB1	Camurati-Engelmann disease	OMIM:131300	MENDELIAN
TGFB1	Camurati-Engelmann disease	ORPHA:1328	UNKNOWN
TGFB1	Cystic fibrosis	ORPHA:586	UNKNOWN
TGFB2	Loeys-Dietz syndrome 4	OMIM:614816	MENDELIAN
TGFB2	Camurati-engelmann disease, type 2	OMIM:606631	MENDELIAN
TGFB2	Loeys-Dietz syndrome	ORPHA:60030	UNKNOWN
TGFB2	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
TGFB3	Loeys-Dietz syndrome 5	OMIM:615582	MENDELIAN
TGFB3	Arrhythmogenic right ventricular dysplasia 1	OMIM:107970	MENDELIAN
TGFB3	Loeys-Dietz syndrome	ORPHA:60030	UNKNOWN
TGFB3	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
TGFB1	Corneal dystrophy, lattice type I	OMIM:122200	MENDELIAN
TGFB1	Corneal dystrophy, Reis-Bucklers type	OMIM:608470	MENDELIAN
TGFB1	Corneal dystrophy, lattice type IIIA	OMIM:608471	MENDELIAN
TGFB1	Corneal dystrophy of bowman layer, type II	OMIM:602082	MENDELIAN
TGFB1	Corneal dystrophy, Avellino type	OMIM:607541	MENDELIAN
TGFB1	Corneal dystrophy, epithelial basement membrane	OMIM:121820	MENDELIAN
TGFB1	Groenouw type I corneal dystrophy	OMIM:121900	MENDELIAN
TGFB1	Granular corneal dystrophy type I	ORPHA:98962	UNKNOWN
TGFB1	Granular corneal dystrophy type II	ORPHA:98963	UNKNOWN
TGFB1	Lattice corneal dystrophy type I	ORPHA:98964	UNKNOWN
TGFB1	Thiel-Behnke corneal dystrophy	ORPHA:98960	UNKNOWN
TGFBR1	Loeys-Dietz syndrome 1	OMIM:609192	MENDELIAN
TGFBR1	Multiple self-healing squamous epithelioma, susceptibility to	OMIM:132800	POLYGENIC
TGFBR1	Loeys-Dietz syndrome	ORPHA:60030	UNKNOWN
TGFBR1	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TGFBR2	Loeys-Dietz syndrome 2	OMIM:610168	MENDELIAN
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6	OMIM:614331	MENDELIAN
TGFBR2	Esophageal cancer, somatic	OMIM:133239	MENDELIAN
TGFBR2	Squamous cell carcinoma of the esophagus	ORPHA:99977	UNKNOWN
TGFBR2	Loeys-Dietz syndrome	ORPHA:60030	UNKNOWN
TGFBR2	Lynch syndrome	ORPHA:144	UNKNOWN
TGFBR2	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	UNKNOWN
TGFBR3	Familial cerebral saccular aneurysm	ORPHA:231160	UNKNOWN
TGFBRAP1	—		UNKNOWN
TGIF1	Holoprosencephaly 4	OMIM:142946	MENDELIAN
TGIF1	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
TGIF1	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
TGIF1	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
TGIF1	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
TGIF1	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
TGIF1	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
TGIF2LY	—		UNKNOWN
TGM1	Ichthyosis, congenital, autosomal recessive 1	OMIM:242300	MENDELIAN
TGM1	Lamellar ichthyosis	ORPHA:313	UNKNOWN
TGM1	Congenital ichthyosiform erythroderma	ORPHA:79394	UNKNOWN
TGM1	Self-improving collodion baby	ORPHA:281122	UNKNOWN
TGM1	Acral self-healing collodion baby	ORPHA:281127	UNKNOWN
TGM1	Bathing suit ichthyosis	ORPHA:100976	UNKNOWN
TGM2	—		UNKNOWN
TGM4	—		UNKNOWN
TGM5	Peeling skin syndrome 2	OMIM:609796	MENDELIAN
TGM5	Acral peeling skin syndrome	ORPHA:263534	UNKNOWN
TGM6	Spinocerebellar ataxia 35	OMIM:613908	MENDELIAN
TGM6	Spinocerebellar ataxia type 35	ORPHA:276193	UNKNOWN
TH	Segawa syndrome, autosomal recessive	OMIM:605407	MENDELIAN
TH	Autosomal recessive dopa-responsive dystonia	ORPHA:101150	UNKNOWN
THADA	—		UNKNOWN
THAP1	Dystonia 6, torsion	OMIM:602629	MENDELIAN
THAP1	Primary dystonia, DYT6 type	ORPHA:98806	UNKNOWN
THBD	Hemolytic uremic syndrome, atypical, susceptibility to, 6	OMIM:612926	POLYGENIC
THBD	Thrombophilia due to thrombomodulin defect	OMIM:614486	MENDELIAN
THBS1	—		UNKNOWN
THBS2	Ehlers-Danlos syndrome, classic-like, 3	OMIM:620865	MENDELIAN
THBS4	—		UNKNOWN
THOC2	Intellectual developmental disorder, X-linked syndromic, Kumar type	OMIM:300957	MENDELIAN
THOC2	Arthrogryposis multiplex congenita 7, X-linked	OMIM:301127	MENDELIAN
THOC2	X-linked intellectual disability-short stature-overweight syndrome	ORPHA:457240	UNKNOWN
THOC6	Beaulieu-Boycott-Innes syndrome	OMIM:613680	MENDELIAN
THOC6	THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome	ORPHA:363444	UNKNOWN
THPO	Thrombocytopenia 9	OMIM:620478	MENDELIAN
THPO	Amegakaryocytic thrombocytopenia, congenital, 2	OMIM:620481	MENDELIAN
THPO	Thrombocythemia 1	OMIM:187950	MENDELIAN
THPO	Familial thrombocytosis	ORPHA:71493	UNKNOWN
THRA	Hypothyroidism, congenital, nongoitrous, 6	OMIM:614450	MENDELIAN
THRB	Thyroid hormone resistance, generalized, autosomal recessive	OMIM:274300	MENDELIAN
THRB	Thyroid hormone resistance, generalized, autosomal dominant	OMIM:188570	MENDELIAN
THRB	Thyroid hormone resistance, selective pituitary	OMIM:145650	MENDELIAN
THRB	Resistance to thyroid hormone due to a mutation in thyroid hormone receptor beta	ORPHA:566243	UNKNOWN
THSD7A	—		UNKNOWN
THSD7B	—		UNKNOWN
TIA1	Welander distal myopathy	OMIM:604454	MENDELIAN
TIA1	Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia	OMIM:619133	MENDELIAN
TIA1	Distal myopathy, Welander type	ORPHA:603	UNKNOWN
TICAM1	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6	OMIM:614850	POLYGENIC
TICAM1	Herpes simplex virus encephalitis	ORPHA:1930	UNKNOWN
TIMELESS	Advance sleep phase syndrome, familial, 4	OMIM:620015	MENDELIAN
TIMM21	—		UNKNOWN
TIMM44	—		UNKNOWN
TIMM8A	Mohr-Tranebjaerg syndrome	OMIM:304700	MENDELIAN
TIMM8A	Mohr-Tranebjaerg syndrome	ORPHA:52368	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TIMP1	—		UNKNOWN
TIMP2	—		UNKNOWN
TIMP3	Sorsby fundus dystrophy	OMIM:136900	MENDELIAN
TIMP3	Sorsby fundus dystrophy	ORPHA:59181	UNKNOWN
TINAG	—		UNKNOWN
TINF2	Dyskeratosis congenita, autosomal dominant 3	OMIM:613990	MENDELIAN
TINF2	Dyskeratosis congenita, autosomal dominant, 1	OMIM:127550	MENDELIAN
TINF2	Revesz syndrome	OMIM:268130	MENDELIAN
TINF2	Hoyeraal-Hreidarsson syndrome	ORPHA:3322	UNKNOWN
TINF2	Revesz syndrome	ORPHA:3088	UNKNOWN
TINF2	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
TIPARP	—		UNKNOWN
TIPIN	—		UNKNOWN
TJP1	—		UNKNOWN
TJP2	Cholestasis, progressive familial intrahepatic 4	OMIM:615878	MENDELIAN
TJP2	Hypercholanemia, familial	OMIM:607748	MENDELIAN
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type)	OMIM:609560	MENDELIAN
TK2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3	OMIM:617069	MENDELIAN
TK2	Autosomal recessive progressive external ophthalmoplegia	ORPHA:254886	UNKNOWN
TLE3	—		UNKNOWN
TLE6	Preimplantation embryonic lethality 1	OMIM:616814	MENDELIAN
TLE6	Female infertility due to oocyte meiotic arrest	ORPHA:488191	UNKNOWN
TLK1	—		UNKNOWN
TLK2	Intellectual developmental disorder, autosomal dominant 57	OMIM:618050	MENDELIAN
TLL1	Atrial septal defect 6	OMIM:613087	MENDELIAN
TLL1	Atrial septal defect, ostium primum type	ORPHA:99106	UNKNOWN
TLL1	Atrial septal defect, ostium secundum type	ORPHA:99103	UNKNOWN
TLR10	—		UNKNOWN
TLR2	Colorectal cancer	OMIM:114500	POLYGENIC
TLR2	Leprosy, susceptibility to	OMIM:246300	POLYGENIC
TLR3	Immunodeficiency 83, susceptibility to viral infections	OMIM:613002	POLYGENIC
TLR3	Herpes simplex virus encephalitis	ORPHA:1930	UNKNOWN
TLR4	Behçet disease	ORPHA:117	UNKNOWN
TLR6	—		UNKNOWN
TLR7	Immunodeficiency 74, COVID19-related, X-linked	OMIM:301051	MENDELIAN
TLR7	Systemic lupus erythematosus 17	OMIM:301080	MENDELIAN
TLR7	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
TLR8	Immunodeficiency 98 with autoinflammation, X-linked	OMIM:301078	MENDELIAN
TLR9	—		UNKNOWN
TLX2	—		UNKNOWN
TM4SF19	—		UNKNOWN
TM4SF20	Specific language impairment 5	OMIM:615432	POLYGENIC
TM9SF4	—		UNKNOWN
TMC1	Deafness, neurosensory, autosomal recessive 7	OMIM:600974	MENDELIAN
TMC1	Deafness, autosomal dominant 36	OMIM:606705	MENDELIAN
TMC3	—		UNKNOWN
TMC6	Epidermodysplasia verruciformis, susceptibility to, 1	OMIM:226400	POLYGENIC
TMC6	Inherited epidermodysplasia verruciformis	ORPHA:302	UNKNOWN
TMC8	Epidermodysplasia verruciformis 2	OMIM:618231	POLYGENIC
TMC8	Inherited epidermodysplasia verruciformis	ORPHA:302	UNKNOWN
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1	OMIM:213980	MENDELIAN
TMCO1	Cerebrofaciothoracic dysplasia	ORPHA:1394	UNKNOWN
TMEM114	—		UNKNOWN
TMEM121	—		UNKNOWN
TMEM126A	Optic atrophy 7 with or without auditory neuropathy	OMIM:612989	MENDELIAN
TMEM127	Pheochromocytoma	OMIM:171300	POLYGENIC
TMEM127	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
TMEM135	—		UNKNOWN
TMEM138	Joubert syndrome 16	OMIM:614465	MENDELIAN
TMEM138	Joubert syndrome with oculorenal defect	ORPHA:2318	UNKNOWN
TMEM165	Congenital disorder of glycosylation, type IIK	OMIM:614727	MENDELIAN
TMEM173	—		UNKNOWN
TMEM185A	FRAXF syndrome	ORPHA:100974	UNKNOWN
TMEM187	—		UNKNOWN
TMEM189	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TMEM189-UBE2V1	—		UNKNOWN
TMEM2	—		UNKNOWN
TMEM216	Joubert syndrome 2	OMIM:608091	MENDELIAN
TMEM216	Meckel syndrome, type 2	OMIM:603194	MENDELIAN
TMEM216	Retinitis pigmentosa 98	OMIM:620996	MENDELIAN
TMEM216	Orofaciodigital syndrome type 6	ORPHA:2754	UNKNOWN
TMEM216	Retinitis pigmentosa	ORPHA:791	UNKNOWN
TMEM216	Joubert syndrome with oculorenal defect	ORPHA:2318	UNKNOWN
TMEM216	Meckel syndrome	ORPHA:564	UNKNOWN
TMEM231	Meckel syndrome 11	OMIM:615397	MENDELIAN
TMEM231	Joubert syndrome 20	OMIM:614970	MENDELIAN
TMEM231	Orofaciodigital syndrome type 6	ORPHA:2754	UNKNOWN
TMEM231	Joubert syndrome with oculorenal defect	ORPHA:2318	UNKNOWN
TMEM231	Meckel syndrome	ORPHA:564	UNKNOWN
TMEM237	Joubert syndrome 14	OMIM:614424	MENDELIAN
TMEM237	Joubert syndrome with renal defect	ORPHA:220497	UNKNOWN
TMEM237	Joubert syndrome with oculorenal defect	ORPHA:2318	UNKNOWN
