Orphanet Report Series

Rare Diseases collection

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List of rare diseases and synonyms

Listed in alphabetical order

www.orpha.net

Instituts thématiques Inserm
Institut national de la santé et de la recherche médicale

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Rare diseases listed in alphabetical order

ORPHA Number	Disease name
289157	1-alpha-hydroxylase deficiency
293948	1p21.3 microdeletion syndrome
1606	1p36 deletion syndrome
250989	1q21.1 microdeletion syndrome
250994	1q21.1 microduplication syndrome
250999	1q41q42 microdeletion syndrome
250999	1q41-q42 microdeletion syndrome
238769	1q44 microdeletion syndrome
976	2,8 dihydroxyadenine urolithiasis
869	2A syndrome
79154	2-aminoadipic 2-oxoadipic aciduria
19	2-hydroxyglutaric acidemia
19	2-hydroxyglutaric aciduria
35123	2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency
79095	2-methylacyl-CoA racemase deficiency
79157	2-methylbutyric aciduria
79157	2-methylbutyryl-CoA dehydrogenase deficiency
255182	2-oxoglutarate complex deficiency
261349	2p15p16.1 microdeletion syndrome
261349	2p15-p16.1 microdeletion syndrome
163693	2p21 deletion
163693	2p21 microdeletion syndrome
228402	2q23.1 microdeletion syndrome
313947	2q23.1 microduplication syndrome
1617	2q24 microdeletion syndrome
251014	2q31.1 microdeletion syndrome
294026	2q31.1 microduplication syndrome
251019	2q32q33 microdeletion syndrome
251019	2q32-q33 microdeletion syndrome
251028	2q33.1 microdeletion syndrome
1001	2q37 microdeletion syndrome
869	3A syndrome
79301	3-beta-hydroxy-delta-5-C27-steroid oxidoreductase deficiency
7	3C syndrome
35123	3-hydroxy-2-methylbutyryl-CoA dehydrogenase deficiency
20	3-hydroxy-3-methylglutaric aciduria
20	3-hydroxy-3-methylglutaryl-CoA lyase deficiency
35701	3-hydroxy-3-methylglutaryl-CoA synthase deficiency
309127	3-hydroxyacyl-CoA dehydrogenase deficiency
939	3-hydroxyisobutyric aciduria
2616	3M syndrome
2616	3-M syndrome
293843	3MC syndrome
6	3-methylcrotonylglycinuria
67046	3-methylglutaconic aciduria type 1
111	3-methylglutaconic aciduria type 2
67047	3-methylglutaconic aciduria type 3
67048	3-methylglutaconic aciduria type 4
66634	3-methylglutaconic aciduria type 5
352328	3-methylglutaconic aciduria with deafness - encephalopathy - Leigh-like syndrome
67046	3-methylglutaconyl-CoA hydratase deficiency
67046	3MG-CoA hydratase deficiency
79351	3-phosphoglycerate dehydrogenase deficiency
79350	3-phosphoserine phosphatase deficiency
65286	3q subtelomere deletion syndrome
1621	3q13 microdeletion syndrome
96095	3q26 microduplication syndrome
356947	3q26q27 microdeletion syndrome
356947	3q26-q27microdeletion syndrome
65286	3q29 microdeletion syndrome
251038	3q29 microduplication
65286	3qter deletion
2118	4-alpha-hydroxyphenylpyruvate hydroxylase deficiency
88637	4H syndrome
2118	4-HPPD deficiency
22	4-hydroxybutyric aciduria
2118	4-hydroxyphenylpyruvic acid dioxygenase deficiency
280	4p- syndrome
238750	4q21 microdeletion syndrome
250977	5-amino-4-imidazole carboxamide ribosiduria
217064	5-fluorouracil intoxication
217064	5-fluorouracil poisoning
33572	5-oxoprolinase deficiency
329802	5p13 microduplication syndrome
86841	5q- syndrome
228384	5q14.3 microdeletion syndrome
314655	5q31.3 microdeletion syndrome
228415	5q35 microduplication syndrome
96125	6p subtelomeric deletion syndrome
251046	6p22 microdeletion syndrome
96125	6p25 microdeletion syndrome
99135	6-phosphogluconate dehydrogenase deficiency
13	6-pyruvoyl-tetrahydropterin synthase deficiency
75857	6q terminal deletion
171829	6q16 deletion syndrome
251056	6q25 microdeletion syndrome
818	7-dehydrocholesterol reductase deficiency
314034	7p22.1 microduplication syndrome
96121	7q11.23 microduplication syndrome
251061	7q31 microdeletion syndrome
96092	8p inverted duplication/deletion syndrome
168953	8p11 myeloproliferative syndrome
251066	8p11.2 deletion syndrome
251071	8p23.1 microdeletion syndrome
251076	8p23.1 microduplication syndrome
228399	8q12 microduplication syndrome
2496	8q13 microdeletion syndrome
284160	8q21.11 microdeletion syndrome
178303	8q22.1 microdeletion syndrome
261112	9p deletion syndrome
261112	9p- syndrome
324313	9p13 microdeletion syndrome
96147	9q subtelomeric deletion syndrome
352665	9q21 microdeletion syndrome
96147	9qSTDS
284169	10p11.21p12.31 microdeletion syndrome
284169	10p12p11 microdeletion syndrome
276413	10q22.3q23.3 microdeletion syndrome
276422	10q22.3q23.3 microduplication syndrome
1307	10q24 microduplication syndrome
168588	11-beta-hydroxysteroid dehydrogenase deficiency type 1
320	11-beta-hydroxysteroid dehydrogenase deficiency type 2
52022	11p11.2 deletion

ORPHA Number	Disease name
300305	11p15.4 microduplication syndrome
313884	12p12.1 microdeletion syndrome
280325	12p13.33 microdeletion syndrome
94063	12q14 microdeletion syndrome
289513	12q15q21.1 microdeletion syndrome
1590	13q32 deletion
261120	14q11.2 microdeletion syndrome
261229	14q11.2 microduplication syndrome
261144	14q12 microdeletion syndrome
264200	14q22q23 microdeletion syndrome
264200	14q22-q23 microdeletion syndrome
314585	15q overgrowth syndrome
261183	15q11.2 microdeletion syndrome
238446	15q11q13 duplication syndrome
238446	15q11-q13 duplication syndrome
238446	15q11q13 microduplication syndrome
238446	15q11-q13 microduplication syndrome
199318	15q13.3 microdeletion syndrome
261190	15q14 microdeletion syndrome
94065	15q24 microdeletion syndrome
1596	15q26 deletion
261211	16p11.2p12.2 microdeletion syndrome
261211	16p11.2-p12.2 microdeletion syndrome
261204	16p11.2p12.2 microduplication syndrome
261236	16p13.11 microdeletion syndrome
261243	16p13.11 microduplication syndrome
96078	16p13.3 microduplication syndrome
352629	16q24.1 microdeletion syndrome
261250	16q24.3 microdeletion syndrome
752	17-beta-hydroxysteroid dehydrogenase 3 deficiency
35123	17b-hydroxysteroid dehydrogenase deficiency type 10
752	17-ketoreductase deficiency
752	17-ketosteroidreductase deficiency
819	17p11.2 microdeletion
1713	17p11.2 microduplication syndrome
217385	17p13.3 duplication syndrome
217385	17p13.3 microduplication syndrome
97685	17q11 microdeletion syndrome
139474	17q11.2 microduplication syndrome
261265	17q12 microdeletion syndrome
261272	17q12 microduplication syndrome
96169	17q21.31 microdeletion syndrome
217340	17q21.31 microduplication syndrome
261279	17q23.1q23.2 microdeletion syndrome
261279	17q23.1-q23.2 microdeletion syndrome
99763	18-hydroxylase deficiency
99763	18-oxidase deficiency
1598	18p- syndrome
1600	18q- syndrome

ORPHA Number	Disease name
254346	19p13.12 microdeletion syndrome
357001	19p13.13 microdeletion syndrome
217346	19q13.11 microdeletion syndrome
313781	20p subtelomeric deletion syndrome
261295	20p12.3 microdeletion syndrome
313781	20p13 microdeletion syndrome
261311	20q13.33 microdeletion syndrome
574	21q deletion
574	21q- syndrome
261323	21q22.11q22.12 microdeletion syndrome
261323	21q22.11-q22.12 microdeletion syndrome
268261	21q22.13q22.2 microdeletion syndrome
268261	21q22.13-q22.2 microdeletion syndrome
567	22q11.2 deletion syndrome
1727	22q11.2 microduplication syndrome
567	22q11DS
48652	22q13 deletion
881	45,X syndrome
881	45,X/46,XX syndrome
1772	45,X/46,XY MGD
1772	45,X/46,XY mixed gonadal dysgenesis
1772	45,X0/46,XY MGD
1772	45,X0/46,XY mixed gonadal dysgenesis
243	46,XX complete gonadal dysgenesis
2973	46,XX disorder of sex development - anorectal anomalies
2975	46,XX disorder of sex development - skeletal anomalies
243	46,XX gonadal dysgenesis
243	46,XX ovarian dysgenesis
2138	46,XX ovotesticular disorder of sex development
2138	46,XX ovotesticular DSD
243	46,XX pure gonadal dysgenesis
393	46,XX testicular disorder of sex development
393	46,XX testicular DSD
199310	46,XX/46,XY chimerism
242	46,XY CGD
242	46,XY complete gonadal dysgenesis
96266	46,XY disorder of sex developement due to partial LH receptor inactivation
96266	46,XY disorder of sex developement due to partial LH resistance
96266	46,XY disorder of sex developement due to partial luteinizing hormone resistance
168558	46,XY disorder of sex development - adrenal insufficiency due to CYP11A1 deficiency
752	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
753	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency

