ORPHA-code	Disease Name
276413	10q22.3q23.3 microdeletion syndrome
261183	15q11.2 microdeletion syndrome
238446	15q11q13 microduplication syndrome
199318	15q13.3 microdeletion syndrome
261211	16p11.2p12.2 microdeletion syndrome
261236	16p13.11 microdeletion syndrome
261243	16p13.11 microduplication syndrome
1713	17p11.2 microduplication syndrome
261265	17q12 microdeletion syndrome
261272	17q12 microduplication syndrome
363958	17q21.31 microdeletion syndrome
261279	17q23.1q23.2 microdeletion syndrome
293948	1p21.3 microdeletion syndrome
1606	1p36 deletion syndrome
250989	1q21.1 microdeletion syndrome
250994	1q21.1 microduplication syndrome
567	22q11.2 deletion syndrome
1727	22q11.2 microduplication syndrome
1001	2q37 microdeletion syndrome
20	3-hydroxy-3-methylglutaric aciduria
65286	3q29 microdeletion syndrome
251038	3q29 microduplication
8	47,XYY syndrome
96072	4p16.3 microduplication syndrome
75857	6q terminal deletion syndrome
314034	7p22.1 microduplication syndrome
96121	7q11.23 microduplication syndrome
251076	8p23.1 duplication syndrome
251071	8p23.1 microdeletion syndrome
915	Aarskog-Scott syndrome
15	Achondroplasia
228285	Acquired cutis laxa
37	Acrodermatitis enteropathica
955	Acroosteolysis dominant type
404448	ADNP-related multiple congenital anomalies-intellectual disability-autism spectrum disorder
100091	Adrenal/paraganglial tumor
139399	Adrenomyeloneuropathy
261619	Alagille syndrome due to a JAG1 point mutation
60	Alpha-1-antitrypsin deficiency
63	Alport syndrome
803	Amyotrophic lateral sclerosis
284984	Aneurysm-osteoarthritis syndrome
72	Angelman syndrome
98794	Angelman syndrome due to maternal 15q11q13 deletion
2346	Angioosteohypertrophic syndrome
77	Aniridia
1114	Aplasia cutis congenita
247	Arrhythmogenic right ventricular cardiomyopathy

109007	Arthrogryposis syndrome
1180	Ataxia-hypogonadism-choroidal dystrophy syndrome
100	Ataxia-telangiectasia
1190	Atelosteogenesis type I
220460	Attenuated familial adenomatous polyposis
2134	Atypical hemolytic-uremic syndrome
93579	Atypical hemolytic-uremic syndrome with H factor anomaly
3095	Atypical Rett syndrome
352490	Autism spectrum disorder due to AUTS2 deficiency
98127	Autosomal anomaly
88918	Autosomal dominant Alport syndrome
99943	Autosomal dominant Charcot-Marie-Tooth disease type 2J
488333	Autosomal dominant Charcot-Marie-Tooth disease type 2W
98808	Autosomal dominant dopa-responsive dystonia
90635	Autosomal dominant non-syndromic sensorineural deafness type DFNA
98672	Autosomal dominant optic atrophy
98673	Autosomal dominant optic atrophy, classic form
88924	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis
100985	Autosomal dominant spastic paraplegia type 4
34149	Autosomal dominant tubulointerstitial kidney disease
91024	Autosomal recessive axonal hereditary motor and sensory neuropathy
139455	Autosomal recessive bestrophinopathy
281097	Autosomal recessive congenital ichthyosis
402041	Autosomal recessive distal renal tubular acidosis
248	Autosomal recessive hypohidrotic ectodermal dysplasia
102015	Autosomal recessive limb-girdle muscular dystrophy
88616	Autosomal recessive non-syndromic intellectual disability
90636	Autosomal recessive non-syndromic sensorineural deafness type DFNB
731	Autosomal recessive polycystic kidney disease
110	Bardet-Biedl syndrome
100976	Bathing suit ichthyosis
98895	Becker muscular dystrophy
116	Beckwith-Wiedemann syndrome
231117	Beckwith-Wiedemann syndrome due to imprinting defect of 11p15
96193	Beckwith-Wiedemann syndrome due to paternal uniparental disomy of chromosome 11
306	Benign familial infantile epilepsy
166311	Benign partial infantile seizures
610	Bethlem myopathy
101070	Bilateral frontoparietal polymicrogyria
1980	Bilateral striopallidodentate calcinosis
122	Birt-Hogg-Dubé syndrome
261572	Blepharophimosis-epicanthus inversus-ptosis due to a point mutation syndrome
3047	Blepharophimosis-intellectual disability syndrome, SBBYS type
97297	Bohring-Opitz syndrome
209905	Brain-lung-thyroid syndrome
2771	Bruck syndrome
130	Brugada syndrome
136	CADASIL
1328	Camurati-Engelmann disease
137667	Capillary malformation-arteriovenous malformation