TMEM237	Meckel syndrome	ORPHA:564	UNKNOWN
TMEM237	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
TMEM240	Spinocerebellar ataxia 21	OMIM:607454	MENDELIAN
TMEM240	Spinocerebellar ataxia type 21	ORPHA:98773	UNKNOWN
TMEM38B	Osteogenesis imperfecta, type XIV	OMIM:615066	MENDELIAN
TMEM39A	—		UNKNOWN
TMEM43	Arrhythmogenic right ventricular dysplasia, familial, 5	OMIM:604400	MENDELIAN
TMEM43	Emery-Dreifuss muscular dystrophy 7, autosomal dominant	OMIM:614302	MENDELIAN
TMEM43	Auditory neuropathy, autosomal dominant 3	OMIM:619832	MENDELIAN
TMEM43	Autosomal dominant Emery-Dreifuss muscular dystrophy	ORPHA:98853	UNKNOWN
TMEM47	—		UNKNOWN
TMEM5	—		UNKNOWN
TMEM67	Rhyns syndrome	OMIM:602152	MENDELIAN
TMEM67	Joubert syndrome 6	OMIM:610688	MENDELIAN
TMEM67	Coach syndrome 1	OMIM:216360	MENDELIAN
TMEM67	Bardet-Biedl syndrome 14	OMIM:615991	POLYGENIC
TMEM67	Meckel syndrome 3	OMIM:607361	MENDELIAN
TMEM67	Nephronophthisis 11	OMIM:613550	MENDELIAN
TMEM67	Joubert syndrome with hepatic defect	ORPHA:1454	UNKNOWN
TMEM67	Senior-Boichis syndrome	ORPHA:84081	UNKNOWN
TMEM67	RHYNS syndrome	ORPHA:140976	UNKNOWN
TMEM67	Meckel syndrome	ORPHA:564	UNKNOWN
TMEM67	Isolated Joubert syndrome	ORPHA:475	UNKNOWN
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	OMIM:614052	MENDELIAN
TMEM70	TMEM70-related mitochondrial encephalo-cardio-myopathy	ORPHA:1194	UNKNOWN
TMEM8A	—		UNKNOWN
TMEM9	—		UNKNOWN
TMEM92	—		UNKNOWN
TMEM98	Nanophthalmos 4	OMIM:615972	MENDELIAN
TMEM98	Isolated complete colobomatous microphthalmia	ORPHA:35612	UNKNOWN
TMEM99	—		UNKNOWN
TMIE	Deafness, autosomal recessive 6	OMIM:600971	MENDELIAN
TMLHE	Autism, susceptibility to, X-linked 6	OMIM:300872	POLYGENIC
TMPO	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
TMPRSS11A	—		UNKNOWN
TMPRSS15	Enterokinase deficiency	OMIM:226200	MENDELIAN
TMPRSS2	—		UNKNOWN
TMPRSS3	Deafness, autosomal recessive 8/10	OMIM:601072	MENDELIAN
TMPRSS5	—		UNKNOWN
TMPRSS6	Iron-Refractory iron deficiency anemia	OMIM:206200	MENDELIAN
TMPRSS6	IRIDA syndrome	ORPHA:209981	UNKNOWN
TMTC3	Lissencephaly 8	OMIM:617255	MENDELIAN
TMTC3	Periventricular nodular heterotopia	ORPHA:98892	UNKNOWN
TMTC3	Cobblestone lissencephaly without muscular or ocular involvement	ORPHA:352682	UNKNOWN
TNC	Deafness, autosomal dominant 56	OMIM:615629	MENDELIAN
TNF	Asthma, susceptibility to	OMIM:600807	POLYGENIC
TNF	Migraine with or without aura, susceptibility to, 1	OMIM:157300	POLYGENIC
TNF	Immunodeficiency 127	OMIM:620977	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
TNFAIP1	—		UNKNOWN
TNFAIP2	—		UNKNOWN
TNFAIP3	Autoinflammatory syndrome, familial, Behcet-like	OMIM:616744	MENDELIAN
TNFAIP3	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
TNFRSF10A	—		UNKNOWN
TNFRSF10B	Squamous cell carcinoma, head and neck	OMIM:275355	MENDELIAN
TNFRSF11A	Familial expansile osteolysis	OMIM:174810	MENDELIAN
TNFRSF11A	Paget disease of bone 2, early-onset	OMIM:602080	POLYGENIC
TNFRSF11A	Osteopetrosis, autosomal recessive 7	OMIM:612301	MENDELIAN
TNFRSF11A	Juvenile Paget disease	ORPHA:2801	UNKNOWN
TNFRSF11A	Dysosteosclerosis	ORPHA:1782	UNKNOWN
TNFRSF11B	Paget disease of bone 5, juvenile-onset	OMIM:239000	MENDELIAN
TNFRSF11B	Familial calcium pyrophosphate deposition	ORPHA:1416	UNKNOWN
TNFRSF11B	Juvenile Paget disease	ORPHA:2801	UNKNOWN
TNFRSF13B	Immunoglobulin A deficiency 2	OMIM:609529	MENDELIAN
TNFRSF13B	Immunodeficiency, common variable, 2	OMIM:240500	MENDELIAN
TNFRSF13C	Immunodeficiency, common variable, 4	OMIM:613494	MENDELIAN
TNFRSF13C	Immunodeficiency, common variable, 2	OMIM:240500	MENDELIAN
TNFRSF14	—		UNKNOWN
TNFRSF1A	Periodic fever, familial, autosomal dominant	OMIM:142680	MENDELIAN
TNFRSF1A	Intermittent hydrarthrosis	ORPHA:329967	UNKNOWN
TNFRSF1A	Tumor necrosis factor receptor 1 associated periodic syndrome	ORPHA:32960	UNKNOWN
TNFRSF1B	SÃ©zary syndrome	ORPHA:3162	UNKNOWN
TNFRSF1B	Classic mycosis fungoides	ORPHA:2584	UNKNOWN
TNFRSF25	—		UNKNOWN
TNFRSF4	Immunodeficiency 16	OMIM:615593	MENDELIAN
TNFRSF9	Immunodeficiency 109 with lymphoproliferation	OMIM:620282	MENDELIAN
TNFSF10	—		UNKNOWN
TNFSF11	Osteopetrosis, autosomal recessive 2	OMIM:259710	MENDELIAN
TNFSF11	Autosomal recessive malignant osteopetrosis	ORPHA:667	UNKNOWN
TNFSF13B	—		UNKNOWN
TNFSF14	—		UNKNOWN
TNFSF15	Primary biliary cholangitis	ORPHA:186	UNKNOWN
TNFSF18	—		UNKNOWN
TNFSF4	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
TNFSF4	Narcolepsy type 1	ORPHA:2073	UNKNOWN
TNFSF8	—		UNKNOWN
TNKS	—		UNKNOWN
TNKS2	—		UNKNOWN
TNNC1	Cardiomyopathy, dilated, 1Z	OMIM:611879	MENDELIAN
TNNC1	Cardiomyopathy, familial hypertrophic, 13	OMIM:613243	MENDELIAN
TNNC1	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
TNNI2	Arthrogryposis, distal, type 2B	OMIM:601680	MENDELIAN
TNNI2	Sheldon-Hall syndrome	ORPHA:1147	UNKNOWN
TNNI2	Distal arthrogryposis type 1	ORPHA:1146	UNKNOWN
TNNI3	Cardiomyopathy, familial restrictive, 1	OMIM:115210	MENDELIAN
TNNI3	Cardiomyopathy, dilated, 2A	OMIM:611880	MENDELIAN
TNNI3	Cardiomyopathy, dilated, 1FF	OMIM:613286	MENDELIAN
TNNI3	Cardiomyopathy, familial hypertrophic, 7	OMIM:613690	MENDELIAN
TNNI3	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
TNNI3	Familial isolated restrictive cardiomyopathy	ORPHA:75249	UNKNOWN
TNNI3K	Cardiac conduction disease with or without dilated cardiomyopathy	OMIM:616117	MENDELIAN
TNNT1	Nemaline myopathy 5C, autosomal dominant	OMIM:620389	MENDELIAN
TNNT1	Nemaline myopathy 5B, autosomal recessive, childhood-onset	OMIM:620386	MENDELIAN
TNNT1	Nemaline myopathy 5, Amish type	OMIM:605355	MENDELIAN
TNNT1	Amish nemaline myopathy	ORPHA:98902	UNKNOWN
TNNT2	Cardiomyopathy, familial hypertrophic, 2	OMIM:115195	MENDELIAN
TNNT2	Cardiomyopathy, dilated, 1D	OMIM:601494	MENDELIAN
TNNT2	Cardiomyopathy, familial restrictive, 3	OMIM:612422	MENDELIAN
TNNT2	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
TNNT2	Familial isolated restrictive cardiomyopathy	ORPHA:75249	UNKNOWN
TNNT3	Arthrogryposis, distal, type 2B2	OMIM:618435	MENDELIAN
TNNT3	Sheldon-Hall syndrome	ORPHA:1147	UNKNOWN
TNNT3	Distal arthrogryposis type 1	ORPHA:1146	UNKNOWN
TNP1	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TNPO2	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies	OMIM:619556	MENDELIAN
TNPO3	Muscular dystrophy, limb-girdle, autosomal dominant 2	OMIM:608423	MENDELIAN
TNPO3	Primary biliary cholangitis	ORPHA:186	UNKNOWN
TNR	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus	OMIM:619653	MENDELIAN
TNRC6B	Global developmental delay with speech and behavioral abnormalities	OMIM:619243	MENDELIAN
TNS2	—		UNKNOWN
TNS3	—		UNKNOWN
TNXB	Vesicoureteral reflux 8	OMIM:615963	MENDELIAN
TNXB	Ehlers-Danlos syndrome, classic-like	OMIM:606408	MENDELIAN
TNXB	Classical-like Ehlers-Danlos syndrome type 1	ORPHA:230839	UNKNOWN
TOMM40	Early-onset autosomal dominant Alzheimer disease	ORPHA:1020	UNKNOWN
TOP1	—		UNKNOWN
TOP1MT	—		UNKNOWN
TOP2A	—		UNKNOWN
TOP3A	Microcephaly, growth restriction, and increased sister chromatid exchange 2	OMIM:618097	MENDELIAN
TOP3A	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5	OMIM:618098	MENDELIAN
TOP3A	Autosomal recessive progressive external ophthalmoplegia	ORPHA:254886	UNKNOWN
TOP3B	—		UNKNOWN
TOPBP1	—		UNKNOWN
TOPORS	Retinitis pigmentosa 31	OMIM:609923	MENDELIAN
TOPORS	Orofaciodigital syndrome type 6	ORPHA:2754	UNKNOWN
TOPORS	Retinitis pigmentosa	ORPHA:791	UNKNOWN
TOR1A	Dystonia 1, torsion, autosomal dominant	OMIM:128100	MENDELIAN
TOR1A	Arthrogryposis multiplex congenita 5	OMIM:618947	MENDELIAN
TOR1A	Early-onset generalized limb-onset dystonia	ORPHA:256	UNKNOWN
TOR1A	Myoclonus-dystonia syndrome	ORPHA:36899	UNKNOWN
TOR1AIP1	Muscular dystrophy, limb-girdle, type 2Y	OMIM:617072	MENDELIAN
TOX3	—		UNKNOWN
TP53	Glioma susceptibility 1	OMIM:137800	POLYGENIC
TP53	Nasopharyngeal carcinoma	OMIM:607107	MENDELIAN
TP53	Pancreatic cancer	OMIM:260350	MENDELIAN
TP53	Papilloma of choroid plexus	OMIM:260500	POLYGENIC
TP53	Basal cell carcinoma 7	OMIM:614740	POLYGENIC
TP53	Osteosarcoma	OMIM:259500	POLYGENIC
TP53	Breast cancer	OMIM:114480	MENDELIAN
TP53	Bone marrow failure syndrome 5	OMIM:618165	MENDELIAN
TP53	Li-Fraumeni syndrome	OMIM:151623	MENDELIAN
TP53	Colorectal cancer	OMIM:114500	POLYGENIC
TP53	Hepatocellular carcinoma	OMIM:114550	MENDELIAN
TP53	Adrenocortical carcinoma, pediatric	OMIM:202300	POLYGENIC
TP53	Adrenocortical carcinoma	ORPHA:1501	UNKNOWN
TP53	Papilloma of choroid plexus	ORPHA:2807	UNKNOWN
TP53	Essential thrombocythemia	ORPHA:3318	UNKNOWN
TP53	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145	UNKNOWN
TP53	Cushing disease	ORPHA:96253	UNKNOWN
TP53	Familial pancreatic carcinoma	ORPHA:1333	UNKNOWN
TP53	Osteosarcoma	ORPHA:668	UNKNOWN
TP53	Li-Fraumeni syndrome	ORPHA:524	UNKNOWN
TP53AIP1	—		UNKNOWN
TP53BP1	—		UNKNOWN
TP53I3	—		UNKNOWN
TP53RK	Galloway-Mowat syndrome 4	OMIM:617730	MENDELIAN
TP53RK	Galloway-Mowat syndrome	ORPHA:2065	UNKNOWN
TP63	Rapp-Hodgkin syndrome	OMIM:129400	MENDELIAN
TP63	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3	OMIM:604292	MENDELIAN
TP63	Limb-Mammary syndrome	OMIM:603543	MENDELIAN
TP63	Adult syndrome	OMIM:103285	MENDELIAN
TP63	Orofacial cleft 8	OMIM:618149	MENDELIAN
TP63	Premature ovarian failure 21	OMIM:620311	MENDELIAN
TP63	Hay-Wells syndrome	OMIM:106260	MENDELIAN
TP63	Split-Hand/foot malformation 4	OMIM:605289	MENDELIAN
TP63	Isolated cleft lip	ORPHA:199302	UNKNOWN
TP63	Cleft lip/palate	ORPHA:199306	UNKNOWN