ORPHA Number	Disease name
96265	46,XY disorder of sex development due to complete LH receptor inactivation
96265	46,XY disorder of sex development due to complete LH resistance
96265	46,XY disorder of sex development due to complete luteinizing hormone receptor inactivation
96265	46,XY disorder of sex development due to complete luteinizing hormone resistance
90796	46,XY disorder of sex development due to isolated 17, 20 lyase deficiency
755	46,XY disorder of sex development due to LH resistance or LHB deficiency
325448	46,XY disorder of sex development due to LHB deficiency
755	46,XY disorder of sex development due to luteinizing hormone resistance or luteinizing hormone beta subunit deficiency
325448	46,XY disorder of sex development due to luteinizing hormone subunit beta deficiency
96265	46,XY DSD due to complete LH receptor inactivation
96265	46,XY DSD due to complete LH resistance
96265	46,XY DSD due to complete luteinizing hormone receptor inactivation
96265	46,XY DSD due to complete luteinizing hormone resistance
755	46,XY DSD due to LH resistance or LHB deficiency
325448	46,XY DSD due to LHB deficiency
755	46,XY DSD due to luteinizing hormone resistance or luteinizing hormone beta subunit deficiency
325448	46,XY DSD due to luteinizing hormone subunit beta deficiency
96266	46,XY DSD due to partial LH receptor inactivation
96266	46,XY DSD due to partial LH resistance
96266	46,XY DSD due to partial luteinizing hormone resistance
168563	46,XY gonadal dysgenesis - motor and sensory neuropathy
325345	46,XY ovotesticular disorder of sex development
325345	46,XY ovotesticular DSD
251510	46,XY partial gonadal dysgenesis
251510	46,XY partial testicular dysgenesis
251510	46,XY PGD
242	46,XY pure gonadal dysgenesis
3375	47,XXX
8	47,XYY syndrome
9	48,XXXX
96263	48,XXXY syndrome
10	48,XXYY
10	48,XXYY syndrome
99329	48,XYYY syndrome

ORPHA Number	Disease name
11	49,XXXXX
96264	49,XXXXY syndrome
261534	49,XXXYY syndrome
99330	49,XYYYY syndrome
869	4A syndrome
869	AAA syndrome
35708	AADC deficiency
91385	AAE
100055	AAE 2
100055	AAE II
1414	Aagenaes syndrome
284460	AAOR
915	Aarskog syndrome
1974	Aarskog-like syndrome
3163	Aarskog-Ose-Pande syndrome
915	Aarskog-Scott syndrome
124	Aase syndrome
916	Aase-Smith I syndrome
124	Aase-Smith II syndrome
916	Aase-Smith syndrome
69663	ABCB4 gene mutation-associated cholelithiasis
800	Aberfeld syndrome
14	Abetalipoproteinemia
920	Ablepharon macrostomia syndrome
99089	Abnormal number of coronary ostia
1138	Abnormal origin of the pulmonary artery
95493	Abnormal origin or aberrant course of coronary artery
1164	ABPA
921	Abruzzo-Erickson syndrome
69739	ABSD
2310	Absence deformity of leg - cataract
99112	Absence of brachiocephalic vein
99112	Absence of innominate vein
1658	Absence of dermatoglyphics - congenital milia
289465	Absence of fingerprints
1658	Absence of fingerprints - congenital milia
101206	Absence of pulmonary valve - Fallot's tetralogy - absence of ductus arteriosus
99048	Absence of pulmonary valve - ventricular septal defect - persistent ductus arteriosus
980	Absence of the pulmonary artery
99114	Absence of the superior caval vein
99114	Absence of the superior vena cava
99114	Absence of the SVC
93322	Absence of tibia
2879	Absence of ulna and fibula
96269	Absence of vagina
294986	Absent foot
295107	Absent foot, bilateral
295105	Absent foot, unilateral
294983	Absent hand
295103	Absent hand, bilateral
295101	Absent hand, unilateral
85201	Absent patellae - scrotal hypoplasia - renal anomalies - facial dysmorphism - intellectual deficit
2951	Absent thumb - short stature - immunodeficiency
988	Absent tibia - polydactyly
3328	Absent tibia - polydactyly - arachnoid cyst
945	Acalvaria
67043	Acanthamoeba keratitis
79468	Acanthokeratolytic verrucous nevus
300504	Acanthoma of the nail matrix
924	Acanthosis nigricans
90301	Acanthosis nigricans - Insulin resistance - muscle cramps - acral enlargement
926	Acatalasemia
561	Accelerated skeletal maturation - peculiar facies - failure to thrive
180182	Accessory breasts
99061	Accessory mitral valve tissue
674	Accessory pancreas
95462	Accessory tricuspid valve tissue
1114	ACCV
48818	Aceruloplasminemia
99736	Acetazolamide-responsive congenital myotonia
99736	Acetazolamide-responsive myotonia
2008	ACFS
869	Achalasia - addisonianism - alacrima syndrome
929	Achalasia - microcephaly
294983	Acheiria
295103	Acheiria, bilateral
295101	Acheiria, unilateral
931	Acheiropodia
931	Acheiropody
49382	ACHM
932	Achondrogenesis
93299	Achondrogenesis type 1A
93298	Achondrogenesis type 1B
93296	Achondrogenesis type 2
93299	Achondrogenesis, Houston-Harris type
93296	Achondrogenesis, Langer-Saldino type
93298	Achondrogenesis, Parenti-Fraccaro type
15	Achondroplasia
935	Achondroplasia - SCID
935	Achondroplasia - severe combined immunodeficiency
935	Achondroplasia - Swiss-type agammaglobulinemia
49382	Achromatopsia
355	Acid beta-glucosidase deficiency
35121	Acid phosphatase deficiency
40366	Acitretin embryofetopathy
79099	Ackerman dermatitis syndrome
2561	Ackerman syndrome
43115	Aconitase deficiency
252175	Acoustic neurilemoma
252175	Acoustic neurinoma
252175	Acoustic neuroma
65759	ACPS 2
65798	ACPS 4
3128	ACPS III
3128	ACPS with leg hypoplasia
306431	Acquired adult-onset immunodeficiency
90065	Acquired aneurysmal subarachnoid hemorrhage
91385	Acquired angioedema
100056	Acquired angioedema type 1
100055	Acquired angioedema type 2
91385	Acquired angioneurotic edema
100056	Acquired angioneurotic edema type 1
100055	Acquired angioneurotic edema type 2
91385	Acquired bradykinine-induced angioedema
91385	Acquired C1 inhibitor deficiency
95626	Acquired CDI
95626	Acquired central diabetes insipidus
91365	Acquired ciliary dyskinesia
228285	Acquired cutis laxa
46487	Acquired epidermolysis bullosa
79086	Acquired generalized lipodystrophy
228247	Acquired Gronblad-Strandberg-Touraine syndrome
231401	Acquired HbH disease
231401	Acquired hemoglobin H disease
73274	Acquired hemophilia
2221	Acquired hypertrichosis lanuginosa
26348	Acquired hypoprothrombinemia
454	Acquired ichthyosis
75564	Acquired idiopathic sideroblastic anemia
37559	Acquired kinky hair syndrome
79086	Acquired lipoatrophic diabetes
206616	Acquired metabolic neuropathy
589	Acquired myasthenia
95626	Acquired neurogenic diabetes insipidus
84142	Acquired neuromyotonia
91385	Acquired non histamine-induced angioedema
314697	Acquired porencephaly