1340	Cardiofaciocutaneous syndrome
3286	Catecholaminergic polymorphic ventricular tachycardia
195	Cat-eye syndrome
1756	Caudal duplication
597	Central core disease
88642	Channelopathy-associated congenital insensitivity to pain
101081	Charcot-Marie-Tooth disease type 1A
138	CHARGE syndrome
167	Chédiak-Higashi syndrome
247667	Childhood-onset hypophosphatasia
68335	Chromosomal anomaly
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form
79239	Classic galactosemia
216866	Classic pantothenate kinase-associated neurodegeneration
79254	Classic phenylketonuria
199306	Cleft lip/palate
228363	CLN6 disease
1465	Coffin-Siris syndrome
193	Cohen syndrome
36383	COL4A1-related familial vascular leukoencephalopathy
97556	Congenital and infantile nephrotic syndrome
115	Congenital contractural arachnodactyly
327	Congenital factor VII deficiency
590	Congenital myasthenic syndrome
201	Cowden syndrome
1520	Craniofrontonasal dysplasia
1522	Craniometaphyseal dysplasia
207	Crouzon disease
586	Cystic fibrosis
2111	Cystic hamartoma of lung and kidney
214	Cystinuria
1652	Dent disease
166260	Dentinogenesis imperfecta type 2
261222	Distal 16p11.2 microdeletion syndrome
261330	Distal 22q11.2 microdeletion syndrome
261337	Distal 22q11.2 microduplication syndrome
139536	Distal hereditary motor neuropathy type 5
96148	Distal monosomy 10q
1590	Distal monosomy 13q
96129	Distal monosomy 19p13.3
96125	Distal monosomy 6p
1636	Distal monosomy 7q36
1642	Distal monosomy 9p
96102	Distal trisomy 10q
1717	Distal trisomy 19q
96096	Distal trisomy 4q
293939	Distal Xq28 microduplication syndrome
870	Down syndrome
262	Duchenne and Becker muscular dystrophy
3306	Duplication/inversion 15q11

1934	Early infantile epileptic encephalopathy
289266	Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation
79373	Ectodermal dysplasia syndrome
1892	Ectrodactyly-polydactyly syndrome
98249	Ehlers-Danlos syndrome
90309	Ehlers-Danlos syndrome type 1
287	Ehlers-Danlos syndrome, classic type
285	Ehlers-Danlos syndrome, hypermobility type
2953	Ehlers-Danlos syndrome, musculocontractural type
286	Ehlers-Danlos syndrome, vascular type
230845	Ehlers-Danlos syndrome, vascular-like type
289	Ellis Van Creveld syndrome
96170	Emanuel syndrome
296	Enchondromatosis
79397	Epidermolysis bullosa simplex with mottled pigmentation
2199	Epidermolytic palmoplantar keratoderma
324	Fabry disease
466950	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to WAC point mutation
269	Facioscapulohumeral dystrophy
733	Familial adenomatous polyposis
229	Familial aortic dissection
404560	Familial atypical multiple mole melanoma syndrome
221061	Familial cerebral cavernous malformation
217607	Familial dilated cardiomyopathy
405	Familial hypocalciuric hypercalcemia
154	Familial isolated dilated cardiomyopathy
75249	Familial isolated restrictive cardiomyopathy
209886	Familial juvenile hyperuricemic nephropathy type 1
768	Familial long QT syndrome
342	Familial Mediterranean fever
741	Familial mitral valve prolapse
569	Familial or sporadic hemiplegic migraine
97	Familial paroxysmal ataxia
98306	Familial partial lipodystrophy
2348	Familial partial lipodystrophy, Dunnigan type
1767	Familial progressive vestibulocochlear dysfunction
1331	Familial prostate cancer
91387	Familial thoracic aortic aneurysm and aortic dissection
95716	Familial thyroid dyshormonogenesis
101039	Female restricted epilepsy with intellectual disability
2019	Femur-fibula-ulna complex
1915	Fetal alcohol syndrome
1906	Fetal valproate syndrome
480773	Fibular aplasia-tibial campomelia-oligosyndactyly syndrome
2044	Floating-Harbor syndrome
908	Fragile X syndrome
93256	Fragile X-associated tremor/ataxia syndrome
95	Friedreich ataxia
282	Frontotemporal dementia
275872	Frontotemporal dementia with motor neuron disease