TP63	Classic bladder exstrophy	ORPHA:93930	UNKNOWN
TP63	ADULT syndrome	ORPHA:978	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TP63	Ankyloblepharon filiforme adnatum-cleft palate syndrome	ORPHA:1072	UNKNOWN
TP63	EEC syndrome	ORPHA:1896	UNKNOWN
TP63	Isolated split hand-split foot malformation	ORPHA:2440	UNKNOWN
TP63	Cleft lip and alveolus	ORPHA:141291	UNKNOWN
TP63	Limb-mammary syndrome	ORPHA:69085	UNKNOWN
TP73	Ciliary dyskinesia, primary, 47, and lissencephaly	OMIM:619466	MENDELIAN
TPH1	—		UNKNOWN
TPH2	Major depressive disorder	OMIM:608516	POLYGENIC
TPH2	Attention deficit-hyperactivity disorder, susceptibility to, 7	OMIM:613003	POLYGENIC
TPI1	Triosephosphate isomerase deficiency	OMIM:615512	MENDELIAN
TPI1	Triose phosphate-isomerase deficiency	ORPHA:868	UNKNOWN
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	OMIM:614458	MENDELIAN
TPM1	Cardiomyopathy, familial hypertrophic, 3	OMIM:115196	MENDELIAN
TPM1	Cardiomyopathy, dilated, 1Y	OMIM:611878	MENDELIAN
TPM1	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
TPM2	Congenital myopathy 23	OMIM:609285	MENDELIAN
TPM2	Arthrogryposis, distal, type 1A	OMIM:108120	MENDELIAN
TPM2	Congenital fiber-type disproportion myopathy	ORPHA:2020	UNKNOWN
TPM2	Typical nemaline myopathy	ORPHA:171436	UNKNOWN
TPM2	Childhood-onset nemaline myopathy	ORPHA:171439	UNKNOWN
TPM2	Sheldon-Hall syndrome	ORPHA:1147	UNKNOWN
TPM2	Distal arthrogryposis type 1	ORPHA:1146	UNKNOWN
TPM2	Cap myopathy	ORPHA:171881	UNKNOWN
TPM3	Nemaline myopathy 1	OMIM:609284	MENDELIAN
TPM3	Myopathy, congenital, with fiber-type disproportion	OMIM:255310	MENDELIAN
TPM3	Congenital fiber-type disproportion myopathy	ORPHA:2020	UNKNOWN
TPM3	Intermediate nemaline myopathy	ORPHA:171433	UNKNOWN
TPM3	Childhood-onset nemaline myopathy	ORPHA:171439	UNKNOWN
TPM3	Cap myopathy	ORPHA:171881	UNKNOWN
TPMT	Thiopurines, poor metabolism of, 1	OMIM:610460	POLYGENIC
TPO	Thyroid hormonogenesis, genetic defect in, 2A	OMIM:274500	MENDELIAN
TPO	Familial thyroid dys hormonogenesis	ORPHA:95716	UNKNOWN
TPP1	Spinocerebellar ataxia, autosomal recessive 7	OMIM:609270	MENDELIAN
TPP1	Ceroid lipofuscinosis, neuronal, 2	OMIM:204500	MENDELIAN
TPP1	Childhood-onset autosomal recessive slowly progressive spinocerebellar ataxia	ORPHA:284324	UNKNOWN
TPP2	Immunodeficiency 78 with autoimmunity and developmental delay	OMIM:619220	MENDELIAN
TPP2	Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome due to TPP2 deficiency	ORPHA:444463	UNKNOWN
TPPP	—		UNKNOWN
TPRN	Deafness, autosomal recessive 79	OMIM:613307	MENDELIAN
TPSB2	—		UNKNOWN
TPTE	—		UNKNOWN
TRA2B	Ramond-Elliott neurodevelopmental syndrome	OMIM:621421	MENDELIAN
TRAC	Immunodeficiency 7, TCR-alpha/beta deficient	OMIM:615387	MENDELIAN
TRADD	—		UNKNOWN
TRAF2	—		UNKNOWN
TRAF3	Immunodeficiency 132B	OMIM:621096	MENDELIAN
TRAF3	Immunodeficiency 132A	OMIM:614849	MENDELIAN
TRAF3	Herpes simplex virus encephalitis	ORPHA:1930	UNKNOWN
TRAF3IP1	—		UNKNOWN
TRAF3IP2	Candidiasis, familial, 8	OMIM:615527	MENDELIAN
TRAF3IP2	Chronic mucocutaneous candidiasis	ORPHA:1334	UNKNOWN
TRAF6	Autosomal dominant hypohidrotic ectodermal dysplasia	ORPHA:1810	UNKNOWN
TRAF7	Cardiac, facial, and digital anomalies with developmental delay	OMIM:618164	MENDELIAN
TRAF7	Meningioma	ORPHA:2495	UNKNOWN
TRAK1	Epileptic encephalopathy, early infantile, 68	OMIM:618201	MENDELIAN
TRAK1	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
TRAK2	—		UNKNOWN
TRAP1	—		UNKNOWN
TRAPPC10	Neurodevelopmental disorder with microcephaly, short stature, and speech delay	OMIM:620027	MENDELIAN
TRAPPC10	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
TRAPPC11	Muscular dystrophy, limb-girdle, autosomal recessive 18	OMIM:615356	MENDELIAN
TRAPPC11	Triple A syndrome	ORPHA:869	UNKNOWN
TRAPPC11	TRAPPC11-related limb-girdle muscular dystrophy R18	ORPHA:369840	UNKNOWN
TRAPPC11	Intellectual disability-hyperkinetic movement-truncal ataxia syndrome	ORPHA:369847	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TRAPPC2	Spondyloepiphyseal dysplasia tarda, X-linked	OMIM:313400	MENDELIAN
TRAPPC2	Spondyloepiphyseal dysplasia tarda	ORPHA:93284	UNKNOWN
TRAPPC6B	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy	OMIM:617862	MENDELIAN
TRAPPC9	Intellectual developmental disorder, autosomal recessive 13	OMIM:613192	MENDELIAN
TRAPPC9	Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome	ORPHA:352530	UNKNOWN
TRBV9	—		UNKNOWN
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 1, with or without atrial dysfunction and/or dilated cardiomyopathy	OMIM:604772	MENDELIAN
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness	OMIM:615441	MENDELIAN
TREH	Trehalase deficiency	OMIM:612119	MENDELIAN
TREH	Trehalase deficiency	ORPHA:103909	UNKNOWN
TREM2	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	OMIM:221770	MENDELIAN
TREM2	Alzheimer disease 17, susceptibility to	OMIM:615080	POLYGENIC
TREM2	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2	OMIM:618193	MENDELIAN
TREM2	Nasu-Hakola disease	ORPHA:2770	UNKNOWN
TREM2	Behavioral variant of frontotemporal dementia	ORPHA:275864	UNKNOWN
TREM2	Early-onset autosomal dominant Alzheimer disease	ORPHA:1020	UNKNOWN
TREM2	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
TREM2	Progressive non-fluent aphasia	ORPHA:100070	UNKNOWN
TREM2	Semantic dementia	ORPHA:100069	UNKNOWN
TRERF1	—		UNKNOWN
TREX1	Chilblain lupus 1	OMIM:610448	MENDELIAN
TREX1	Aicardi-Goutieres syndrome 1	OMIM:225750	MENDELIAN
TREX1	Systemic lupus erythematosus	OMIM:152700	POLYGENIC
TREX1	Vasculopathy, retinal, with cerebral leukodystrophy	OMIM:192315	MENDELIAN
TREX1	Aicardi-Goutieres syndrome	ORPHA:51	UNKNOWN
TREX1	Systemic lupus erythematosus	ORPHA:536	UNKNOWN
TREX1	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	ORPHA:247691	UNKNOWN
TREX2	—		UNKNOWN
TRHR	Hypothyroidism, congenital, nongoitrous, 7	OMIM:618573	MENDELIAN
TRHR	Resistance to thyrotropin-releasing hormone syndrome	ORPHA:99832	UNKNOWN
TRIB1	—		UNKNOWN
TRIB2	—		UNKNOWN
TRIB3	—		UNKNOWN
TRIL	—		UNKNOWN
TRIM17	—		UNKNOWN
TRIM2	Charcot-Marie-Tooth disease, axonal, type 2R	OMIM:615490	MENDELIAN
TRIM21	—		UNKNOWN
TRIM23	—		UNKNOWN
TRIM28	Wilms tumor 7	OMIM:621332	MENDELIAN
TRIM28	Nephroblastoma	ORPHA:654	UNKNOWN
TRIM32	Muscular dystrophy, limb-girdle, type 2H	OMIM:254110	MENDELIAN
TRIM32	Bardet-Biedl syndrome 11	OMIM:615988	MENDELIAN
TRIM32	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
TRIM32	TRIM32-related limb-girdle muscular dystrophy R8	ORPHA:1878	UNKNOWN
TRIM33	Developmental dysplasia of the hip 4	OMIM:621311	MENDELIAN
TRIM33	Dislocation of the hip-dysmorphism syndrome	ORPHA:2412	UNKNOWN
TRIM37	Mulibrey nanism	OMIM:253250	MENDELIAN
TRIM37	Mulibrey nanism	ORPHA:2576	UNKNOWN
TRIM5	—		UNKNOWN
TRIM63	Cardiomyopathy, familial hypertrophic, 31	OMIM:621270	MENDELIAN
TRIM71	Hydrocephalus, congenital communicating, 1	OMIM:618667	MENDELIAN
TRIM8	Focal segmental glomerulosclerosis and neurodevelopmental syndrome	OMIM:619428	MENDELIAN
TRIM8	Early infantile developmental and epileptic encephalopathy	ORPHA:1934	UNKNOWN
TRIO	Intellectual developmental disorder, autosomal dominant 63, with macrocephaly	OMIM:618825	MENDELIAN
TRIO	Intellectual developmental disorder, autosomal dominant 44, with microcephaly	OMIM:617061	MENDELIAN
TRIO	Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome	ORPHA:476126	UNKNOWN
TRIOBP	Deafness, autosomal recessive 28	OMIM:609823	MENDELIAN
TRIP11	Osteochondrodysplasia	OMIM:184260	MENDELIAN
TRIP11	Achondrogenesis, type 1A	OMIM:200600	MENDELIAN
TRIP11	Odontochondrodysplasia	ORPHA:166272	UNKNOWN
TRIP11	Achondrogenesis type 1A	ORPHA:93299	UNKNOWN
TRIP12	Clark-Baraitser syndrome	OMIM:617752	MENDELIAN
TRIP13	Mosaic variegated aneuploidy syndrome 3	OMIM:617598	MENDELIAN
TRIP13	Oocyte maturation defect 9	OMIM:619011	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
TRIP13	Mosaic variegated aneuploidy syndrome	ORPHA:1052	UNKNOWN
TRIP13	Nephroblastoma	ORPHA:654	UNKNOWN
TRIT1	Combined oxidative phosphorylation deficiency 35	OMIM:617873	MENDELIAN
TRMT1	Intellectual developmental disorder, autosomal recessive 68	OMIM:618302	MENDELIAN
TRMT10A	Microcephaly, short stature, and impaired glucose metabolism 1	OMIM:616033	MENDELIAN
TRMT10A	Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome	ORPHA:391408	UNKNOWN
TRMT44	—		UNKNOWN
TRMU	Deafness, aminoglycoside-induced	OMIM:580000	MENDELIAN
TRMU	Deafness, aminoglycoside-induced	OMIM:580000	POLYGENIC
TRMU	Liver failure, infantile, transient	OMIM:613070	MENDELIAN
TRMU	Mitochondrial myopathy with reversible cytochrome C oxidase deficiency	ORPHA:254864	UNKNOWN
TRNT1	Retinitis pigmentosa and erythrocytic microcytosis	OMIM:616959	MENDELIAN
TRNT1	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay	OMIM:616084	MENDELIAN
TROAP	—		UNKNOWN
TROVE2	—		UNKNOWN
TRPA1	Episodic pain syndrome, familial, 1	OMIM:615040	MENDELIAN
TRPC3	Spinocerebellar ataxia 41	OMIM:616410	MENDELIAN
TRPC3	Spinocerebellar ataxia type 41	ORPHA:458798	UNKNOWN
TRPC4	—		UNKNOWN
TRPC5	—		UNKNOWN
TRPC6	Focal segmental glomerulosclerosis 2	OMIM:603965	MENDELIAN
TRPC6	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
TRPM1	Night blindness, congenital stationary (complete), 1C, autosomal recessive	OMIM:613216	MENDELIAN
TRPM2	—		UNKNOWN
TRPM3	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures	OMIM:620224	MENDELIAN
TRPM3	Cataract 50 with or without glaucoma	OMIM:620253	MENDELIAN