ORPHA Number	Disease name
729	Acquired primary erythocytosis
26348	Acquired prothrombin deficiency
228247	Acquired pseudoxanthoma elasticum
228247	Acquired PXE
206575	Acquired rippling muscle disease
238547	Acquired secondary erythrocytosis
238547	Acquired secondary polycythemia
93585	Acquired thrombotic thrombocytopenic purpura
93585	Acquired thrombotic thrombocytopenic purpura due to anti-ADAMTS 13 antibodies
99147	Acquired Von Willebrand disease
99147	Acquired Von Willebrand syndrome
97360	Acral dysostosis with facial and genital abnormalities
158673	Acral dystrophic epidermolysis bullosa
90396	Acral persistent papular mucinosis
281127	Acral self-healing collodion baby
281127	Acral SHCB
945	Acrania
36	Acrocallosal syndrome
63446	Acrocapitofemoral dysplasia
2008	Acro-cardio-facial syndrome
221054	Acrocephalopolydactylous dysplasia
221054	Acrocephalopolydactyly
65759	Acrocephalopolysyndactyly type 2
3128	Acrocephalopolysyndactyly type 3
65798	Acrocephalopolysyndactyly type 4
87	Acrocephalosyndactyly type 1
794	Acrocephalosyndactyly type 3
710	Acrocephalosyndactyly type 5
1526	Acro-cephalo-synostosis
949	Acrocraniofacial dysostosis
955	Acro-dento-osteo-dysplasia
163931	Acrodermatitis continua suppurativa of Hallopeau
37	Acrodermatitis enteropathica, zinc deficiency type
978	Acro-dermato-ungual-lacrimal-tooth syndrome
950	Acrodysostosis
280651	Acrodysostosis with multiple hormone resistance
950	Acrodysplasia
2956	Acrodysplasia scoliosis
1786	Acrofacial dysostosis, Catania type
246	Acrofacial dysostosis, Genee-Wiedmann type
64542	Acrofacial dysostosis, Kennedy-Teebi type
1787	Acrofacial dysostosis, Palagonia type
1788	Acrofacial dysostosis, Rodriguez type
952	Acrofacial dysostosis, Weyers type
1784	Acro-fronto-facio-nasal dysostosis
2211	Acro-fronto-facio-nasal dysostosis type 2
2211	Acro-fronto-facio-nasal syndrome type 2
2500	Acrogeria
2500	Acrogeria, Gottron type
38	Acrokeratoelastoidosis of Costa
166113	Acrokeratosis of Bazex
166113	Acrokeratosis paraneoplastica
79151	Acrokeratosis verruciformis of Hopf
965	Acromegaloid facial appearance syndrome
963	Acromegaly
39	Acromelanosis
1827	Acromelic frontonasal dysplasia
968	Acromesomelic dwarfism
953	Acromesomelic dysplasia, Brahimi-Bacha type
2098	Acromesomelic dysplasia, Grebe type
968	Acromesomelic dysplasia, Hunter-Thomson type
40	Acromesomelic dysplasia, Maroteaux type
2500	Acrometageria
969	Acromicric dysplasia
955	Acroosteolysis, dominant type
2980	Acro-oto-ocular syndrome
85203	Acro-pectoral syndrome
956	Acro-pectoro-renal dysplasia
957	Acropectorovertebral dysplasia
41	Acropigmentation of Dohi
1133	Acrorenal defect - ectodermal dysplasia - diabetes
971	Acrorenal syndrome
958	Acro-renal-mandibular syndrome
959	Acro-renal-ocular syndrome
85203	ACRP syndrome
36	ACS
87	ACS 1
794	ACS 3
710	ACS 5
361	ACTH resistance
189427	ACTH-independent macronodular adrenal hyperplasia
98904	Actin myopathy
254395	Actinic lichen planus
254395	Actinic LP
330061	Actinic prurigo
163696	Action myoclonus - renal failure syndrome
101089	Activation-induced cytidine deaminase deficiency
73423	Acute ackee fruit intoxication
95409	Acute adrenal failure
95409	Acute adrenal insufficiency
95409	Acute adrenocortical insufficiency
73423	Acute akee fruit intoxication
99870	Acute and disseminated Langerhans cell histiocytosis
284460	Acute annular outer retinopathy
86849	Acute basophilic leukemia
69736	Acute bilateral depigmentation of the iris
98837	Acute biphenotypic leukemia
2901	Acute brachial plexus neuritis
83597	Acute disseminated encephalitis
83597	Acute disseminated encephalomyelitis
163703	Acute encephalitis with refractory repetitive partial seizures
318	Acute erythroid leukemia
243367	Acute fatty liver of pregnancy
3243	Acute febrile neutrophilic dermatosis
293173	Acute generalized exanthematous pustulosis
99920	Acute graft versus host disease
90062	Acute hepatic failure
95157	Acute hepatic porphyria
98916	Acute idiopathic demyelinating polyneuropathy
217371	Acute infantile liver failure due to synthesis defect of mitochondrial DNA-encoded proteins
217371	Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins
98916	Acute inflammatory demyelinating polyradiculoneuropathy
98916	Acute inflammatory polyneuropathy
79276	Acute intermittent porphyria
79126	Acute interstitial pneumonia
79126	Acute interstitial pneumonitis
73423	Acute intoxication by Blighia sapida
86851	Acute leukemia of ambiguous lineage
86851	Acute leukemia of indeterminate lineage
90062	Acute liver failure
178320	Acute lung injury
513	Acute lymphoblastic leukemia
513	Acute lymphoblastic leukemia/lymphoma
513	Acute lymphocytic leukemia
518	Acute megakaryoblastic leukemia
99887	Acute megakaryoblastic leukemia in Down syndrome
329469	Acute megakaryoblastic leukemia without Down syndrome
514	Acute monoblastic leukemia
514	Acute monocytic leukemia
98918	Acute motor axonal neuropathy
98917	Acute motor-sensory axonal GBS
98917	Acute motor-sensory axonal Guillain-Barré syndrome
98917	Acute motor-sensory axonal neuropathy

ORPHA Number	Disease name
228157	Acute multiple sclerosis, Marburg type
228157	Acute multiple sclerosis, Marburg variant
98833	Acute myeloblastic leukemia type 1
98834	Acute myeloblastic leukemia type 2
520	Acute myeloblastic leukemia type 3
514	Acute myeloblastic leukemia type 5
318	Acute myeloblastic leukemia type 6
518	Acute myeloblastic leukemia type 7
98834	Acute myeloblastic leukemia with maturation
98833	Acute myeloblastic leukemia without maturation
86843	Acute myelodysplasia with myelofibrosis
86843	Acute myelofibrosis
519	Acute myelogenous leukemia
519	Acute myeloid leukemia
102379	Acute myeloid leukemia and myelodysplastic syndromes related to alkylating agent
164726	Acute myeloid leukemia and myelodysplastic syndromes related to radiation
102381	Acute myeloid leukemia and myelodysplastic syndromes related to topoisomerase type 2 inhibitor
98831	Acute myeloid leukemia with 11q23 abnormalities
98829	Acute myeloid leukemia with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22)
319480	Acute myeloid leukemia with CEBPA somatic mutations
86845	Acute myeloid leukemia with multilineage dysplasia
520	Acute myeloid leukemia with t(15;17)(q22;q12);(PML/RARalpha) and variants
102724	Acute myeloid leukemia with t(8;21)(q22;q22) translocation
517	Acute myelomonocytic leukemia
86843	Acute myelosclerosis
263524	Acute necrotizing encephalopathy of childhood
247546	Acute neonatal citrullinemia type 1
247546	Acute neonatal citrullinemia type I
77260	Acute neuronopathic Gaucher disease
163703	Acute non-herpetic encephalitis with severe refractory status epilepticus
519	Acute non-lymphoblastic leukemia
35889	Acute opioid poisoning
231457	Acute panautonomic GBS
231457	Acute panautonomic Guillain-Barré syndrome
231457	Acute panautonomic neuropathy
231457	Acute pandysautonomia
86843	Acute panmyelosis with myelofibrosis
90064	Acute peripheral arterial occlusion
43119	Acute poisoning by drugs with membrane-stabilizing effect
520	Acute promyelocytic leukemia
98918	Acute pure motor GBS
98918	Acute pure motor Guillain-Barré syndrome
231450	Acute pure sensory GBS
231450	Acute pure sensory Guillain-Barré syndrome
231450	Acute pure sensory neuropathy
3099	Acute rheumatic fever
90059	Acute sensorineural hearing loss by acute acoustic trauma or sudden deafness or surgery induced acoustic trauma
231466	Acute sensory ataxic GBS
231466	Acute sensory ataxic Guillain-Barré syndrome
231466	Acute sensory ataxic neuropathy
139417	Acute transverse myelitis
43117	Acute tricyclic antidepressant poisoning
91500	Acute tubulointerstitial nephritis and uveitis syndrome
98835	Acute undifferentiated leukemia
284454	Acute zonal occult outer retinopathy
137754	ACY1D
141	ACY2 deficiency
99901	Acyl-CoA dehydrogenase 9 deficiency
99736	ACZ-responsive congenital myotonia
99736	ACZ-responsive myotonia
93608	AD dRTA
428	AD hypocalcemia
314889	AD pRTA
277	ADA deficiency
973	Adactylia of hand, unilateral
295118	Adactyly of foot, bilateral
295116	Adactyly of foot, unilateral
216796	Adair-Dighton syndrome
1034	ADAM syndrome
55881	Adamantinoma
974	Adams-Oliver syndrome
88619	ADANE
94145	ADCA1
94148	ADCA3
94149	ADCA4
314404	ADCA-DN
94145	ADCAI
94148	ADCAIII
94149	ADCAIV
90348	ADCL
86814	ADCME
169189	AD-CNM
85138	Addison disease
2953	Adducted thumb - clubfoot syndrome
2952	Adducted thumbs - arthrogryposis, Christian type
2953	Adducted thumbs - arthrogryposis, Dundar type
101046	ADEAF
83597	ADEM
976	Adenine phosphoribosyltransferase deficiency
213772	Adenocarcinoma of the cervix uteri
95512	Adenohypophysitis
213828	Adenoid basal carcinoma of the cervix uteri
213823	Adenoid cystic carcinoma of the cervix uteri
213741	Adenoid cystic carcinoma of the corpus uteri
26790	Adenomucinosis
213792	Adenosarcoma of the cervix uteri
213600	Adenosarcoma of the corpus uteri
45	Adenosine monophosphate deaminase deficiency
28	Adenosylcobalamin deficiency
91127	Adenovirus infection in immunocompromised patients
46	Adenylosuccinase deficiency
46	Adenylosuccinate lyase deficiency
1810	AD-HED
137817	Adhesive arachnoiditis
2314	AD-HIES
89937	ADHR
36397	Adiposalgia
36397	Adipose tissue rheumatism
36397	Adiposis dolorosa
289290	ADK hypermethioninemia
101046	ADLTE
178464	ADMERF
98784	ADNFLE
329211	ADNIV
1544	Adolescent benign focal crisis
3153	Adolescent idiopathic scoliosis
306588	ADOS
36355	ADP platelet receptor P2Y12 deficiency
2924	ADPCLD
101046	ADPEAF
254892	adPEO
95409	Adrenal crisis
463	Adrenal incidentaloma
869	Adrenal insufficiency - achalasia - alacrima
1501	Adrenocortical carcinoma
231625	Adrenocortical carcinoma with pure aldosterone hypersecretion