2065	Galloway-Mowat syndrome
36387	Generalized epilepsy with febrile seizures-plus
3221	Generalized resistance to thyroid hormone
183497	Genetic neuromuscular disease
358	Gitelman syndrome
25	Glutaryl-CoA dehydrogenase deficiency
366	Glycogen storage disease due to glycogen debranching enzyme deficiency
354	GM1 gangliosidosis
374	Goldenhar syndrome
377	Gorlin syndrome
380	Greig cephalopolysyndactyly syndrome
181393	Growth hormone insensitivity syndrome
2108	Hallermann-Streiff syndrome
73229	HANAC syndrome
79230	Hemochromatosis type 2
93616	Hemoglobin H disease
766	Hemolytic anemia due to red cell pyruvate kinase deficiency
98878	Hemophilia A
100050	Hereditary angioedema type 1
145	Hereditary breast and ovarian cancer syndrome
227535	Hereditary breast cancer
676	Hereditary chronic pancreatitis
26106	Hereditary diffuse gastric cancer
313808	Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia
774	Hereditary hemorrhagic telangiectasia
640	Hereditary neuropathy with liability to pressure palsies
29072	Hereditary pheochromocytoma-paraganglioma
685	Hereditary spastic paraplegia
231512	Hermansky-Pudlak syndrome without pulmonary fibrosis
388	Hirschsprung disease
392	Holt-Oram syndrome
391665	Homozygous familial hypercholesterolemia
399	Huntington disease
217569	Hypertrophic cardiomyopathy
429	Hypochondroplasia
2237	Hypoparathyroidism-deafness-renal disease syndrome
70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency
200418	Immunodeficiency with factor I anomaly
464	Incontinentia pigmenti
319462	Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations
289548	Inherited isolated adrenal insufficiency due to partial CYP11A1 deficiency
252190	Inherited nervous system cancer-predisposing syndrome
464311	Intellectual disability syndrome due to a DYRK1A point mutation
391372	Intellectual disability-severe speech delay-mild dysmorphism syndrome
1478	Interatrial communication
85200	Ischio-vertebral syndrome
250923	Isolated aniridia
1885	Isolated ectopia lentis
231679	Isolated growth hormone deficiency type II
2345	Isolated Klippel-Feil syndrome

3366	Isolated trigonocephaly
474	Jeune syndrome
475	Joubert syndrome
79404	Junctional epidermolysis bullosa, Herlitz type
2322	Kabuki syndrome
478	Kallmann syndrome
480	Kearns-Sayre syndrome
261494	Kleefstra syndrome
96147	Kleefstra syndrome due to 9q34 microdeletion
261652	Kleefstra syndrome due to a point mutation
96169	Koolen-De Vries syndrome
99749	Kostmann syndrome
89838	KRT14-related epidermolysis bullosa simplex
79314	L-2-hydroxyglutaric aciduria
104	Leber hereditary optic neuropathy
99718	Leber plus disease
54260	Left ventricular noncompaction
137605	Legius syndrome
240	Léri-Weill dyschondrosteosis
2612	Linear nevus sebaceus syndrome
48471	Lissencephaly
60030	Loeys-Dietz syndrome
69663	Low phospholipid associated cholelithiasis
295051	Lower limb hypertrophy
144	Lynch syndrome
275761	Lysosomal acid lipase deficiency
79457	Maculopapular cutaneous mastocytosis
163634	Maffucci syndrome
79113	Mandibulofacial dysostosis-microcephaly syndrome
284993	Marfan and Marfan-related disorder
558	Marfan syndrome
284963	Marfan syndrome type 1
96181	Maternal uniparental disomy of chromosome 6
562	McCune-Albright syndrome
85281	MECP2 duplication syndrome
42	Medium chain acyl-CoA dehydrogenase deficiency
60040	Megalencephaly-capillary malformation-polymicrogyria syndrome
550	MELAS
512	Metachromatic leukodystrophy
79282	Methylmalonic acidemia with homocystinuria, type cblC
79253	Mild phenylketonuria
552	MODY
96168	Monosomy 13q34
1598	Monosomy 18p
1600	Monosomy 18q
281	Monosomy 5p
96061	Mosaic trisomy 8
99776	Mosaic trisomy 9
2152	Mowat-Wilson syndrome
261552	Mowat-Wilson syndrome due to a ZEB2 point mutation