TRPM4	Progressive familial heart block, type IB	OMIM:604559	MENDELIAN
TRPM4	Erythrokeratoderma variabilis et progressiva 6	OMIM:618531	MENDELIAN
TRPM4	Progressive symmetric erythrokeratodermia	ORPHA:316	UNKNOWN
TRPM4	Brugada syndrome	ORPHA:130	UNKNOWN
TRPM4	Hereditary progressive cardiac conduction defect	ORPHA:871	UNKNOWN
TRPM6	Hypomagnesemia 1, intestinal	OMIM:602014	MENDELIAN
TRPM7	Amyotrophic lateral sclerosis-parkinsonism/dementia complex 1	OMIM:105500	POLYGENIC
TRPM7	Autosomal dominant macrothrombocytopenia	ORPHA:140957	UNKNOWN
TRPS1	Trichorhinophalangeal syndrome, type I	OMIM:190350	MENDELIAN
TRPS1	Trichorhinophalangeal syndrome, type III	OMIM:190351	MENDELIAN
TRPS1	Trichorhinophalangeal syndrome type 1	ORPHA:77258	UNKNOWN
TRPS1	Trichorhinophalangeal syndrome type 2	ORPHA:502	UNKNOWN
TRPV1	—		UNKNOWN
TRPV3	Palmoplantar keratoderma, nonepidermolytic, focal 2	OMIM:616400	MENDELIAN
TRPV3	Olmsted syndrome 1	OMIM:614594	MENDELIAN
TRPV3	Mutilating palmoplantar keratoderma with periorificial keratotic plaques	ORPHA:659	UNKNOWN
TRPV4	Spondylometaphyseal dysplasia, Kozlowski type	OMIM:184252	MENDELIAN
TRPV4	Digital arthropathy-brachydactyly, familial	OMIM:606835	MENDELIAN
TRPV4	Neuronopathy, distal hereditary motor, type VIII	OMIM:600175	MENDELIAN
TRPV4	Scapuloperoneal spinal muscular atrophy	OMIM:181405	MENDELIAN
TRPV4	Parastremmatic dwarfism	OMIM:168400	MENDELIAN
TRPV4	Hereditary motor and sensory neuropathy, type IIC	OMIM:606071	MENDELIAN
TRPV4	Brachyolmia type 3	OMIM:113500	MENDELIAN
TRPV4	Metatropic dysplasia	OMIM:156530	MENDELIAN
TRPV4	Avascular necrosis of femoral head, primary, 2	OMIM:617383	MENDELIAN
TRPV4	Spondyloepiphyseal dysplasia, Maroteaux type	OMIM:184095	MENDELIAN
TRPV4	Autosomal dominant congenital benign spinal muscular atrophy	ORPHA:1216	UNKNOWN
TRPV4	Familial avascular necrosis of femoral head	ORPHA:86820	UNKNOWN
TRPV4	Metatropic dysplasia	ORPHA:2635	UNKNOWN
TRPV4	Spondylometaphyseal dysplasia, Kozlowski type	ORPHA:93314	UNKNOWN
TRPV4	Autosomal dominant brachyolmia	ORPHA:93304	UNKNOWN
TRPV4	Familial digital arthropathy-brachydactyly	ORPHA:85169	UNKNOWN
TRPV5	—		UNKNOWN
TRRAP	Deafness, autosomal dominant 75	OMIM:618778	MENDELIAN
TRRAP	Developmental delay with or without dysmorphic facies and autism	OMIM:618454	MENDELIAN
TSC1	Focal cortical dysplasia of taylor	OMIM:607341	MENDELIAN
TSC1	Lymphangioleiomyomatosis	OMIM:606690	MENDELIAN
TSC1	Tuberous sclerosis-1	OMIM:191100	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
TSC1	Tuberous sclerosis complex	ORPHA:805	UNKNOWN
TSC1	Lymphangioleiomyomatosis	ORPHA:538	UNKNOWN
TSC2	Tuberous sclerosis-2	OMIM:613254	MENDELIAN
TSC2	Focal cortical dysplasia of taylor	OMIM:607341	MENDELIAN
TSC2	Lymphangioleiomyomatosis	OMIM:606690	MENDELIAN
TSC2	Tuberous sclerosis complex	ORPHA:805	UNKNOWN
TSC2	Lymphangioleiomyomatosis	ORPHA:538	UNKNOWN
TSEN15	Pontocerebellar hypoplasia, type 2F	OMIM:617026	MENDELIAN
TSEN15	Pontocerebellar hypoplasia type 2	ORPHA:2524	UNKNOWN
TSEN2	Pontocerebellar hypoplasia, type 2B	OMIM:612389	MENDELIAN
TSEN2	Pontocerebellar hypoplasia type 2	ORPHA:2524	UNKNOWN
TSEN34	Pontocerebellar hypoplasia, type 2C	OMIM:612390	MENDELIAN
TSEN34	Pontocerebellar hypoplasia type 2	ORPHA:2524	UNKNOWN
TSEN54	Pontocerebellar hypoplasia, type 5	OMIM:610204	MENDELIAN
TSEN54	Pontocerebellar hypoplasia, type 4	OMIM:225753	MENDELIAN
TSEN54	Pontocerebellar hypoplasia, type 2A	OMIM:277470	MENDELIAN
TSEN54	Pontocerebellar hypoplasia type 2	ORPHA:2524	UNKNOWN
TSEN54	Pontocerebellar hypoplasia type 4	ORPHA:166063	UNKNOWN
TSFM	Combined oxidative phosphorylation deficiency 3	OMIM:610505	MENDELIAN
TSG101	—		UNKNOWN
TSGA10	Spermatogenic failure 26	OMIM:617961	MENDELIAN
TSHB	Hypothyroidism, congenital, nongoitrous, 4	OMIM:275100	MENDELIAN
TSHB	Isolated thyroid-stimulating hormone deficiency	ORPHA:90674	UNKNOWN
TSHR	Hyperthyroidism, nonautoimmune	OMIM:609152	MENDELIAN
TSHR	Hyperthyroidism, familial gestational	OMIM:603373	MENDELIAN
TSHR	Hypothyroidism, congenital, nongoitrous, 1	OMIM:275200	MENDELIAN
TSHR	Hypothyroidism due to TSH receptor mutations	ORPHA:90673	UNKNOWN
TSHR	Familial gestational hyperthyroidism	ORPHA:99819	UNKNOWN
TSHR	Thyroid hypoplasia	ORPHA:95720	UNKNOWN
TSHR	Athyreosis	ORPHA:95713	UNKNOWN
TSHR	Familial hyperthyroidism due to mutations in TSH receptor	ORPHA:424	UNKNOWN
TSHZ1	Aural atresia, congenital	OMIM:607842	MENDELIAN
TSHZ3	19q13.11 microdeletion syndrome	ORPHA:217346	UNKNOWN
TSLP	—		UNKNOWN
TSPAN12	Exudative vitreoretinopathy 5	OMIM:613310	MENDELIAN
TSPAN12	Familial exudative vitreoretinopathy	ORPHA:891	UNKNOWN
TSPAN17	—		UNKNOWN
TSPAN7	Intellectual developmental disorder, X-linked 58	OMIM:300210	MENDELIAN
TSPEAR	Tooth agenesis, selective, 10	OMIM:620173	MENDELIAN
TSPEAR	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis	OMIM:618180	MENDELIAN
TSPEAR	Deafness, autosomal recessive 98	OMIM:614861	MENDELIAN
TSPO	—		UNKNOWN
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome	OMIM:608800	MENDELIAN
TSPYL1	Sudden infant death-dysgenesis of the testes syndrome	ORPHA:168593	UNKNOWN
TSR2	Diamond-Blackfan anemia 14 with mandibulofacial dysostosis	OMIM:300946	MENDELIAN
TSR2	Diamond-Blackfan anemia	ORPHA:124	UNKNOWN
TSSC4	—		UNKNOWN
TSSK2	—		UNKNOWN
TSSK3	—		UNKNOWN
TSSK4	—		UNKNOWN
TST	—		UNKNOWN
TTBK2	Spinocerebellar ataxia 11	OMIM:604432	MENDELIAN
TTBK2	Spinocerebellar ataxia type 11	ORPHA:98767	UNKNOWN
TTC19	Mitochondrial complex III deficiency, nuclear type 2	OMIM:615157	MENDELIAN
TTC21B	Short-Rib thoracic dysplasia 4 with or without polydactyly	OMIM:613819	MENDELIAN
TTC21B	Nephronophthisis 12	OMIM:613820	MENDELIAN
TTC21B	Jeune syndrome	ORPHA:474	UNKNOWN
TTC37	—		UNKNOWN
TTC5	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism	OMIM:619244	MENDELIAN
TTC7A	Gastrointestinal defects and immunodeficiency syndrome	OMIM:243150	MENDELIAN
TTC7A	Combined immunodeficiency-multiple intestinal atresia	ORPHA:436252	UNKNOWN
TTC8	Retinitis pigmentosa 51	OMIM:613464	MENDELIAN
TTC8	Bardet-Biedl syndrome 8	OMIM:615985	MENDELIAN
TTC8	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
TTC8	Retinitis pigmentosa	ORPHA:791	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TTF2	—		UNKNOWN
TTI2	Intellectual developmental disorder, autosomal recessive 39	OMIM:615541	MENDELIAN
TTI2	Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome	ORPHA:391307	UNKNOWN
TTLL1	—		UNKNOWN
TTLL11	—		UNKNOWN
TTLL5	Cone-Rod dystrophy 19	OMIM:615860	MENDELIAN
TTLL5	Cone rod dystrophy	ORPHA:1872	UNKNOWN
TTLL9	—		UNKNOWN
TTN	Congenital myopathy 5 with cardiomyopathy	OMIM:611705	MENDELIAN
TTN	Myopathy, myofibrillar, 9, with early respiratory failure	OMIM:603689	MENDELIAN
TTN	Muscular dystrophy, limb-girdle, autosomal recessive 10	OMIM:608807	MENDELIAN
TTN	Cardiomyopathy, dilated, 1G	OMIM:604145	MENDELIAN
TTN	Cardiomyopathy, familial hypertrophic, 9	OMIM:613765	MENDELIAN
TTN	Tibial muscular dystrophy, tardive	OMIM:600334	MENDELIAN
TTN	Hereditary myopathy with early respiratory failure	ORPHA:178464	UNKNOWN
TTN	Hereditary atrial fibrillation	ORPHA:334	UNKNOWN
TTN	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
TTN	Autosomal recessive centronuclear myopathy	ORPHA:169186	UNKNOWN
TTN	Classic multiminicore myopathy	ORPHA:324604	UNKNOWN
TTN	Tibial muscular dystrophy	ORPHA:609	UNKNOWN
TPPA	Ataxia with isolated vitamin E deficiency	OMIM:277460	MENDELIAN
TPPA	Ataxia with vitamin E deficiency	ORPHA:96	UNKNOWN
TTR	Carpal tunnel syndrome	OMIM:115430	MENDELIAN
TTR	Dystransthyretinemic euthyroidal hyperthyroxinemia	OMIM:145680	MENDELIAN
TTR	Amyloidosis, hereditary, transthyretin-related	OMIM:105210	MENDELIAN
TTR	ATTRV30M amyloidosis	ORPHA:85447	UNKNOWN
TTR	ATTRV122I amyloidosis	ORPHA:85451	UNKNOWN
TTY13	—		UNKNOWN
TUB	Retinal dystrophy and obesity	OMIM:616188	MENDELIAN
TUB	Retinitis pigmentosa	ORPHA:791	UNKNOWN
TUBA1A	Lissencephaly 3	OMIM:611603	MENDELIAN
TUBA1A	Lissencephaly due to TUBA1A mutation	ORPHA:171680	UNKNOWN
TUBA1A	Fetal akinesia deformation sequence	ORPHA:994	UNKNOWN
TUBA1A	Congenital fibrosis of extraocular muscles	ORPHA:45358	UNKNOWN
TUBA1A	Tubulinopathy-associated dysgyria	ORPHA:467166	UNKNOWN
TUBA3C	—		UNKNOWN
TUBA4A	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia	OMIM:616208	MENDELIAN
TUBA4A	Oocyte/zygote/embryo maturation arrest 23	OMIM:621231	MENDELIAN
TUBA4A	Spastic ataxia 11, autosomal dominant	OMIM:621226	MENDELIAN
TUBA4A	Congenital myopathy 26	OMIM:621225	MENDELIAN
TUBA8	Macrothrombocytopenia, isolated, 2, autosomal dominant	OMIM:619840	MENDELIAN
TUBA8	Polymicrogyria with optic nerve hypoplasia	ORPHA:250972	UNKNOWN
TUBA8	Autosomal dominant macrothrombocytopenia	ORPHA:140957	UNKNOWN
TUBB	Cortical dysplasia, complex, with other brain malformations 6	OMIM:615771	MENDELIAN
TUBB	Skin creases, congenital symmetric circumferential, 1	OMIM:156610	MENDELIAN
TUBB	Multiple benign circumferential skin creases on limbs	ORPHA:2505	UNKNOWN
TUBB1	Macrothrombocytopenia, autosomal dominant, tubb1-related	OMIM:613112	MENDELIAN
TUBB1	Autosomal dominant macrothrombocytopenia	ORPHA:140957	UNKNOWN
TUBB2A	Cortical dysplasia, complex, with other brain malformations 5	OMIM:615763	MENDELIAN
TUBB2B	Cortical dysplasia, complex, with other brain malformations 7	OMIM:610031	MENDELIAN
TUBB2B	Congenital fibrosis of extraocular muscles	ORPHA:45358	UNKNOWN
TUBB2B	Polymicrogyria due to TUBB2B mutation	ORPHA:300573	UNKNOWN
TUBB2B	Tubulinopathy-associated dysgyria	ORPHA:467166	UNKNOWN