ORPHA Number	Disease name
99889	Adrenocorticotropic hormone secretion syndrome
189427	Adrenocorticotropic hormone-independent macronodular adrenal hyperplasia
139399	Adrenomyeloneuropathy
977	Adrenomyodystrophy
228169	ADSD
46	ADSL deficiency
2688	Adult idiopathic neutropenia
70578	Adult acute respiratory distress syndrome
70578	Adult ARDS
93605	Adult Bartter syndrome
157846	Adult basal ganglia disease
93668	Adult chronic recurrent multifocal osteomyelitis
93668	Adult CRMO
2666	Adult familial nephronophtisis - spastic quadriparesia
309169	Adult GM2 gangliosidosis 0 variant
874	Adult heart tumor
210159	Adult hepatocellular carcinoma
247676	Adult hypophosphatasia
178487	Adult intestinal botulism
178487	Adult intestinal colonization botulism
178487	Adult intestinal toxemia botulism
178487	Adult intestinal toxin-mediated botulism
206448	Adult Krabbe disease
79262	Adult NCL
79262	Adult neuronal ceroid lipofuscinosis
247676	Adult phosphoethanolaminuria
206583	Adult polyglucosan body disease
902	Adult progeria
99874	Adult pulmonary Langerhans cell histiocytosis
98872	Adult pure red cell aplasia
247676	Adult Rathburn disease
829	Adult Still's disease
978	ADULT syndrome
86875	Adult T-cell leukemia/lymphoma
79280	Adult-onset Alpha-N-acetylgalactosaminidase deficiency
99027	Adult-onset autosomal dominant leukodystrophy
284289	Adult-onset autosomal recessive cerebellar ataxia
329336	Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy
247585	Adult-onset citrin deficiency
247573	Adult-onset citrullinemia type 1
247585	Adult-onset citrullinemia type 2
247573	Adult-onset citrullinemia type I
329336	Adult-onset CPEO with mitochondrial myopathy
221	Adult-onset dermatomyositis
329478	Adult-onset distal myopathy due to VCP mutation
199351	Adult-onset dystonia-parkinsonism
93963	Adult-onset focal torsion dystonia
99000	Adult-onset foveomacular vitelliform dystrophy
79257	Adult-onset GM1 gangliosidosis
93963	Adult-onset idiopathic torsion dystonia
306431	Adult-onset immunodeficiency with anti-interferon-gamma autoantibodies
313808	Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia
329314	Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency
329314	Adult-onset multiple mtDNA deletion syndrome due to DGUOK deficiency
171442	Adult-onset nemaline myopathy
276608	Adult-onset non-insulinoma persistent hyperinsulinemic hypoglycemia
206572	Adult-onset overlap myositis
35689	Adult-onset PLS
732	Adult-onset polymyositis
35689	Adult-onset primary lateral sclerosis
209335	Adult-onset proximal spinal muscular atrophy, autosomal dominant
324982	Adult-onset SAPHO syndrome
324982	Adult-onset synovitis-acne-pustulosis-hyperostosis-osteitis syndrome
3086	ADVIRC
682	Adynamia episodica hereditaria
1071	AEC syndrome
281139	AEI
163703	AERRPS
220460	AFAP
313772	AFG3L2-associated spastic ataxia-neuropathy syndrome
243367	AFLP
139507	African iron overload
101334	African tick typhus
3385	African trypanosomiasis
83617	Agammaglobulinemia - microcephaly - craniosynostosis - severe dermatitis
33110	Agammaglobulinemia, non-Bruton type
388	Aganglionic megacolon
180142	Agenesis and aplasia of uterine body
52055	Agenesis of the corpus callosum - intellectual deficit - coloboma - micrognathia
99114	Agenesis of the superior caval vein
99114	Agenesis of the superior vena cava
99114	Agenesis of the SVC
293173	AGEP
873	Aggressive fibromatosis
86873	Aggressive NK-cell leukemia
86873	Aggressive NK-cell lymphoma
98850	Aggressive systemic mastocytosis
989	Aglossia - adactylia
990	Agnathia - holoprosencephaly - situs inversus
824	Agnogenic myeloid metaplasia
100070	Agramatic variant of PPA
100070	Agramatic variant of primary progressive aphasia
2131	AHC
59	AHDS
50812	Ahn-Lerman-Sagie syndrome
79443	AHO - PHP Ia
79445	AHO - PPHP
2134	aHUS
93581	aHUS with anti-factor H antibodies
93578	aHUS with B factor anomaly
93575	aHUS with C3 anomaly
357008	aHUS with DGKE deficiency
93579	aHUS with H factor anomaly
93580	aHUS with I factor anomaly
93576	aHUS with MCP/CD46 anomaly
217023	aHUS with thrombomodulin anomaly
50	Aicardi syndrome
51	Aicardi-Goutières syndrome
250977	AICA-ribosiduria
101089	AID deficiency
98916	AIDP
90081	AIDS wasting syndrome
178333	AIED
86886	AILT
189427	AIMAH
103919	AIP
280302	AIP type 1
280315	AIP type 2
754	AIS
75564	AISA
33355	AK2 deficiency
1417	Akaba-Hayasaka syndrome
38	AKE
85443	AL amyloidosis
2232	Al Awadi-Farag-Teebi syndrome
2879	Al Awadi-Raas-Rothschild syndrome
2725	Al Gazali-Al Talabani syndrome
2865	Al Gazali-Aziz-Salem syndrome
2153	Al Gazali-Donnai-Muller syndrome
2725	Al Gazali-Lytle syndrome
2773	Al Gazali-Nair syndrome
100924	ALAD porphyria
52	Alagille syndrome