261537	Mowat-Wilson syndrome due to monosomy 2q22
88949	MUC1-related autosomal dominant tubulointerstitial kidney disease
217085	Mucopolysaccharidosis type 2, severe form
587	Muir-Torre syndrome
652	Multiple endocrine neoplasia type 1
321	Multiple osteochondromas
98473	Muscular dystrophy
247798	MUTYH-related attenuated familial adenomatous polyposis
2588	Myhre syndrome
1942	Myoclonic-astastic epilepsy
2614	Nail-patella syndrome
223	Nephrogenic diabetes insipidus
636	Neurofibromatosis type 1
363700	Neurofibromatosis type 1 due to NF1 mutation or intragenic deletion
637	Neurofibromatosis type 2
93921	Neurofibromatosis type 3
2149	Nodular neuronal heterotopia
206656	Non-dystrophic myopathy
217598	Non-familial hypertrophic cardiomyopathy
329918	Non-immunoglobulin-mediated membranoproliferative glomerulonephritis
93457	Non-syndromic limb reduction defect
648	Noonan syndrome
500	Noonan syndrome with multiple lentigines
2701	Noonan syndrome-like disorder with loose anagen hair
432	Normosmic congenital hypogonadotropic hypogonadism
71529	Obesity due to melanocortin 4 receptor deficiency
198	Occipital horn syndrome
55	Oculocutaneous albinism
79432	Oculocutaneous albinism type 2
2710	Oculodentodigital dysplasia
2712	Oculofaciocardiodental syndrome
270	Oculopharyngeal muscular dystrophy
99798	Oligodontia
2750	Orofaciodigital syndrome type 1
666	Osteogenesis imperfecta
216796	Osteogenesis imperfecta type 1
216812	Osteogenesis imperfecta type 3
216820	Osteogenesis imperfecta type 4
216828	Osteogenesis imperfecta type 5
684	Paramyotonia congenita of Von Eulenburg
98809	Paroxysmal kinesigenic dyskinesia
447	Paroxysmal nocturnal hemoglobinuria
262092	Partial deletion of the long arm of chromosome 11
262101	Partial deletion of the long arm of chromosome 13
262128	Partial deletion of the long arm of chromosome 16
262010	Partial deletion of the long arm of chromosome 2
262173	Partial deletion of the long arm of chromosome 21
262182	Partial deletion of the long arm of chromosome 22
262029	Partial deletion of the long arm of chromosome 4
263756	Partial deletion of the long arm of chromosome X

261857	Partial deletion of the short arm of chromosome 1
316244	Partial deletion of the short arm of chromosome 12
261956	Partial deletion of the short arm of chromosome 16
261974	Partial deletion of the short arm of chromosome 18
261983	Partial deletion of the short arm of chromosome 19
261866	Partial deletion of the short arm of chromosome 2
262833	Partial duplication of the long arm of chromosome 1
262950	Partial duplication of the long arm of chromosome 15
262968	Partial duplication of the long arm of chromosome 17
262860	Partial duplication of the long arm of chromosome 4
263783	Partial duplication of the long arm of chromosome X
262794	Partial duplication of the short arm of chromosome 16
262698	Partial duplication of the short arm of chromosome 2
263775	Partial duplication of the short arm of chromosome X
262083	Partial monosomy of the long arm of chromosome 10
262959	Partial trisomy of the long arm of chromosome 16
262869	Partial trisomy of the long arm of chromosome 5
262905	Partial trisomy of the long arm of chromosome 9
2836	PEHO syndrome
280229	Pelizaeus-Merzbacher disease in female carriers
705	Pendred syndrome
98892	Periventricular nodular heterotopia
42775	PHACE syndrome
2875	Phakomatosis pigmentovascularis
221150	Pitt-Hopkins-like syndrome
95496	Pituitary stalk interruption syndrome
2897	Pityriasis rubra pilaris
2911	Poland syndrome
98913	Postsynaptic congenital myasthenic syndromes
739	Prader-Willi syndrome
177910	Prader-Willi syndrome due to imprinting mutation
98754	Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15
98793	Prader-Willi syndrome due to paternal 15q11q13 deletion
177901	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1
93446	Primary bone dysplasia with decreased bone density
93598	Primary hyperoxaluria type 1
30924	Primary hypomagnesemia with secondary hypocalcemia
1214	Progressive hemifacial atrophy
261197	Proximal 16p11.2 microdeletion syndrome
370079	Proximal 16p11.2 microduplication syndrome
606	Proximal myotonic myopathy
83330	Proximal spinal muscular atrophy type 1
750	Pseudoachondroplasia
94089	Pseudohypoparathyroidism type 1B
758	Pseudoxanthoma elasticum
306498	PTEN hamartoma tumor syndrome
320335	Pure or complex hereditary spastic paraplegia
760	Purine nucleoside phosphorylase deficiency
93419	Rare bone disease
97929	Rare cardiac disease