TUBB2B	Dysequilibrium syndrome	ORPHA:1766	UNKNOWN
TUBB3	Cortical dysplasia, complex, with other brain malformations 1	OMIM:614039	MENDELIAN
TUBB3	Fibrosis of extraocular muscles, congenital, 3A, with or without extraocularinvolvement	OMIM:600638	MENDELIAN
TUBB3	Congenital fibrosis of extraocular muscles	ORPHA:45358	UNKNOWN
TUBB3	Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation	ORPHA:300570	UNKNOWN
TUBB3	Tubulinopathy-associated dysgyria	ORPHA:467166	UNKNOWN
TUBB4A	Dystonia 4, torsion, autosomal dominant	OMIM:128101	MENDELIAN
TUBB4A	Leukodystrophy, hypomyelinating, 6	OMIM:612438	MENDELIAN
TUBB4A	Primary dystonia, DYT4 type	ORPHA:98805	UNKNOWN
TUBB8	Oocyte maturation defect 2	OMIM:616780	MENDELIAN
TUBB8	Female infertility due to oocyte meiotic arrest	ORPHA:488191	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
TUBG1	Cortical dysplasia, complex, with other brain malformations 4	OMIM:615412	MENDELIAN
TUBG1	15q11.2 microdeletion syndrome	ORPHA:261183	UNKNOWN
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, 3	OMIM:616335	MENDELIAN
TUBGCP4	Autosomal recessive chorioretinopathy-microcephaly syndrome	ORPHA:2518	UNKNOWN
TUBGCP5	—		UNKNOWN
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1	OMIM:251270	MENDELIAN
TUBGCP6	Autosomal recessive chorioretinopathy-microcephaly syndrome	ORPHA:2518	UNKNOWN
TUFM	Combined oxidative phosphorylation deficiency 4	OMIM:610678	MENDELIAN
TULP1	Retinitis pigmentosa 14	OMIM:600132	MENDELIAN
TULP1	Leber congenital amaurosis 15	OMIM:613843	MENDELIAN
TULP1	Leber congenital amaurosis	ORPHA:65	UNKNOWN
TULP1	Retinitis pigmentosa	ORPHA:791	UNKNOWN
TULP3	Hepatorenocardiac degenerative fibrosis	OMIM:619902	MENDELIAN
TULP4	—		UNKNOWN
TUSC1	—		UNKNOWN
TUSC3	Intellectual developmental disorder, autosomal recessive 7	OMIM:611093	MENDELIAN
TWIST1	Robinow-Sorauf syndrome	OMIM:180750	MENDELIAN
TWIST1	Sweeney-Cox syndrome	OMIM:617746	MENDELIAN
TWIST1	Saethre-Chotzen syndrome	OMIM:101400	MENDELIAN
TWIST1	Craniosynostosis 1	OMIM:123100	MENDELIAN
TWIST1	Non-syndromic sagittal craniosynostosis	ORPHA:35093	UNKNOWN
TWIST1	Non-syndromic bicoronal craniosynostosis	ORPHA:35099	UNKNOWN
TWIST1	Saethre-Chotzen syndrome	ORPHA:794	UNKNOWN
TWIST2	Ablepharon-Macrostomia syndrome	OMIM:200110	MENDELIAN
TWIST2	Focal facial dermal dysplasia 3, Setleis type	OMIM:227260	MENDELIAN
TWIST2	Barber-Say syndrome	OMIM:209885	MENDELIAN
TWIST2	Barber-Say syndrome	ORPHA:1231	UNKNOWN
TWIST2	Ablepharon macrostomia syndrome	ORPHA:920	UNKNOWN
TWIST2	Focal facial dermal dysplasia type III	ORPHA:1807	UNKNOWN
TWNK	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3	OMIM:609286	MENDELIAN
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	OMIM:271245	MENDELIAN
TWNK	Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis	OMIM:607459	MENDELIAN
TWNK	Perrault syndrome 5	OMIM:616138	MENDELIAN
TWNK	Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome	ORPHA:70595	UNKNOWN
TWNK	Infantile-onset spinocerebellar ataxia	ORPHA:1186	UNKNOWN
TWNK	Autosomal dominant progressive external ophthalmoplegia	ORPHA:254892	UNKNOWN
TXLNB	—		UNKNOWN
TXN2	Combined oxidative phosphorylation deficiency 29	OMIM:616811	MENDELIAN
TXN2	Combined oxidative phosphorylation defect type 29	ORPHA:478029	UNKNOWN
TXNIP	—		UNKNOWN
TXNL4A	Burn-Mckeown syndrome	OMIM:608572	MENDELIAN
TXNL4A	Burn-McKeown syndrome	ORPHA:1200	UNKNOWN
TXNRD2	Glucocorticoid deficiency 5	OMIM:617825	MENDELIAN
TXNRD2	Familial glucocorticoid deficiency	ORPHA:361	UNKNOWN
TXNRD2	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
TYK2	Immunodeficiency 35	OMIM:611521	MENDELIAN
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	OMIM:603041	MENDELIAN
TYMP	Mitochondrial neurogastrointestinal encephalomyopathy	ORPHA:298	UNKNOWN
TYMS	Dyskeratosis congenita, digenic	OMIM:620040	MENDELIAN
TYMS	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
TYR	Albinism, oculocutaneous, type IB	OMIM:606952	MENDELIAN
TYR	Albinism, oculocutaneous, type IA	OMIM:203100	MENDELIAN
TYR	Oculocutaneous albinism type 1A	ORPHA:79431	UNKNOWN
TYR	Oculocutaneous albinism type 1B	ORPHA:79434	UNKNOWN
TYR	Waardenburg syndrome type 2	ORPHA:895	UNKNOWN
TYRO3	—		UNKNOWN
TYROBP	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	OMIM:221770	MENDELIAN
TYROBP	Nasu-Hakola disease	ORPHA:2770	UNKNOWN
TYRP1	Albinism, oculocutaneous, type III	OMIM:203290	MENDELIAN
TYRP1	Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)	OMIM:612271	MENDELIAN
TYRP1	Oculocutaneous albinism type 3	ORPHA:79433	UNKNOWN
U2AF1	—		UNKNOWN
UACA	—		UNKNOWN
UBA1	VEXAS syndrome, somatic	OMIM:301054	MENDELIAN
UBA1	Spinal muscular atrophy, X-linked 2	OMIM:301830	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
UBA1	Infantile-onset X-linked spinal muscular atrophy	ORPHA:1145	UNKNOWN
UBA3	—		UNKNOWN
UBA7	—		UNKNOWN
UBAC2	Behçet disease	ORPHA:117	UNKNOWN
UBAP2L	Neurodevelopmental disorder with impaired language, behavioral abnormalities, and dysmorphic facies	OMIM:620494	MENDELIAN
UBB	Cleft hard palate	ORPHA:101023	UNKNOWN
UBB	Submucosal cleft palate	ORPHA:155878	UNKNOWN
UBB	Bifid uvula	ORPHA:99771	UNKNOWN
UBB	Cleft velum	ORPHA:99772	UNKNOWN
UBD	—		UNKNOWN
UBE2A	Intellectual developmental disorder, X-linked syndromic, Nascimento type	OMIM:300860	MENDELIAN
UBE2A	X-linked intellectual disability, Nascimento type	ORPHA:163956	UNKNOWN
UBE2B	—		UNKNOWN
UBE2I	—		UNKNOWN
UBE2N	—		UNKNOWN
UBE2NL	—		UNKNOWN
UBE2T	Fanconi anemia, complementation group T	OMIM:616435	MENDELIAN
UBE2T	Fanconi anemia	ORPHA:84	UNKNOWN
UBE2V1	—		UNKNOWN
UBE2V2	—		UNKNOWN
UBE3A	Angelman syndrome	OMIM:105830	MENDELIAN
UBE3A	15q11q13 microduplication syndrome	ORPHA:238446	UNKNOWN
UBE3A	Angelman syndrome due to maternal 15q11q13 deletion	ORPHA:98794	UNKNOWN
UBE3A	Angelman syndrome due to paternal uniparental disomy of chromosome 15	ORPHA:98795	UNKNOWN
UBE3A	Angelman syndrome due to a point mutation	ORPHA:411511	UNKNOWN
UBE3A	Angelman syndrome due to imprinting defect in 15q11-q13	ORPHA:411515	UNKNOWN
UBE3B	Kaufman oculocerebrofacial syndrome	OMIM:244450	MENDELIAN
UBE3B	Oculocerebrofacial syndrome, Kaufman type	ORPHA:2707	UNKNOWN
UBE3C	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities	OMIM:620270	MENDELIAN
UBIAD1	Corneal dystrophy, crystalline, of schnyder	OMIM:121800	MENDELIAN
UBN2	—		UNKNOWN
UBQLN1	—		UNKNOWN
UBQLN2	Amyotrophic lateral sclerosis 15 with or without frontotemporal dementia	OMIM:300857	MENDELIAN
UBQLN2	Juvenile amyotrophic lateral sclerosis	ORPHA:300605	UNKNOWN
UBQLN2	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
UBR1	Johanson-Blizzard syndrome	OMIM:243800	MENDELIAN
UBR1	Johanson-Blizzard syndrome	ORPHA:2315	UNKNOWN
UBR2	—		UNKNOWN
UBR3	—		UNKNOWN
UBR5	Neurodevelopmental disorder with speech delay and behavioral abnormalities	OMIM:621372	MENDELIAN
UBR7	Li-Campeau syndrome	OMIM:619189	MENDELIAN
UBTF	Neurodegeneration, childhood-onset, with brain atrophy	OMIM:617672	MENDELIAN
UBTF	Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder	ORPHA:500180	UNKNOWN
UCHL1	Spastic paraplegia 79A, autosomal dominant	OMIM:620221	MENDELIAN
UCHL1	Spastic paraplegia 79, autosomal recessive	OMIM:615491	MENDELIAN
UCHL1	Parkinson disease 5, susceptibility to	OMIM:613643	POLYGENIC
UCHL1	Young-onset Parkinson disease	ORPHA:2828	UNKNOWN
UCN	—		UNKNOWN
UCP1	—		UNKNOWN
UCP2	Hyperinsulinism due to UCP2 deficiency	ORPHA:276556	UNKNOWN
UCP3	Obesity	OMIM:601665	POLYGENIC
UFD1L	—		UNKNOWN
UGCG	—		UNKNOWN
UGGT1	Congenital disorder of glycosylation, type IICC	OMIM:621381	MENDELIAN
UGGT2	—		UNKNOWN
UGT1A1	Crigler-Najjar syndrome, type I	OMIM:218800	MENDELIAN
UGT1A1	Hyperbilirubinemia, familial transient neonatal	OMIM:237900	MENDELIAN
UGT1A1	Crigler-Najjar syndrome, type II	OMIM:606785	MENDELIAN
UGT1A1	Gilbert syndrome	OMIM:143500	MENDELIAN
UGT1A1	Crigler-Najjar syndrome type 1	ORPHA:79234	UNKNOWN
UGT1A1	Crigler-Najjar syndrome type 2	ORPHA:79235	UNKNOWN
UGT1A10	—		UNKNOWN
UGT1A3	—		UNKNOWN
UGT1A4	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
UGT1A5	—		UNKNOWN
UGT1A6	—		UNKNOWN
UGT1A7	—		UNKNOWN
UGT1A8	—		UNKNOWN
UGT1A9	—		UNKNOWN
UGT2A1	—		UNKNOWN
UGT2A3	—		UNKNOWN
UGT2B10	—		UNKNOWN
UGT2B15	—		UNKNOWN
UGT2B28	—		UNKNOWN
UGT2B4	—		UNKNOWN
UGT2B7	—		UNKNOWN
UGT8	—		UNKNOWN
UHRF1BP1	—		UNKNOWN
UIMC1	—		UNKNOWN
ULK2	—		UNKNOWN
ULK4	—		UNKNOWN
UMOD	Tubulointerstitial kidney disease, autosomal dominant, 1	OMIM:162000	MENDELIAN
UMPS	Orotic aciduria	OMIM:258900	MENDELIAN
UMPS	Hereditary orotic aciduria	ORPHA:30	UNKNOWN
UNC119	Immunodeficiency 13	OMIM:615518	MENDELIAN
UNC119	Cone-rod dystrophy 24	OMIM:620342	MENDELIAN
UNC119	Cone rod dystrophy	ORPHA:1872	UNKNOWN
UNC13A	Intellectual development disorder with seizures and dysmorphic facies	OMIM:621457	MENDELIAN
UNC13A	Neurodevelopmental disorder with speech delay, movement abnormalities, and seizures	OMIM:621456	MENDELIAN
UNC13A	Neurodevelopmental disorder with hypotonia, epilepsy, and absent speech	OMIM:621455	MENDELIAN
UNC13A	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
UNC13B	—		UNKNOWN
UNC13C	—		UNKNOWN
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3	OMIM:608898	MENDELIAN
UNC13D	Familial hemophagocytic lymphohistiocytosis	ORPHA:540	UNKNOWN
UNC45B	Cataract 43	OMIM:616279	MENDELIAN
UNC45B	Myofibrillar myopathy 11	OMIM:619178	MENDELIAN
UNC5C	—		UNKNOWN
UNC5CL	—		UNKNOWN
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	OMIM:616801	MENDELIAN