ORPHA Number	Disease name
261600	Alagille syndrome due to 20p12 microdeletion
261619	Alagille syndrome due to a JAG1 point mutation
261629	Alagille syndrome due to a NOTCH2 point mutation
261600	Alagille syndrome due to del(20)(p12)
261600	Alagille syndrome due to monosomy 20p12
52	Alagille-Watson syndrome
261619	Alagille-Watson syndrome due to a JAG1 point mutation
261629	Alagille-Watson syndrome due to a NOTCH2 point mutation
261600	Alagille-Watson syndrome due to monosomy 20p12
178333	Åland Islands eye disease
2007	Alar cartilages hypoplasia - coloboma - telecanthus
53	Albers-Schönberg osteopetrosis
998	Albinism-deafness syndrome
665	Albright hereditary osteodystrophy
79443	Albright hereditary osteodystrophy - PHP Ia
79445	Albright hereditary osteodystrophy - PPHP
1001	Albright hereditary osteodystrophy 3
1001	Albright hereditary osteodystrophy-like syndrome
98841	ALCL
1915	Alcohol antenatal infection
36899	Alcohol-responsive dystonia
43	ALD
35664	ALDH18A1-related DeBarsy syndrome
99763	Aldosterone synthase deficiency
99764	Aldosterone synthase deficiency unlinked to CYP11B2
99764	Aldosterone synthase deficiency unlinked to the aldosterone synthase gene
85332	Aldred syndrome
158799	Aleukemic mast cell leukemia
58	Alexander disease
261112	Alfi syndrome
79327	ALG1-CDG syndrome
79326	ALG2-CDG syndrome
79321	ALG3-CDG syndrome
79320	ALG6-CDG syndrome
79325	ALG8-CDG syndrome
79328	ALG9-CDG syndrome
280071	ALG11-CDG syndrome
79324	ALG12-CDG syndrome
324422	ALG13-CDG syndrome
99995	Algodystrophy
300903	ALK- ALCL
300903	ALK- anaplastic large cell lymphoma
300895	ALK+ ALCL
300895	ALK+ anaplastic large cell lymphoma
56	Alkaptonuria
300903	ALK-negative anaplastic large cell lymphoma
300895	ALK-positive anaplastic large cell lymphoma
513	ALL
1526	Allain-Babin-Demarquez syndrome
59	Allan-Herndon-Dudley syndrome
1164	Allergic aspergillosis
1164	Allergic bronchopulmonary aspergillosis
869	Allgrove syndrome
93925	Alobar holoprosencephaly
2316	Alopecia - anosmia - deafness - hypogonadism
1005	Alopecia - contractures - dwarfism - intellectual deficit
202	Alopecia - deafness - hypogonadism
1008	Alopecia - epilepsy - pyorrhea - intellectual deficit
1008	Alopecia - epilepsy - pyorrhea - mental subnormality
1014	Alopecia - intellectual deficit - hypergonadotropic hypogonadism
157954	Alopecia - progressive neurological defect - endocrinopathy
1006	Alopecia antibody deficiency
700	Alopecia totalis
701	Alopecia universalis
2574	Alopecia-epilepsy-oligophrenia syndrome, Moynahan type
2850	Alopecia-intellectual deficit syndrome
726	Alpers progressive sclerosing poliodystrophy
726	Alpers syndrome
726	Alpers-Huttenlocher syndrome
734	Alpha delta granule deficiency
734	Alpha dense granule deficiency
100025	Alpha heavy-chain disease
721	Alpha storage pool deficiency
98791	Alpha thalassemia - intellectual deficit syndrome linked to chromosome 16
98791	Alpha thalassemia - intellectual deficit syndrome, deletion type
98791	Alpha thalassemia - retardation syndrome
847	Alpha thalassemia - retardation syndrome, X-linked
847	Alpha thalassemia - X-linked intellectual deficit syndrome
365	Alpha-1,4-glucosidase acid deficiency
308604	Alpha-1,4-glucosidase acid deficiency, adult onset
308552	Alpha-1,4-glucosidase acid deficiency, infantile onset
308573	Alpha-1,4-glucosidase acid deficiency, juvenile onset
93594	Alpha-1-antichymotrypsin deficiency
60	Alpha-1-antitrypsin deficiency
79154	Alpha-aminoadipic aciduria
280553	Alpha-B crystallin-related myofibrillar myopathy
98910	Alpha-crystallinopathy
324	Alpha-galactosidase A deficiency
100025	Alpha-HCD
31	Alpha-ketoglutarate dehydrogenase deficiency
349	Alpha-L-fucosidase deficiency
579	Alpha-L-iduronidase deficiency
61	Alpha-mannosidosis
309288	Alpha-mannosidosis, adult form
309282	Alpha-mannosidosis, infantile form
134	Alpha-methyl-acetoacetyl-CoA thiolase deficiency
79095	Alpha-methyl-acyl-CoA racemase deficiency
3137	Alpha-N-acetylgalactosaminidase deficiency
79279	Alpha-N-acetylgalactosaminidase deficiency type 1
79280	Alpha-N-acetylgalactosaminidase deficiency type 2
79281	Alpha-N-acetylgalactosaminidase deficiency type 3
62	Alpha-sarcoglycanopathy
846	Alpha-thalassemia
231401	Alpha-thalassemia - myelodysplastic syndrome
163596	Alpha-thalassemia hydrops fetalis
93616	Alpha-thalassemia intermedia
163596	Alpha-thalassemia major
63	Alport deafness-nephropathy
63	Alport syndrome
86818	Alport syndrome - intellectual deficit - midface hypoplasia - elliptocytosis
3261	ALPS
268114	ALPS type 4
268114	ALPS type IV
275517	ALPS with recurrent infections
803	ALS
357043	ALS4
86815	ALSG
313808	ALSP
64	Alström syndrome
99971	ALT
2131	Alternating hemiplegia in childhood
2131	Alternating hemiplegia of childhood
210122	Alveolar capillary dysplasia with misalignment of pulmonary veins
210122	Alveolar capillary dysplasia with misalignment of pulmonary vessels
284	Alveolar echinococcosis

ORPHA Number	Disease name
99756	Alveolar rhabdomyosarcoma
163699	Alveolar soft-part sarcoma
3354	Alves-dos Santos-Castelo syndrome
306542	ALX1-related frontonasal dysplasia
228390	ALX4-related FNDAG
79095	AMACR deficiency
98918	AMAN
1021	Amaurosis - hypertrichosis
65	Amaurosis congenita of Leber
1023	Ambras syndrome
294969	Amelia of lower limb
295059	Amelia of lower limb, bilateral
295057	Amelia of lower limb, unilateral
294967	Amelia of upper limb
295055	Amelia of upper limb, bilateral
295053	Amelia of upper limb, unilateral
314422	Ameloblastic carcinoma
314419	Ameloblastoma
1946	Amelo-cerebro-hypohidrotic syndrome
88661	Amelogenesis imperfecta
1031	Amelogenesis imperfecta - nephrocalcinosis
171836	Amelogenesis imperfecta and gingival hyperplasia syndrome
100031	Amelogenesis imperfecta type 1
100033	Amelogenesis imperfecta type 2
100032	Amelogenesis imperfecta type 3
100034	Amelogenesis imperfecta type 4
1028	Amelo-onycho-hypohidrotic syndrome
83595	American mountain fever
3386	American trypanosomiasis
2116	Aminoaciduria, Hartnup type
141	Aminoacylase 2 deficiency
1908	Aminopterin embryopathy syndrome
221120	Aminopterin syndrome-like sine aminopterin
1908	Aminopterin/methotrexate embryofetopathy
1245	Amish brittle hair syndrome
171714	Amish infantile epilepsy syndrome
99742	Amish lethal microcephaly
98902	Amish nemaline myopathy
518	AMKL
519	AML
319480	AML with CEBPA somatic mutations
514	AML-M5
318	AML-M6
86818	AMME complex
86818	AMME syndrome
1034	Amniotic bands
1034	Amniotic deformity - adhesion - mutilation syndrome

ORPHA Number	Disease name
251663	aMOA
67	Amoebiasis due to Entamoeba histolytica
68	Amoebiasis due to free-living amoebae
45	AMP deaminase deficiency
1035	Ampola syndrome
66529	Ampulla cardiomyopathy
300557	Ampullary carcinoma
300557	Ampulloma
98917	AMSAN
366	Amylo-1,6-glucosidase deficiency
49804	Amyloid lichen
85445	Amyloidosis AA
319635	Amyloidosis cutis dyschromia
319635	Amyloidosis cutis dyschromica
85450	Amyloidosis, Ostertag type
367	Amylopectinosis
1037	Amyoplasia congenita
803	Amyotrophic lateral sclerosis
357043	Amyotrophic lateral sclerosis type 4
94091	Amyotrophic lateral sclerosis, hemiplegic type
90020	Amyotrophic lateral sclerosis-parkinsonism-dementia complex
90020	Amyotrophic lateral sclerosis-parkinsonism-dementia of Guam
2615	Amyotrophy - fat tissue anomaly
100082	Anal endocrine tumor
228113	Anal fistula
761	Anaphylactoid purpura
251589	Anaplastic astrocytoma
251646	Anaplastic ependymoma
251957	Anaplastic ganglioglioma
98841	Anaplastic large cell lymphoma
251663	Anaplastic oligoastrocytoma
251630	Anaplastic oligodendroglioma
142	Anaplastic thyroid carcinoma
251855	Anaplastic/large cell medulloblastoma
93347	Anauxetic dysplasia
79262	ANCL
78	Ancylostomiasis
1496	Andermann syndrome
37553	Andersen cardiodysrhythmic periodic paralysis
367	Andersen disease
37553	Andersen syndrome
37553	Andersen-Tawil syndrome
71	Anderson disease
324	Anderson-Fabry disease
99916	Androblastoma
754	Androgen insensitivity syndrome
754	Androgen resistance syndrome
329813	Androgenetic/biparental mosaicism