93890	Rare developmental defect during embryogenesis
180772	Rare disease with autism
285014	Rare disease with thoracic aortic aneurysm and aortic dissection
370106	Rare disorder with dystonia and other neurologic or systemic manifestation
97978	Rare endocrine disease
99739	Rare familial disorder with hypertrophic cardiomyopathy
98054	Rare genetic cardiac disease
98053	Rare genetic disease
101435	Rare genetic eye disease
71859	Rare genetic neurological disorder
183763	Rare genetic syndromic intellectual disability
477811	Rare hypercholesterolemia
87277	Rare intellectual disability
182070	Rare neurodegenerative disease
101685	Rare non-syndromic intellectual disability
79386	Rare skin tumor or hamartoma
102369	Rare syndromic intellectual disability
461	Recessive X-linked ichthyosis
93111	Renal cysts and diabetes syndrome
790	Retinoblastoma
778	Rett syndrome
1442	Ring chromosome 18 syndrome
2909	Rothmund-Thomson syndrome
794	Saethre-Chotzen syndrome
84081	Senior-Boichis syndrome
3157	Septo-optic dysplasia spectrum
83618	Severe dilated cardiomyopathy due to lamin A/C mutation
1505	Short rib-polydactyly syndrome
314795	SHOX-related short stature
232	Sickle cell anemia
813	Silver-Russell syndrome
231144	Silver-Russell syndrome due to 11p15 microduplication
231140	Silver-Russell syndrome due to an imprinting defect of 11p15
819	Smith-Magenis syndrome
821	Sotos syndrome
98756	Spinocerebellar ataxia type 2
98764	Spinocerebellar ataxia type 27
64753	Spinocerebellar ataxia with axonal neuropathy type 2
2440	Split hand-split foot malformation
273	Steinert myotonic dystrophy
828	Stickler syndrome
50942	Striate palmoplantar keratoderma
3205	Sturge-Weber syndrome
99796	Subcortical band heterotopia
3193	Supravalvular aortic stenosis
1570	Symbrachydactyly of hands and feet
217595	Syndrome associated with hypertrophic cardiomyopathy
85279	Syndromic X-linked intellectual disability due to JARID1C mutation
404443	Tall stature-intellectual disability-facial dysmorphism syndrome
252018	Teratoma of the central nervous system

884	Tetrasomy 12p
3307	Tetrasomy 18p
614	Thomsen and Becker disease
93573	Thrombotic microangiopathy
3339	Toriello-Lacassie-Droste syndrome
857	Townes-Brocks syndrome
420611	Transient myeloproliferative syndrome
32960	TRAPS syndrome
861	Treacher-Collins syndrome
3377	Trismus-pseudocamptodactyly syndrome
3378	Trisomy 13
236	Trisomy 9p
3375	Trisomy X
805	Tuberous sclerosis complex
881	Turner syndrome
3138	Ulnar-mammary syndrome
308	Unverricht-Lundborg disease
231169	Usher syndrome type 1
887	VACTERL/VATER association
247871	Vitiligo-associated autoimmune disease
892	Von Hippel-Lindau disease
3440	Waardenburg syndrome
897	Waardenburg-Shah syndrome
904	Williams syndrome
905	Wilson disease
280	Wolf-Hirschhorn syndrome
411590	Wolfram-like syndrome
88917	X-linked Alport syndrome
64747	X-linked Charcot-Marie-Tooth disease
101075	X-linked Charcot-Marie-Tooth disease type 1
35173	X-linked dominant chondrodysplasia punctata
181	X-linked hypohidrotic ectodermal dysplasia
89936	X-linked hypophosphatemia
281210	X-linked ichthyosis syndrome
163937	X-linked intellectual disability, Najm type
85287	X-linked intellectual disability, Siderius type
85328	X-linked intellectual disability, Turner type
163976	X-linked intellectual disability, Van Esch type
383	X-linked mixed deafness with perilymphatic gusher
777	X-linked non-syndromic intellectual disability