UNC93A	—		UNKNOWN
UNC93B1	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1	OMIM:610551	POLYGENIC
UNC93B1	Herpes simplex virus encephalitis	ORPHA:1930	UNKNOWN
UNG	Immunodeficiency with hyper IgM, type 5	OMIM:608106	MENDELIAN
UNKL	—		UNKNOWN
UPB1	Beta-ureidopropionase deficiency	OMIM:613161	MENDELIAN
UPB1	Beta-ureidopropionase deficiency	ORPHA:65287	UNKNOWN
UPF2	—		UNKNOWN
UPF3B	Intellectual developmental disorder, X-linked syndromic 14	OMIM:300676	MENDELIAN
UPF3B	Lujan-Fryns syndrome	ORPHA:776	UNKNOWN
UQCRB	Mitochondrial complex III deficiency, nuclear type 3	OMIM:615158	MENDELIAN
UQCRC1	Parkinsonism with polyneuropathy	OMIM:619279	MENDELIAN
UQCRC2	Mitochondrial complex III deficiency, nuclear type 5	OMIM:615160	MENDELIAN
UQCRFS1	Mitochondrial complex III deficiency, nuclear type 10	OMIM:618775	MENDELIAN
UQCRH	Mitochondrial complex III deficiency, nuclear type 11	OMIM:620137	MENDELIAN
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4	OMIM:615159	MENDELIAN
UROC1	Urocanase deficiency	OMIM:276880	MENDELIAN
UROC1	Urocanic aciduria	ORPHA:210128	UNKNOWN
UROD	Porphyria cutanea tarda	OMIM:176100	MENDELIAN
UROD	Hepatoerythropoietic porphyria	ORPHA:95159	UNKNOWN
UROS	Porphyria, congenital erythropoietic	OMIM:263700	MENDELIAN
UROS	Congenital erythropoietic porphyria	ORPHA:79277	UNKNOWN
USB1	Poikiloderma with neutropenia	OMIM:604173	MENDELIAN
USB1	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
USH1C	Usher syndrome, type I	OMIM:276900	MENDELIAN
USH1C	Usher syndrome, type IC	OMIM:276904	MENDELIAN
USH1C	Deafness, neurosensory, autosomal recessive 18	OMIM:602092	MENDELIAN
USH1C	Usher syndrome type 1	ORPHA:231169	UNKNOWN
USH1G	Usher syndrome, type IG	OMIM:606943	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
USH1G	Usher syndrome type 1	ORPHA:231169	UNKNOWN
USH2A	Usher syndrome, type IIA	OMIM:276901	MENDELIAN
USH2A	Retinitis pigmentosa 39	OMIM:613809	MENDELIAN
USH2A	Retinitis pigmentosa	ORPHA:791	UNKNOWN
USH2A	Usher syndrome type 2	ORPHA:231178	UNKNOWN
USP1	—		UNKNOWN
USP15	—		UNKNOWN
USP24	—		UNKNOWN
USP26	Spermatogenic failure, X-linked, 6	OMIM:301101	MENDELIAN
USP27X	Intellectual developmental disorder, X-linked 105	OMIM:300984	MENDELIAN
USP3	—		UNKNOWN
USP30	—		UNKNOWN
USP34	—		UNKNOWN
USP44	—		UNKNOWN
USP46	—		UNKNOWN
USP53	Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss	OMIM:619658	MENDELIAN
USP54	—		UNKNOWN
USP7	Chromosome 16p13.2 deletion syndrome	OMIM:616863	MENDELIAN
USP7	Hao-Fountain syndrome due to 16p13.2 microdeletion	ORPHA:500055	UNKNOWN
USP8	Pituitary adenoma 4, ACTH-secreting, somatic	OMIM:219090	MENDELIAN
USP8	Autosomal recessive spastic paraplegia type 59	ORPHA:401795	UNKNOWN
USP8	Cushing disease	ORPHA:96253	UNKNOWN
USP9X	Intellectual developmental disorder, X-linked 99	OMIM:300919	MENDELIAN
USP9X	Intellectual developmental disorder, X-linked 99, syndromic, female-restricted	OMIM:300968	MENDELIAN
USP9X	X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability	ORPHA:480880	UNKNOWN
USP9Y	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
USP9Y	Chromosome Y microdeletion syndrome	ORPHA:1646	UNKNOWN
UST	—		UNKNOWN
UTF1	—		UNKNOWN
UTP14C	—		UNKNOWN
UTP6	—		UNKNOWN
UTRN	—		UNKNOWN
UTS2	—		UNKNOWN
UVSSA	UV-sensitive syndrome 3	OMIM:614640	MENDELIAN
VAMP1	Myasthenic syndrome, congenital, 25, presynaptic	OMIM:618323	MENDELIAN
VAMP1	Spastic ataxia 1, autosomal dominant	OMIM:108600	MENDELIAN
VAMP1	Autosomal dominant spastic ataxia type 1	ORPHA:251282	UNKNOWN
VAMP2	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements	OMIM:618760	MENDELIAN
VAMP7	46,XY partial gonadal dysgenesis	ORPHA:251510	UNKNOWN
VANGL1	Sacral defect with anterior meningocele	OMIM:600145	MENDELIAN
VANGL1	Neural tube defects, susceptibility to	OMIM:182940	POLYGENIC
VANGL1	Caudal regression syndrome	ORPHA:3027	UNKNOWN
VANGL2	Neural tube defects, susceptibility to	OMIM:182940	MENDELIAN
VANGL2	Isolated exencephaly	ORPHA:563612	UNKNOWN
VANGL2	Isolated anencephaly	ORPHA:563609	UNKNOWN
VAPB	Amyotrophic lateral sclerosis 8	OMIM:608627	MENDELIAN
VAPB	Spinal muscular atrophy, late-onset, Finkel type	OMIM:182980	MENDELIAN
VAPB	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
VAPB	Autosomal dominant adult-onset proximal spinal muscular atrophy	ORPHA:209335	UNKNOWN
VARS2	Combined oxidative phosphorylation deficiency 20	OMIM:615917	MENDELIAN
VAV3	—		UNKNOWN
VAX1	Microphthalmia, syndromic 11	OMIM:614402	MENDELIAN
VAX2	—		UNKNOWN
VCAM1	—		UNKNOWN
VCAN	Wagner vitreoretinopathy	OMIM:143200	MENDELIAN
VCAN	Wagner disease	ORPHA:898	UNKNOWN
VCL	Cardiomyopathy, dilated, 1W	OMIM:611407	MENDELIAN
VCL	Cardiomyopathy, familial hypertrophic, 15	OMIM:613255	MENDELIAN
VCL	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
VCP	Charcot-Marie-Tooth disease, axonal, type 2Y	OMIM:616687	MENDELIAN
VCP	Inclusion body myopathy with early-onset paget disease with or without frontotemporal dementia 1	OMIM:167320	MENDELIAN
VCP	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6	OMIM:613954	MENDELIAN
VCP	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	ORPHA:52430	UNKNOWN
VCP	Autosomal dominant Charcot-Marie-Tooth disease type 2Y	ORPHA:435387	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
VCP	Behavioral variant of frontotemporal dementia	ORPHA:275864	UNKNOWN
VCP	Frontotemporal dementia with motor neuron disease	ORPHA:275872	UNKNOWN
VCP	Adult-onset distal myopathy due to VCP mutation	ORPHA:329478	UNKNOWN
VCP	Spastic paraplegia-Paget disease of bone syndrome	ORPHA:329475	UNKNOWN
VCP	Amyotrophic lateral sclerosis	ORPHA:803	UNKNOWN
VCP	Progressive non-fluent aphasia	ORPHA:100070	UNKNOWN
VCX3A	—		UNKNOWN
VCY	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
VDAC1	—		UNKNOWN
VDR	Rickets, vitamin D-resistant, type IIA	OMIM:277440	MENDELIAN
VDR	Hypocalcemic vitamin D-resistant rickets	ORPHA:93160	UNKNOWN
VEGFC	Lymphatic malformation 4	OMIM:615907	MENDELIAN
VEZF1	Cardiomyopathy, dilated, 10O	OMIM:620247	MENDELIAN
VEZF1	Familial isolated dilated cardiomyopathy	ORPHA:154	UNKNOWN
VHL	Pheochromocytoma	OMIM:171300	MENDELIAN
VHL	von Hippel-Lindau syndrome	OMIM:193300	MENDELIAN
VHL	Renal cell carcinoma, nonpapillary	OMIM:144700	MENDELIAN
VHL	Erythrocytosis, familial, 2	OMIM:263400	MENDELIAN
VHL	Von Hippel-Lindau disease	ORPHA:892	UNKNOWN
VHL	Sporadic pheochromocytoma/secretory paraganglioma	ORPHA:276621	UNKNOWN
VHL	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	UNKNOWN
VIL1	—		UNKNOWN
VIM	Cataract 30, multiple types	OMIM:116300	MENDELIAN
VIMP	—		UNKNOWN
VIP	—		UNKNOWN
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2	OMIM:613404	MENDELIAN
VIPAS39	Arthrogryposis-renal dysfunction-cholestasis syndrome	ORPHA:2697	UNKNOWN
VIPR2	—		UNKNOWN
VKORC1	Coumarin resistance	OMIM:122700	MENDELIAN
VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, 2	OMIM:607473	MENDELIAN
VKORC1	Hereditary combined deficiency of vitamin K-dependent clotting factors	ORPHA:98434	UNKNOWN
VLDLR	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1	OMIM:224050	MENDELIAN
VLDLR	Dysequilibrium syndrome	ORPHA:1766	UNKNOWN
VMA21	Myopathy, X-linked, with excessive autophagy	OMIM:310440	MENDELIAN
VNN1	—		UNKNOWN
VPREB1	—		UNKNOWN
VPS13A	Choreoacanthocytosis	OMIM:200150	MENDELIAN
VPS13A	Choreoacanthocytosis	ORPHA:2388	UNKNOWN
VPS13B	Cohen syndrome	OMIM:216550	MENDELIAN
VPS13B	Cohen syndrome	ORPHA:193	UNKNOWN
VPS33B	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive	OMIM:620009	MENDELIAN
VPS33B	Cholestasis, progressive familial intrahepatic, 12	OMIM:620010	MENDELIAN
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1	OMIM:208085	MENDELIAN
VPS33B	Arthrogryposis-renal dysfunction-cholestasis syndrome	ORPHA:2697	UNKNOWN
VPS35	Parkinson disease 17	OMIM:614203	POLYGENIC
VPS35	Hereditary late-onset Parkinson disease	ORPHA:411602	UNKNOWN
VPS37A	Spastic paraplegia 53, autosomal recessive	OMIM:614898	MENDELIAN
VPS37A	Autosomal recessive spastic paraplegia type 53	ORPHA:319199	UNKNOWN
VPS45	Neutropenia, severe congenital, 5, autosomal recessive	OMIM:615285	MENDELIAN
VPS53	Pontocerebellar hypoplasia, type 2E	OMIM:615851	MENDELIAN
VPS54	—		UNKNOWN
VRK1	Neuronopathy, distal hereditary motor, autosomal recessive 10	OMIM:620542	MENDELIAN
VRK1	Pontocerebellar hypoplasia type 1A	OMIM:607596	MENDELIAN
VRK1	Pontocerebellar hypoplasia type 1	ORPHA:2254	UNKNOWN
VSIG1	—		UNKNOWN
VSIG4	—		UNKNOWN
VSX1	Craniofacial anomalies and anterior segment dysgenesis syndrome	OMIM:614195	MENDELIAN
VSX1	Keratoconus 1	OMIM:148300	MENDELIAN
VSX1	Posterior polymorphous corneal dystrophy	ORPHA:98973	UNKNOWN
VSX2	Microphthalmia with coloboma 3	OMIM:610092	MENDELIAN
VSX2	Microphthalmia, isolated 2	OMIM:610093	MENDELIAN
VTCN1	—		UNKNOWN
VTN	—		UNKNOWN
VWA3B	Spinocerebellar ataxia, autosomal recessive 22	OMIM:616948	MENDELIAN
VWF	von Willebrand disease, type 1	OMIM:193400	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
VWF	von Willebrand disease, type 3	OMIM:277480	MENDELIAN
VWF	Von willebrand disease, type 2	OMIM:613554	MENDELIAN
WAC	Desanto-Shinawi syndrome	OMIM:616708	MENDELIAN
WAC	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion	ORPHA:284169	UNKNOWN
WAC	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to WAC point mutation	ORPHA:466950	UNKNOWN
WAS	Wiskott-Aldrich syndrome	OMIM:301000	MENDELIAN
WAS	Neutropenia, severe congenital, X-linked	OMIM:300299	MENDELIAN
WAS	Thrombocytopenia 1	OMIM:313900	MENDELIAN