ORPHA Number	Disease name
157954	ANE syndrome
263524	ANEC
1044	Anemia due to adenosine triphosphatase deficiency
284984	Aneurysm - osteoarthritis syndrome
1054	Aneurysm of sinus of Valsalva
95484	Aneurysm or dilatation of ascending aorta
353344	Aneurysmal telangiectasia
72	Angelman syndrome
98794	Angelman syndrome due to maternal 15q11q13 deletion
98794	Angelman syndrome due to maternal monosomy 15q11q13
98795	Angelman syndrome due to paternal uniparental disomy of chromosome 15
63442	Angel-shaped phalango-epiphyseal dysplasia
251671	Angiocentric glioma
86879	Angiocentric T-cell lymphoma
79093	Angiodysgenetic necrotizing myelopathy
98839	Angioendotheliomatosis proliferans systemisata
160	Angiofollicular ganglionic hyperplasia
160	Angiofollicular lymph hyperplasia
86886	Angioimmunoblastic T-cell lymphoma
324	Angiokeratoma corporis diffusum
95429	Angioma serpiginosum
2346	Angio-osteohypertrophic syndrome
75508	Angio-osteohypotrophic syndrome
263413	Angiosarcoma
74	Angiostrongyliasis
98839	Angiotropic large cell lymphoma
76	Anguilluliasis
76	Anguillulosis
238468	Anhidrotic ectodermal dysplasia
69088	Anhidrotic ectodermal dysplasia - immunodeficiency - osteopetrosis - lymphedema
98813	Anhidrotic ectodermal dysplasia with immunodeficiency
77	Aniridia
1069	Aniridia - absent patella
1065	Aniridia - cerebellar ataxia - intellectual deficit
1067	Aniridia - ptosis - intellectual deficit - familial obesity
1064	Aniridia - renal agenesis - psychomotor retardation
1068	Aniridia-intellectual deficit syndrome
1070	Anisakiasis
86873	ANKCL
1071	Ankyloblepharon - ectodermal defects - cleft lip/palate
1074	Ankyloblepharon filiforme - imperforate anus

ORPHA Number	Disease name
1072	Ankyloblepharon filiforme adnatum - cleft palate
2206	Ankylosing vertebral hyperostosis with tylosis
1077	Ankylosis of teeth
78	Ankylostomiasis
254411	Annular atrophic lichen planus
254411	Annular atrophic LP
281139	Annular epidermolytic ichthyosis
254424	Annular lichen planus
254424	Annular LP
675	Annular pancreas
229	Annuloaortic ectasia
99797	Anodontia
101932	Anomaly of the mitral subvalvular apparatus
95463	Anomaly of the tricuspid subvalvular apparatus
99055	Anomaly of the tricuspid valve chordae
79143	Anonychia
1094	Anonychia - microcephaly
90390	Anonychia - onychodystrophy
1487	Anonychia - onychodystrophy with hypoplasia or absence of distal phalanges
94150	Anonychia congenita totalis
69125	Anonychia with flexural pigmentation
99987	Anophthalmia - esophageal-genital syndrome
1101	Anophthalmia - megalocornea - cardiopathy - skeletal anomalies
2470	Anophthalmia - pulmonary hypoplasia
1106	Anophthalmia - syndactyly
1104	Anophthalmia plus syndrome
77298	Anophthalmia/microphthalmia - esophageal atresia
1882	ANOTHER syndrome
93976	Anotia
2987	Antecubital pterygium syndrome
93604	Antenatal Bartter syndrome
70596	Antenatal Epstein-Barr virus infection
178148	Antenatal multiminicore disease with arthrogryposis multiplex congenita
1931	Anterior encephalocele
98961	Anterior limiting membrane dystrophy type I
98960	Anterior limiting membrane dystrophy type II
95512	Anterior pituitary hypophysitis
98988	Anterior polar cataract
98988	Anterior subcapsular cataract
90079	Anthracycline extravasations
36412	Anti-C1q vasculitis
375	Anti-glomerular basement membrane antibody-mediated disease
2194	Anti-HLA hyperimmunization
2821	Antinolo-Nieto-Borrego syndrome
206569	Anti-SRP myopathy
81	Antisynthetase syndrome
83	Antley-Bixler syndrome
1190	AO1
56305	AO3
1168	AOA1
64753	AOA2
1190	AOI*
70590	AOI*
56305	AOIII
1457	Aorta coarctation
2037	Aorta-pulmonary artery fistula
60030	Aortic aneurysm syndrome due to TGF-beta receptors anomalies
1110	Aortic arch anomaly - peculiar facies - intellectual deficit
1132	Aortic arch defects
2299	Aortic arch interruption
99079	Aortic arch syndrome
88636	Aortic dilatation - joint hypermobility - arterial tortuosity
95448	Aortic valve atresia
101043	Aortic valve dysplasia
99071	Aorto-left ventricular tunnel
99086	Aorto-pulmonary coronary arterial course
99070	Aorto-right ventricular tunnel
3400	Aorto-ventricular tunnel
974	AOS*
284984	AOS*
280763	AP4 deficiency syndrome
206583	APBD
247806	APC-related AFAP
247806	APC-related attenuated familial adenomatous polyposis
247806	APC-related attenuated familial polyposis coli
247806	APC-related attenuated FAP
3453	APECED syndrome
87	Apert syndrome
162521	Apertura pyriformis with holoprosencephaly
1112	Aphalangy - hemivertebrae - urogenital-intestinal dysgenesis
1113	Aphalangy - syndactyly - microcephaly
49	Aphallia
324540	Aphonia - deafness - retinal dystrophy - bifid halluces - intellectual deficit
324540	Aphonia - deafness - retinal dystrophy - duplicated halluces - intellectual deficit
66529	Apical ballooning syndrome
324530	aPLAID
1117	Aplasia cutis - myopia
3339	Aplasia cutis congenita - epibulbar dermoids
1116	Aplasia cutis congenita - intestinal lymphangiectasia
1114	Aplasia cutis congenita verticis
86815	Aplasia of lacrimal and salivary glands
3329	Aplasia of tibia with split-hand/split-foot deformity
2879	Aplasia/hypoplasia of limbs and pelvis
70590	Apnea of infancy
99981	Apnea of prematurity
294986	Apodia
295107	Apodia, bilateral
295105	Apodia, unilateral
93560	Apolipoprotein AI amyloidosis
425	Apolipoprotein A-I deficiency
238269	Apolipoprotein AII amyloidosis
320	Apparent mineralocorticoid excess
100079	Appendiceal endocrine tumor
1201	Apple peel syndrome
1126	Aprosencephaly cerebellar dysgenesis
976	APRT deficiency
3143	APS type 2
227982	APS type 3
227990	APS type 4
3453	APS1
3143	APS2
227982	APS3
227990	APS4
101206	APV/ADA, Fallot type
99048	APV/PDA, non-Fallot type
93611	AR dRTA with hearing loss
93609	AR dRTA without deafness
93609	AR dRTA without hearing loss
93611	AR dRTA wth deafness
93607	AR pRTA
1129	Arachnodactyly - abnormal ossification - intellectual deficit
1130	Arachnodactyly - intellectual deficit - dysmorphism
2356	Arachnoid cyst
137817	Arachnoiditis
324442	ARAN-NM
344	Arbovirus fever
2697	ARC syndrome
88644	ARCA1
139485	ARCA2
90349	ARCL1
90350	ARCL2
357074	ARCL2, classic type
357074	ARCL2, Debré type

*Caution: one same acronym may correspond to different diseases in medical terms. Please refer to the full name of the disease to get the correct Orpha code.