WAS	X-linked severe congenital neutropenia	ORPHA:86788	UNKNOWN
WAS	Wiskott-Aldrich syndrome	ORPHA:906	UNKNOWN
WASF1	Neurodevelopmental disorder with absent language and variable seizures	OMIM:618707	MENDELIAN
WASF3	—		UNKNOWN
WBSR17	—		UNKNOWN
WDFY3	Microcephaly 18, primary, autosomal dominant	OMIM:617520	MENDELIAN
WDFY4	—		UNKNOWN
WDPCP	Congenital heart defects, hamartomas of tongue, and polysyndactyly	OMIM:217085	MENDELIAN
WDPCP	Bardet-Biedl syndrome 15	OMIM:615992	MENDELIAN
WDPCP	Bardet-Biedl syndrome	ORPHA:110	UNKNOWN
WDPCP	Heart defect-tongue hamartoma-polysyndactyly syndrome	ORPHA:1338	UNKNOWN
WDR11	Hypogonadotropic hypogonadism 7 without anosmia	OMIM:146110	MENDELIAN
WDR11	Intellectual developmental disorder, autosomal recessive 78	OMIM:620237	MENDELIAN
WDR11	Hypogonadotropic hypogonadism 14 with or without anosmia	OMIM:614858	MENDELIAN
WDR11	Pituitary stalk interruption syndrome	ORPHA:95496	UNKNOWN
WDR11	Kallmann syndrome	ORPHA:478	UNKNOWN
WDR11	Normosmic congenital hypogonadotropic hypogonadism	ORPHA:432	UNKNOWN
WDR13	—		UNKNOWN
WDR19	Senior-Loken syndrome 8	OMIM:616307	MENDELIAN
WDR19	Cranioectodermal dysplasia 4	OMIM:614378	MENDELIAN
WDR19	Short-Rib thoracic dysplasia 5 with or without polydactyly	OMIM:614376	MENDELIAN
WDR19	Nephronophthisis 13	OMIM:614377	MENDELIAN
WDR19	Spermatogenic failure 72	OMIM:619867	MENDELIAN
WDR19	Senior-Loken syndrome	ORPHA:3156	UNKNOWN
WDR19	Cranioectodermal dysplasia	ORPHA:1515	UNKNOWN
WDR19	Jeune syndrome	ORPHA:474	UNKNOWN
WDR26	Skraban-Deardorff syndrome	OMIM:617616	MENDELIAN
WDR26	Intellectual disability-seizures-abnormal gait-facial dysmorphism syndrome	ORPHA:513456	UNKNOWN
WDR27	—		UNKNOWN
WDR31	—		UNKNOWN
WDR34	—		UNKNOWN
WDR35	Short-Rib thoracic dysplasia 7 with or without polydactyly	OMIM:614091	MENDELIAN
WDR35	Cranioectodermal dysplasia 2	OMIM:613610	MENDELIAN
WDR35	Cranioectodermal dysplasia	ORPHA:1515	UNKNOWN
WDR35	Short rib-polydactyly syndrome, Verma-Naumoff type	ORPHA:93271	UNKNOWN
WDR36	Glaucoma 1, open angle, G	OMIM:609887	MENDELIAN
WDR4	Microcephaly, growth deficiency, seizures, and brain malformations	OMIM:618346	MENDELIAN
WDR4	Galloway-Mowat syndrome 6	OMIM:618347	MENDELIAN
WDR4	Galloway-Mowat syndrome	ORPHA:2065	UNKNOWN
WDR45	Neurodegeneration with brain iron accumulation 5	OMIM:300894	MENDELIAN
WDR45	Beta-propeller protein-associated neurodegeneration	ORPHA:329284	UNKNOWN
WDR45	Infantile epileptic spasms syndrome	ORPHA:697160	UNKNOWN
WDR45B	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures	OMIM:617977	MENDELIAN
WDR48	Autosomal recessive spastic paraplegia type 60	ORPHA:401800	UNKNOWN
WDR5	—		UNKNOWN
WDR60	—		UNKNOWN
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations	OMIM:604317	MENDELIAN
WDR62	Autosomal recessive primary microcephaly	ORPHA:2512	UNKNOWN
WDR65	—		UNKNOWN
WDR66	—		UNKNOWN
WDR72	Amelogenesis imperfecta, hypomaturation type, iia3	OMIM:613211	MENDELIAN
WDR73	Galloway-Mowat syndrome 1	OMIM:251300	MENDELIAN
WDR73	CAMOS syndrome	ORPHA:83472	UNKNOWN
WDR73	Galloway-Mowat syndrome	ORPHA:2065	UNKNOWN
WDR81	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2	OMIM:610185	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
WDR81	Hydrocephalus, congenital, 3, with brain anomalies	OMIM:617967	MENDELIAN
WDR81	Dysequilibrium syndrome	ORPHA:1766	UNKNOWN
WDR93	—		UNKNOWN
WEE2	Oocyte maturation defect 5	OMIM:617996	MENDELIAN
WEE2	Female infertility due to oocyte meiotic arrest	ORPHA:488191	UNKNOWN
WFS1	Deafness, autosomal dominant 6	OMIM:600965	MENDELIAN
WFS1	Cataract, nuclear total	OMIM:116400	MENDELIAN
WFS1	Wolfram-like syndrome, autosomal dominant	OMIM:614296	MENDELIAN
WFS1	Wolfram syndrome 1	OMIM:222300	MENDELIAN
WFS1	Wolfram-like syndrome	ORPHA:411590	UNKNOWN
WFS1	Wolfram syndrome	ORPHA:3463	UNKNOWN
WHSC1	—		UNKNOWN
WHSC1L1	—		UNKNOWN
WIPF1	Wiskott-Aldrich syndrome 2	OMIM:614493	MENDELIAN
WISP3	—		UNKNOWN
WNK1	Neuropathy, hereditary sensory and autonomic, type II	OMIM:201300	MENDELIAN
WNK1	Pseudohypoaldosteronism, type IIC	OMIM:614492	MENDELIAN
WNK1	Hereditary sensory and autonomic neuropathy type 2	ORPHA:970	UNKNOWN
WNK3	Prieto syndrome	OMIM:309610	MENDELIAN
WNK4	Pseudohypoaldosteronism, type IIB	OMIM:614491	MENDELIAN
WNT1	Osteogenesis imperfecta, type XV	OMIM:615220	MENDELIAN
WNT1	Idiopathic juvenile osteoporosis	ORPHA:85193	UNKNOWN
WNT10A	Odontoonychodermal dysplasia	OMIM:257980	MENDELIAN
WNT10A	Schopf-Schulz-Passarge syndrome	OMIM:224750	MENDELIAN
WNT10A	Tooth agenesis, selective, 4	OMIM:150400	MENDELIAN
WNT10A	Autosomal recessive hypohidrotic ectodermal dysplasia	ORPHA:248	UNKNOWN
WNT10A	Schöpf-Schulz-Passarge syndrome	ORPHA:50944	UNKNOWN
WNT10A	Oligodontia	ORPHA:99798	UNKNOWN
WNT10B	Split-Hand/foot malformation 6	OMIM:225300	MENDELIAN
WNT10B	Tooth agenesis, selective, 8	OMIM:617073	MENDELIAN
WNT10B	Isolated split hand-split foot malformation	ORPHA:2440	UNKNOWN
WNT10B	Oligodontia	ORPHA:99798	UNKNOWN
WNT16	—		UNKNOWN
WNT3	Tetraamelia, autosomal recessive	OMIM:273395	MENDELIAN
WNT3	Tetraamelia-multiple malformations syndrome	ORPHA:3301	UNKNOWN
WNT4	Mullerian aplasia and hyperandrogenism	OMIM:158330	MENDELIAN
WNT4	46,XX sex reversal with dysgenesis of kidneys, adrenals, and lungs	OMIM:611812	MENDELIAN
WNT4	Mayer-Rokitansky-Küster-Hauser syndrome type 2	ORPHA:2578	UNKNOWN
WNT4	SERKAL syndrome	ORPHA:139466	UNKNOWN
WNT4	Müllerian aplasia and hyperandrogenism	ORPHA:247768	UNKNOWN
WNT5A	Robinow syndrome, autosomal dominant 1	OMIM:180700	MENDELIAN
WNT5A	Autosomal dominant Robinow syndrome	ORPHA:3107	UNKNOWN
WNT5B	—		UNKNOWN
WNT6	—		UNKNOWN
WNT7A	Ulna and fibula, absence of, with severe limb deficiency	OMIM:276820	MENDELIAN
WNT7A	Santos syndrome	OMIM:613005	MENDELIAN
WNT7A	Fibular aplasia or hypoplasia, femoral bowing and poly-, syn-, and oligodactyly	OMIM:228930	MENDELIAN
WNT7A	Phocomelia, Schinzel type	ORPHA:2879	UNKNOWN
WNT9A	—		UNKNOWN
WNT9B	Renal agenesis, bilateral	ORPHA:1848	UNKNOWN
WRAP53	Dyskeratosis congenita, autosomal recessive, 3	OMIM:613988	MENDELIAN
WRAP53	Dyskeratosis congenita	ORPHA:1775	UNKNOWN
WRN	Werner syndrome	OMIM:277700	MENDELIAN
WRN	Werner syndrome	ORPHA:902	UNKNOWN
WT1	Frasier syndrome	OMIM:136680	MENDELIAN
WT1	Nephrotic syndrome, type 4	OMIM:256370	MENDELIAN
WT1	Denys-Drash syndrome	OMIM:194080	MENDELIAN
WT1	Wilms tumor, aniridia, genitourinary anomalies and impaired intellectual development syndrome	OMIM:194072	MENDELIAN
WT1	Wilms tumor 1	OMIM:194070	MENDELIAN
WT1	Mesothelioma, malignant	OMIM:156240	MENDELIAN
WT1	Meacham syndrome	OMIM:608978	MENDELIAN
WT1	Aniridia	OMIM:106210	MENDELIAN
WT1	Frasier syndrome	ORPHA:347	UNKNOWN
WT1	Desmoplastic small round cell tumor	ORPHA:83469	UNKNOWN
WT1	46,XY partial gonadal dysgenesis	ORPHA:251510	UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
WT1	Denys-Drash syndrome	ORPHA:220	UNKNOWN
WT1	46,XY complete gonadal dysgenesis	ORPHA:242	UNKNOWN
WT1	Meacham syndrome	ORPHA:3097	UNKNOWN
WT1	WAGR syndrome	ORPHA:893	UNKNOWN
WT1	Nephroblastoma	ORPHA:654	UNKNOWN
WT1	Hereditary steroid-resistant nephrotic syndrome	ORPHA:656	UNKNOWN
WT1-AS	—		UNKNOWN
WWC2	—		UNKNOWN
WWC3	—		UNKNOWN
WWOX	Developmental and epileptic encephalopathy 28	OMIM:616211	MENDELIAN
WWOX	Spinocerebellar ataxia, autosomal recessive 12	OMIM:614322	MENDELIAN
WWOX	Esophageal cancer, somatic	OMIM:133239	MENDELIAN
WWOX	46,XY partial gonadal dysgenesis	ORPHA:251510	UNKNOWN
WWOX	Squamous cell carcinoma of the esophagus	ORPHA:99977	UNKNOWN
WWOX	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency	ORPHA:284282	UNKNOWN
XDH	Xanthinuria, type I	OMIM:278300	MENDELIAN
XG	—		UNKNOWN
XIAP	Lymphoproliferative syndrome, X-linked, 1	OMIM:308240	MENDELIAN
XIAP	Lymphoproliferative syndrome, X-linked, 2	OMIM:300635	MENDELIAN
XIAP	X-linked lymphoproliferative disease due to XIAP deficiency	ORPHA:538934	UNKNOWN
XIRP1	—		UNKNOWN
XIST	X-inactivation, familial skewed	OMIM:300087	MENDELIAN
XK	McLeod syndrome	OMIM:300842	MENDELIAN
XKR4	—		UNKNOWN
XKR6	—		UNKNOWN
XKRY	Spermatogenic failure, Y-linked, 2	OMIM:415000	MENDELIAN
XPA	Xeroderma pigmentosum, complementation group A	OMIM:278700	MENDELIAN
XPA	Xeroderma pigmentosum	ORPHA:910	UNKNOWN
XPC	Xeroderma pigmentosum, group C	OMIM:278720	MENDELIAN
XPC	Xeroderma pigmentosum	ORPHA:910	UNKNOWN
XPNPEP2	Acquired angioedema	OMIM:300909	POLYGENIC
XPNPEP2	Renin-angiotensin-aldosterone system-blocker-induced angioedema	ORPHA:100057	UNKNOWN
XPNPEP3	Nephronophthisis-Like nephropathy 1	OMIM:613159	MENDELIAN
XPO1	2p15p16.1 microdeletion syndrome	ORPHA:261349	UNKNOWN
XPO5	—		UNKNOWN
XRCC1	Spinocerebellar ataxia, autosomal recessive 26	OMIM:617633	MENDELIAN
XRCC2	Premature ovarian failure 17	OMIM:619146	MENDELIAN
XRCC2	Spermatogenic failure 50	OMIM:619145	MENDELIAN
XRCC2	Fanconi anemia, complementation group U	OMIM:617247	MENDELIAN
XRCC2	Fanconi anemia	ORPHA:84	UNKNOWN
XRCC2	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
XRCC3	Breast cancer	OMIM:114480	POLYGENIC
XRCC4	Short stature, microcephaly, and endocrine dysfunction	OMIM:616541	MENDELIAN
XRCC4	Microcephalic primordial dwarfism-insulin resistance syndrome	ORPHA:436182	UNKNOWN
XRCC5	—		UNKNOWN
XRCC6	—		UNKNOWN
XYLT1	Pseudoxanthoma elasticum	OMIM:264800	POLYGENIC
XYLT1	Desbuquois dysplasia 2	OMIM:615777	MENDELIAN
XYLT1	XYLT1-CDG	ORPHA:370930	UNKNOWN
XYLT1	Desbuquois syndrome	ORPHA:1425	UNKNOWN
XYLT2	Spondyloocular syndrome	OMIM:605822	MENDELIAN