ORPHA Number	Disease name
357064	ARCL2, progeroid type
357058	ARCL2A
357064	ARCL2B
64749	AR-CMT1
91024	AR-CMT2
98856	AR-CMT2B1
101101	AR-CMT2B2
101102	AR-CMT2C
101097	ARCMT2K
324442	ARCMT2-NM
169186	AR-CNM
1133	AREDYLD syndrome
101096	Aregenerative anemia
75377	Areolar atrophy of the macula
319223	Argentine hemorrhagic fever
319223	Argentinian hemorrhagic fever
90	Arginase deficiency
35704	Arginine:glycine amidinotransferase deficiency
90	Argininemia
23	Argininosuccinase deficiency
247525	Argininosuccinate synthase deficiency
247525	Argininosuccinate synthetase deficiency
247525	Argininosuccinic acid synthase deficiency
247525	Argininosuccinic acid synthetase deficiency
23	Argininosuccinic aciduria
60014	Argyria
97342	Argyrophilic grain disease
248	AR-HED
169446	AR-HIES
331226	AR-HIES due to TYK2 deficiency
289176	ARHR
331226	AR-hyper IgE syndrome due to TYK2 deficiency
79235	Arias syndrome
2318	Arima syndrome
950	Arkless-Graham syndrome
85276	Armfield syndrome
167635	Arndt-Gottron disease
268882	Arnold-Chiari malformation type 1
1136	Arnold-Chiari malformation type 2
268882	Arnold-Chiari malformation type I
1136	Arnold-Chiari malformation type II
91	Aromatase deficiency
178345	Aromatase excess syndrome
35708	Aromatic L-amino acid decarboxylase deficiency
254886	arPEO
99916	Arrhenoblastoma
1134	Arrhinia
1135	Arrhinia - choanal atresia - microphthalmia
247	Arrhythmogenic right ventricular cardiomyopathy
247	Arrhythmogenic right ventricular dysplasia
98	ARSACS
314603	ARSAL
583	ARSB deficiency
357107	Arterial cervical rib syndrome
357107	Arterial costoclavicular syndrome
1682	Arterial dissection - lentiginosis
357107	Arterial hyperabduction syndrome
97599	Arterial hypertension due to renal artery stenosis secondary to vasculitis
357107	Arterial scalenus anticus syndrome
357107	Arterial thoracic outlet compression syndrome
357107	Arterial thoracic outlet syndrome
3342	Arterial tortuosity syndrome
357107	Arterial TOS
52	Arteriohepatic dysplasia
261619	Arteriohepatic dysplasia due to a JAG1 point mutation
261629	Arteriohepatic dysplasia due to a NOTCH2 point mutation
261600	Arteriohepatic dysplasia due to monosomy 20p12
29207	Arthritis urethritica
3200	Arthrogryposis - ectodermal dysplasia - other anomalies
1139	Arthrogryposis - epileptic seizures - migrational brain disorder
1485	Arthrogryposis - hyperkeratosis, lethal form
2697	Arthrogryposis - renal dysfunction - cholestasis
65720	Arthrogryposis - severe scoliosis
1155	Arthrogryposis due to muscular dystrophy
1037	Arthrogryposis multiplex congenita
994	Arthrogryposis multiplex congenita - pulmonary hypoplasia
1150	Arthrogryposis multiplex congenita - whistling face
1154	Arthrogryposis with oculomotor limitation and electroretinal anomalies
1144	Arthrogryposis-like hand anomaly - sensorineural deafness
1149	Arthrogryposis-like syndrome
2848	Arthropathy-camptodactyly syndrome
1187	Arts syndrome
247	ARVC
247	ARVD
512	Arylsulfatase A deficiency
309271	Arylsulfatase A deficiency, adult form
309263	Arylsulfatase A deficiency, juvenile form
309256	Arylsulfatase A deficiency, late infantile form
583	Arylsulfatase B deficiency
231466	ASAN
583	ASB deficiency
2302	Asbestos intoxication
2302	Asbestosis
1253	Ascher syndrome
1478	ASD
352490	ASD due to AUTS2 deficiency
99104	ASD, coronary sinus type
99106	ASD, ostium primum type
99103	ASD, ostium secundum type
99105	ASD, sinus venosus type
54251	Aseptic abscesses syndrome
97337	Aseptic necrosis of patella
3314	Aseptic necrosis of phalangeal epiphyses
2380	Aseptic necrosis of the capital femoral epiphysis
97336	Aseptic necrosis of the capital humerus
97332	Aseptic necrosis of the lunate bone
2054	Aseptic necrosis of the tarsal bone
97335	Aseptic necrosis of the tibial tubercle
57194	Aseptic osteitis
54251	Aseptic systemic abscesses
137686	Asherman syndrome
141	Aspartoacylase deficiency
93	Aspartylglucosaminidase deficiency
93	Aspartylglucosaminuria
63442	ASPED
1163	Aspergillosis
474	Asphyxiating thoracic dystrophy of the newborn
247525	ASS deficiency
221120	ASSA
85175	Astley-Kendall dysplasia
251679	Astroblastoma
94	Astrocytic tumor
94	Astrocytoma
647	AT V1
251891	AT/RT
137639	Ataxia - delayed dentition - hypomyelination
1227	Ataxia - diabetes - goiter - gonadal insufficiency
1180	Ataxia - hypogonadism - choroidal dystrophy
1168	Ataxia - oculomotor apraxia type 1
64753	Ataxia - oculomotor apraxia type 2
2585	Ataxia - pancytopenia
1184	Ataxia - photosensitivity - short stature
1178	Ataxia - tapetoretinal degeneration

ORPHA Number	Disease name
96	Ataxia with isolated vitamin E deficiency
96	Ataxia with vitamin E deficiency
1188	Ataxia-deafness-retardation syndrome
100	Ataxia-telangiectasia
647	Ataxia-telangiectasia, variant 1
251347	Ataxia-telangiectasia-like disorder
1183	Ataxo-opso-myoclonus syndrome
2953	ATCS
3469	Atelencephaly
1190	Atelosteogenesis type I
56304	Atelosteogenesis type II
56305	Atelosteogenesis type III
69739	Athabaskan brainstem dysgenesis syndrome
1192	Atherosclerosis- deafness - diabetes - epilepsy - nephropathy
95713	Athyreosis
250977	ATIC deficiency
1193	Atkin-Flaitz syndrome
99666	Atlantoaxial subluxation
251347	ATLD
86875	ATLL
139423	ATM/TM
231401	ATMDS
163934	Atopic keratoconjunctivitis
357107	ATOS
98791	ATR syndrome linked to chromosome 16
98791	ATR syndrome, deletion type
98791	ATR-16 syndrome
30391	Atresia of bile ducts
1201	Atresia of small intestine
105	Atresia of urethra
99141	Atrésie choanale postérieure - lymphoedème
1344	Atrial cardiomyopathy with heart block
99107	Atrial septal aneurysm
1478	Atrial septal defect
1479	Atrial septal defect - atrioventricular conduction defects
99104	Atrial septal defect, coronary sinus type
99106	Atrial septal defect, ostium primum type
99103	Atrial septal defect, ostium secundum type
99105	Atrial septal defect, sinus venosus type
1344	Atrial stand still
844	Atrial tachyarrhythmia with short PR interval
1211	Atrichia - mental and growth delay
86819	Atrichia with papular lesions
392	Atriodigital dysplasia type 1
1350	Atriodigital dysplasia type 2
1342	Atriodigital dysplasia type 3
168796	Atrio-digital dysplasia, Slovenian type
1352	Atrioventricular defect - blepharophimosis -radial defects
86813	Atrophia areata
649	Atrophia bulborum hereditaria
254449	Atrophic lichen planus
254449	Atrophic LP
79100	Atrophoderma vermiculata
71289	ATRUS syndrome
847	ATR-X syndrome
3342	ATS
86818	ATS-MR
352723	Attenuated Chédiak-Higashi syndrome
220460	Attenuated familial adenomatous polyposis
220460	Attenuated familial polyposis coli
220460	Attenuated FAP
85451	ATTR cardiomyopathy
95487	Atypical arterial duct
199627	Atypical autism
352723	Atypical Chédiak-Higashi syndrome
98824	Atypical chronic myeloid leukemia
1456	Atypical coarctation of aorta
314466	Atypical Demons-Meigs syndrome
314721	Atypical dentin dysplasia due to SMOC2 deficiency
309252	Atypical Gaucher disease due to saposin C deficiency
289863	Atypical glycine encephalopathy
98961	Atypical granular corneal dystrophy
238523	Atypical HCS
2134	Atypical hemolytic uremic syndrome
93581	Atypical hemolytic uremic syndrome with anti-factor H antibodies
93578	Atypical hemolytic uremic syndrome with B factor anomaly
93575	Atypical hemolytic uremic syndrome with C3 anomaly
357008	Atypical hemolytic uremic syndrome with DGKE deficiency
93579	Atypical hemolytic uremic syndrome with H factor anomaly
93580	Atypical hemolytic uremic syndrome with I factor anomaly
93576	Atypical hemolytic uremic syndrome with MCP/CD46 anomaly
217023	Atypical hemolytic uremic syndrome with thrombomodulin anomaly
2134	Atypical HUS
93581	Atypical HUS with anti-factor H antibodies
93578	Atypical HUS with B factor anomaly
93575	Atypical HUS with C3 anomaly
357008	Atypical HUS with DGKE deficiency
93579	Atypical HUS with H factor anomaly
93580	Atypical HUS with I factor anomaly
93576	Atypical HUS with MCP/CD46 anomaly
217023	Atypical HUS with thrombomodulin anomaly
238523	Atypical hypotonia - cystinuria syndrome
86797	Atypical lichen myxedematosus
99971	Atypical lipoma
99971	Atypical lipomatous tumor
247768	Atypical Mayer-Rokitansky-Küster-Hauser syndrome
314466	Atypical Meigs syndrome
247768	Atypical MRKH syndrome
261501	Atypical Norrie disease due to del(X) (p11.3)
261501	Atypical Norrie disease due to monosomy Xp11.3
261501	Atypical Norrie disease due to Xp11.3 microdeletion
216873	Atypical pantothenate kinase associated neurodegeneration
251902	Atypical papilloma of choroid plexus
95487	Atypical patent ductus arteriosus
79474	Atypical progeroid syndrome
99750	Atypical progressive supranuclear palsy
99750	Atypical PSP
3095	Atypical Rett syndrome
247768	Atypical Rokitansky syndrome
3095	Atypical RTT
99966	Atypical teratoid tumor
251891	Atypical teratoid/rhabdoid tumor
90393	Atypical tuberous myxedema of Jadassohn-Dosseker
79474	Atypical Werner syndrome
16	Atypical X-linked achromatopsia
166415	Audiogenic seizures
1074	Aughton-Hufnagle syndrome
1488	Aural atresia - multiple congenital anomalies - intellectual deficit
77300	Auricular abnormalities - cleft lip with or without cleft palate - ocular abnormalities
137888	Auriculo-condylar syndrome
71270	Auriculoocular anomalies - cleft lip
114	Auriculoosteodysplasia
1995	Ausems-Wittebol Post-Hennekam syndrome
585	Austin type juvenile sulfatidosis
137911	Autism - facial port-wine stain
352490	Autism spectrum disorder due to AUTS2 deficiency
308410	Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency
324636	Autoerythrocyte sensitization syndrome
85138	Autoimmune Addison's disease