XYLT2	Pseudoxanthoma elasticum	OMIM:264800	POLYGENIC
XYLT2	Spondylo-ocular syndrome	ORPHA:85194	UNKNOWN
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development	OMIM:120433	MENDELIAN
YAP1	Uveal coloboma-cleft lip and palate-intellectual disability	ORPHA:1473	UNKNOWN
YARS	—		UNKNOWN
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2	OMIM:613561	MENDELIAN
YARS2	Mitochondrial myopathy and sideroblastic anemia	ORPHA:2598	UNKNOWN
YBX2	—		UNKNOWN
YES1	—		UNKNOWN
YTHDF2	—		UNKNOWN
YWHAE	17p13.3 microduplication syndrome	ORPHA:217385	UNKNOWN
YWHAE	Miller-Dieker syndrome	ORPHA:531	UNKNOWN
YWHAG	Epileptic encephalopathy, early infantile, 56	OMIM:617665	MENDELIAN

Gene_symbol	Disease_name	Disease_ID	Association_type
YWHAG	Non-specific early-onset epileptic encephalopathy	ORPHA:442835	UNKNOWN
YY1	Gabriele-De vries syndrome	OMIM:617557	MENDELIAN
YY1	Insulinoma	ORPHA:97279	UNKNOWN
YY1	Gabriele-de Vries syndrome	ORPHA:506358	UNKNOWN
YY2	—		UNKNOWN
ZAN	—		UNKNOWN
ZAP70	Immunodeficiency 48	OMIM:269840	MENDELIAN
ZAP70	Autoimmune disease, multisystem, infantile-onset, 2	OMIM:617006	MENDELIAN
ZAP70	Combined immunodeficiency due to ZAP70 deficiency	ORPHA:911	UNKNOWN
ZBTB11	Intellectual developmental disorder, autosomal recessive 69	OMIM:618383	MENDELIAN
ZBTB16	Acute promyelocytic leukemia	ORPHA:520	UNKNOWN
ZBTB17	—		UNKNOWN
ZBTB18	Intellectual developmental disorder, autosomal dominant 22	OMIM:612337	MENDELIAN
ZBTB18	Distal deletion 1q syndrome	ORPHA:36367	UNKNOWN
ZBTB2	—		UNKNOWN
ZBTB20	Primrose syndrome	OMIM:259050	MENDELIAN
ZBTB20	Intellectual disability-cataracts-calcified pinnae-myopathy syndrome	ORPHA:3042	UNKNOWN
ZBTB21	—		UNKNOWN
ZBTB24	Immunodeficiency-Centromeric instability-facial anomalies syndrome2	OMIM:614069	MENDELIAN
ZBTB24	ICF syndrome	ORPHA:2268	UNKNOWN
ZBTB38	—		UNKNOWN
ZBTB40	—		UNKNOWN
ZBTB41	—		UNKNOWN
ZBTB42	Lethal congenital contracture syndrome 6	OMIM:616248	MENDELIAN
ZBTB7A	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin	OMIM:619769	MENDELIAN
ZC3H12B	—		UNKNOWN
ZC3H14	Intellectual developmental disorder, autosomal recessive 56	OMIM:617125	MENDELIAN
ZC3H3	—		UNKNOWN
ZC3HAV1	—		UNKNOWN
ZC4H2	Wieacker-Wolff syndrome	OMIM:314580	MENDELIAN
ZC4H2	Wieacker-Wolff syndrome, female-restricted	OMIM:301041	MENDELIAN
ZC4H2	Wieacker-Wolff syndrome	ORPHA:3454	UNKNOWN
ZCCHC12	—		UNKNOWN
ZCCHC13	—		UNKNOWN
ZCCHC16	—		UNKNOWN
ZCCHC8	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 5	OMIM:618674	MENDELIAN
ZDHHC15	—		UNKNOWN
ZDHHC17	—		UNKNOWN
ZDHHC2	—		UNKNOWN
ZDHHC8	—		UNKNOWN
ZDHHC9	Intellectual developmental disorder, X-linked syndromic, Raymond type	OMIM:300799	MENDELIAN
ZDHHC9	Lujan-Fryns syndrome	ORPHA:776	UNKNOWN
ZEB1	Corneal dystrophy, posterior polymorphous, 3	OMIM:609141	MENDELIAN
ZEB1	Corneal dystrophy, fuchs endothelial, 6	OMIM:613270	MENDELIAN
ZEB1	Posterior polymorphous corneal dystrophy	ORPHA:98973	UNKNOWN
ZEB1	Fuchs endothelial corneal dystrophy	ORPHA:98974	UNKNOWN
ZEB2	Mowat-Wilson syndrome	OMIM:235730	MENDELIAN
ZEB2	Mowat-Wilson syndrome due to a ZEB2 point mutation	ORPHA:261552	UNKNOWN
ZEB2	Mowat-Wilson syndrome due to monosomy 2q22	ORPHA:261537	UNKNOWN
ZEB2	Large/giant congenital melanocytic nevus	ORPHA:626	UNKNOWN
ZFAT	Autoimmune thyroid disease, susceptibility to, 3	OMIM:608175	POLYGENIC
ZFHX3	Atrial fibrillation 8, susceptibility to	OMIM:613055	POLYGENIC
ZFHX3	Epilepsy, idiopathic generalized 20	OMIM:621500	MENDELIAN
ZFHX3	Prostate cancer	OMIM:176807	MENDELIAN
ZFHX3	Spinocerebellar ataxia 4	OMIM:600223	MENDELIAN
ZFP36	—		UNKNOWN
ZFP36L1	—		UNKNOWN
ZFP36L2	Oocyte maturation defect 13	OMIM:620154	MENDELIAN
ZFP36L2	Female infertility due to oocyte meiotic arrest	ORPHA:488191	UNKNOWN
ZFP541	—		UNKNOWN
ZFP57	Diabetes mellitus, transient neonatal, 1	OMIM:601410	MENDELIAN
ZFP57	Transient neonatal diabetes mellitus	ORPHA:99886	UNKNOWN
ZFP69	—		UNKNOWN
ZFP90	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
ZFPL1	—		UNKNOWN
ZFPM2	Diaphragmatic hernia 3	OMIM:610187	MENDELIAN
ZFPM2	Tetralogy of Fallot	OMIM:187500	MENDELIAN
ZFPM2	46,xy sex reversal 9	OMIM:616067	MENDELIAN
ZFPM2	46,XY partial gonadal dysgenesis	ORPHA:251510	UNKNOWN
ZFPM2	Tetralogy of Fallot	ORPHA:3303	UNKNOWN
ZFPM2	Congenital diaphragmatic hernia	ORPHA:2140	UNKNOWN
ZFR	Autosomal recessive spastic paraplegia type 71	ORPHA:401840	UNKNOWN
ZFX	Intellectual developmental disorder, X-linked syndromic 37	OMIM:301118	MENDELIAN
ZFY	—		UNKNOWN
ZFYVE26	Spastic paraplegia 15, autosomal recessive	OMIM:270700	MENDELIAN
ZFYVE26	Kjellin syndrome	ORPHA:100996	UNKNOWN
ZFYVE27	—		UNKNOWN
ZHX3	—		UNKNOWN
ZIC1	Craniosynostosis 6	OMIM:616602	MENDELIAN
ZIC1	Structural brain anomalies with impaired intellectual development and craniosynostosis	OMIM:618736	MENDELIAN
ZIC1	Isolated Dandy-Walker malformation	ORPHA:217	UNKNOWN
ZIC1	Non-syndromic bicoronal craniosynostosis	ORPHA:35099	UNKNOWN
ZIC2	Holoprosencephaly 5	OMIM:609637	MENDELIAN
ZIC2	Septopreoptic holoprosencephaly	ORPHA:280195	UNKNOWN
ZIC2	Semilobar holoprosencephaly	ORPHA:220386	UNKNOWN
ZIC2	Microform holoprosencephaly	ORPHA:280200	UNKNOWN
ZIC2	Midline interhemispheric variant of holoprosencephaly	ORPHA:93926	UNKNOWN
ZIC2	Lobar holoprosencephaly	ORPHA:93924	UNKNOWN
ZIC2	Alobar holoprosencephaly	ORPHA:93925	UNKNOWN
ZIC3	VACTERL association, X-linked	OMIM:314390	MENDELIAN
ZIC3	Heterotaxy, visceral, 1, X-linked	OMIM:306955	MENDELIAN
ZIC4	Isolated Dandy-Walker malformation	ORPHA:217	UNKNOWN
ZIC5	—		UNKNOWN
ZKSCAN5	—		UNKNOWN
ZMIZ1	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies	OMIM:618659	MENDELIAN
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy	OMIM:608612	MENDELIAN
ZMPSTE24	Restrictive dermopathy 1	OMIM:275210	MENDELIAN
ZMPSTE24	Hutchinson-Gilford progeria syndrome	ORPHA:740	UNKNOWN
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy	ORPHA:90154	UNKNOWN
ZMPSTE24	Restrictive dermopathy	ORPHA:1662	UNKNOWN
ZMYM2	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities	OMIM:619522	MENDELIAN
ZMYM3	Intellectual developmental disorder, X-linked 112	OMIM:301111	MENDELIAN
ZMYM5	—		UNKNOWN
ZMYM6	—		UNKNOWN
ZMYND10	Ciliary dyskinesia, primary, 22	OMIM:615444	MENDELIAN
ZMYND10	Primary ciliary dyskinesia	ORPHA:244	UNKNOWN
ZMYND11	Intellectual developmental disorder, autosomal dominant 30	OMIM:616083	MENDELIAN
ZMYND15	Spermatogenic failure 14	OMIM:615842	MENDELIAN
ZMYND15	Male infertility with azoospermia or oligozoospermia due to single gene mutation	ORPHA:399805	UNKNOWN
ZMYND8	—		UNKNOWN
ZNF141	Polydactyly, postaxial, type A6	OMIM:615226	MENDELIAN
ZNF157	—		UNKNOWN
ZNF175	—		UNKNOWN
ZNF202	—		UNKNOWN
ZNF213	—		UNKNOWN
ZNF217	—		UNKNOWN
ZNF224	—		UNKNOWN
ZNF24	—		UNKNOWN
ZNF292	Intellectual developmental disorder, autosomal dominant 64	OMIM:619188	MENDELIAN
ZNF311	—		UNKNOWN
ZNF335	Microcephaly 10, primary, autosomal recessive	OMIM:615095	MENDELIAN
ZNF335	Microcephalic primordial dwarfism due to ZNF335 deficiency	ORPHA:329228	UNKNOWN
ZNF33A	—		UNKNOWN
ZNF347	—		UNKNOWN
ZNF350	—		UNKNOWN
ZNF365	Narcolepsy type 2	ORPHA:83465	UNKNOWN
ZNF365	Narcolepsy type 1	ORPHA:2073	UNKNOWN
ZNF385B	—		UNKNOWN
ZNF395	—		UNKNOWN

Gene_symbol	Disease_name	Disease_ID	Association_type
ZNF407	SIMHA syndrome	OMIM:619557	MENDELIAN
ZNF408	Retinitis pigmentosa 72	OMIM:616469	MENDELIAN
ZNF408	Exudative vitreoretinopathy 6	OMIM:616468	MENDELIAN
ZNF408	Familial exudative vitreoretinopathy	ORPHA:891	UNKNOWN
ZNF408	Retinitis pigmentosa	ORPHA:791	UNKNOWN
ZNF41	—		UNKNOWN
ZNF419	—		UNKNOWN
ZNF420	—		UNKNOWN
ZNF423	Nephronophthisis 14	OMIM:614844	MENDELIAN
ZNF423	Joubert syndrome with oculorenal defect	ORPHA:2318	UNKNOWN
ZNF433	—		UNKNOWN
ZNF44	—		UNKNOWN
ZNF45	—		UNKNOWN
ZNF451	—		UNKNOWN
ZNF462	Weiss-Kruszka syndrome	OMIM:618619	MENDELIAN
ZNF462	Weiss-Kruszka Syndrome	ORPHA:502430	UNKNOWN
ZNF469	Brittle cornea syndrome	OMIM:229200	MENDELIAN
ZNF469	Brittle cornea syndrome	ORPHA:90354	UNKNOWN
ZNF480	—		UNKNOWN
ZNF507	—		UNKNOWN
ZNF513	Retinitis pigmentosa 58	OMIM:613617	MENDELIAN
ZNF513	Retinitis pigmentosa	ORPHA:791	UNKNOWN
ZNF517	—		UNKNOWN
ZNF526	Dentici-Novelli neurodevelopmental syndrome	OMIM:619877	MENDELIAN
ZNF582	—		UNKNOWN
ZNF592	CAMOS syndrome	ORPHA:83472	UNKNOWN
ZNF599	—		UNKNOWN
ZNF610	—		UNKNOWN
ZNF627	—		UNKNOWN
ZNF638	—		UNKNOWN
ZNF644	Myopia 21, autosomal dominant	OMIM:614167	MENDELIAN
ZNF668	Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies	OMIM:620194	MENDELIAN
ZNF674	—		UNKNOWN
ZNF683	—		UNKNOWN
ZNF703	—		UNKNOWN
ZNF711	Intellectual developmental disorder, X-linked 97	OMIM:300803	MENDELIAN
ZNF713	—		UNKNOWN
ZNF750	Seborrhea-Like dermatitis with psoriasiform elements	OMIM:610227	MENDELIAN
ZNF75D	—		UNKNOWN
ZNF77	—		UNKNOWN
ZNF778	—		UNKNOWN
ZNF780B	—		UNKNOWN
ZNF783	—		UNKNOWN
ZNF79	—		UNKNOWN
ZNF80	—		UNKNOWN
ZNF804A	—		UNKNOWN
ZNF813	—		UNKNOWN
ZNF865	—		UNKNOWN
ZNHIT6	—		UNKNOWN
ZNRF1	—		UNKNOWN
ZP1	Oocyte maturation defect	OMIM:615774	MENDELIAN
ZP2	Oocyte maturation defect 6	OMIM:618353	MENDELIAN
ZP3	Oocyte maturation defect 3	OMIM:617712	MENDELIAN
ZBPB	Spermatogenic failure 66	OMIM:619799	MENDELIAN
ZBPB2	—		UNKNOWN
ZPLD1	—		UNKNOWN
ZRSR2	Orofaciodigital syndrome XXI	OMIM:301132	MENDELIAN
ZSCAN29	—		UNKNOWN
ZSWIM6	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features	OMIM:617865	MENDELIAN
ZSWIM6	Acromelic frontonasal dysostosis	OMIM:603671	MENDELIAN
ZSWIM6	Acromelic frontonasal dysplasia	ORPHA:1827	UNKNOWN
ZXDA	—		UNKNOWN