ORPHA Number	Disease name
85138	Autoimmune adrenalitis
94075	Autoimmune enteropathy
37042	Autoimmune enteropathy type 1
103916	Autoimmune enteropathy type 2
103917	Autoimmune enteropathy type 3
90033	Autoimmune hemolytic anemia, warm type
2137	Autoimmune hepatitis
36913	Autoimmune hypoparathyroidism
3453	Autoimmune hypoparathyroidism - chronic candidiasis - Addison's disease
3453	Autoimmune hypoparathyroidism - chronic candidosis - Addison's disease
3261	Autoimmune lymphoproliferative syndrome
268114	Autoimmune lymphoproliferative syndrome type 4
268114	Autoimmune lymphoproliferative syndrome type IV
275517	Autoimmune lymphoproliferative syndrome with recurrent infections
589	Autoimmune myasthenia gravis
206569	Autoimmune necrotizing myopathy
103919	Autoimmune pancreatitis
280302	Autoimmune pancreatitis type 1
280315	Autoimmune pancreatitis type 2
747	Autoimmune PAP
3453	Autoimmune polyendocrine syndrome type 1
3143	Autoimmune polyendocrine syndrome type 2
227982	Autoimmune polyendocrine syndrome type 3
227990	Autoimmune polyendocrine syndrome type 4
3453	Autoimmune polyendocrinopathy - candidiasis - ectodermal dystrophy syndrome
3453	Autoimmune polyendocrinopathy - candidosis - ectodermal dystrophy syndrome
3453	Autoimmune polyendocrinopathy type 1
3143	Autoimmune polyendocrinopathy type 2
227982	Autoimmune polyendocrinopathy type 3
227990	Autoimmune polyendocrinopathy type 4
3453	Autoimmune polyglandular syndrome type 1
3143	Autoimmune polyglandular syndrome type 2
227982	Autoimmune polyglandular syndrome type 3
227990	Autoimmune polyglandular syndrome type 4
747	Autoimmune pulmonary alveolar proteinosis
71203	Autoimmune thrombocytopenia
3143	Autoimmune thyroid disease and/or type 1 diabetes - Addison disease
324530	Autoinflammation and PLCG2-associated antibody deficiency and immune dysregulation
210115	Autoinflammatory disease due to interleukin-1 receptor antagonist deficiency
329173	Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis
33110	Autosomal agammaglobulinemia
280365	Autosomal codominant severe lipodystrophic laminopathy
88918	Autosomal dominant Alport syndrome
1810	Autosomal dominant anhidrotic ectodermal dysplasia
314399	Autosomal dominant aplasia and myelodysplasia
314399	Autosomal dominant aplastic anemia and myelodysplasia
64746	Autosomal dominant axonal Charcot-Marie-Tooth disease
1945	Autosomal dominant BECRS
1216	Autosomal dominant benign distal spinal muscular atrophy
314652	Autosomal dominant beta2-microglobulinic amyloidosis
93304	Autosomal dominant brachyolmia
169189	Autosomal dominant centronuclear myopathy
94145	Autosomal dominant cerebellar ataxia type 1
94148	Autosomal dominant cerebellar ataxia type 3
94149	Autosomal dominant cerebellar ataxia type 4
94145	Autosomal dominant cerebellar ataxia type I
94148	Autosomal dominant cerebellar ataxia type III
94149	Autosomal dominant cerebellar ataxia type IV
314404	Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome
64746	Autosomal dominant Charcot-Marie-Tooth disease type 2
324611	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to KIF5A mutation
99946	Autosomal dominant Charcot-Marie-Tooth disease type 2A1
99947	Autosomal dominant Charcot-Marie-Tooth disease type 2A2
99936	Autosomal dominant Charcot-Marie-Tooth disease type 2B
99937	Autosomal dominant Charcot-Marie-Tooth disease type 2C
99938	Autosomal dominant Charcot-Marie-Tooth disease type 2D
99939	Autosomal dominant Charcot-Marie-Tooth disease type 2E
99940	Autosomal dominant Charcot-Marie-Tooth disease type 2F
99941	Autosomal dominant Charcot-Marie-Tooth disease type 2G
99942	Autosomal dominant Charcot-Marie-Tooth disease type 2I
99943	Autosomal dominant Charcot-Marie-Tooth disease type 2J
99944	Autosomal dominant Charcot-Marie-Tooth disease type 2K
99945	Autosomal dominant Charcot-Marie-Tooth disease type 2L
228179	Autosomal dominant Charcot-Marie-Tooth disease type 2M
228174	Autosomal dominant Charcot-Marie-Tooth disease type 2N
284232	Autosomal dominant Charcot-Marie-Tooth disease type 2O
300319	Autosomal dominant Charcot-Marie-Tooth disease type 2P
329258	Autosomal dominant Charcot-Marie-Tooth disease type 2Q
98975	Autosomal dominant CHED
306561	Autosomal dominant childhood-onset cortical cataract
306561	Autosomal dominant childhood-onset progressive cortical cataract
1455	Autosomal dominant coarctation of aorta
1216	Autosomal dominant congenital benign spinal muscular atrophy
98975	Autosomal dominant congenital hereditary endothelial dystrophy
86814	Autosomal dominant cortical myoclonus and epilepsy
90348	Autosomal dominant cutis laxa
75381	Autosomal dominant cystoid macular edema
65753	Autosomal dominant demyelinating Charcot-Marie-Tooth disease
2337	Autosomal dominant diffuse palmoplantar keratoderma, Norrbotten type
139518	Autosomal dominant distal juvenile spinal muscular atrophy type 1
93608	Autosomal dominant distal renal tubular acidosis
98808	Autosomal dominant dopa-responsive dystonia
231568	Autosomal dominant dystrophic epidermolysis bullosa, Pasini and Cockayne-Touraine types
300576	Autosomal dominant ectodermal dysplasia-cancer predisposition syndrome syndrome
98853	Autosomal dominant Emery-Dreifuss muscular dystrophy
101046	Autosomal dominant epilepsy with auditory features

ORPHA Number	Disease name
73229	Autosomal dominant familial hematuria - retinal arteriolar tortuosity - contractures
100988	Autosomal dominant familial spastic paraplegia type 3
329466	Autosomal dominant focal dystonia, DYT25
93963	Autosomal dominant focal dystonia, DYT7 type
2024	Autosomal dominant gingival fibromatosis
2024	Autosomal dominant gingival hyperplasia
139491	Autosomal dominant hereditary hemochromatosis
2314	Autosomal dominant HIES
2314	Autosomal dominant hyper IgE syndrome
2314	Autosomal dominant hyperimmunoglobulin E syndrome
276580	Autosomal dominant hyperinsulinemic hypoglycemia due to Kir6.2 deficiency
276575	Autosomal dominant hyperinsulinemic hypoglycemia due to SUR1 deficiency
276580	Autosomal dominant hyperinsulinism due to Kir6.2 deficiency
276575	Autosomal dominant hyperinsulinism due to SUR1 deficiency
428	Autosomal dominant hypocalcemia
1810	Autosomal dominant hypohidrotic ectodermal dysplasia
89937	Autosomal dominant hypophosphatemia
89937	Autosomal dominant hypophosphatemic rickets
90114	Autosomal dominant intermediate Charcot-Marie-Tooth disease
100043	Autosomal dominant intermediate Charcot-Marie-Tooth disease type A
100044	Autosomal dominant intermediate Charcot-Marie-Tooth disease type B
100045	Autosomal dominant intermediate Charcot-Marie-Tooth disease type C
100046	Autosomal dominant intermediate Charcot-Marie-Tooth disease type D
93114	Autosomal dominant intermediate Charcot-Marie-Tooth disease type E
352670	Autosomal dominant intermediate Charcot-Marie-Tooth disease type F
324585	Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain
90635	Autosomal dominant isolated neurosensory deafness type DFNA
90635	Autosomal dominant isolated neurosensory hearing loss type DFNA
90635	Autosomal dominant isolated sensorineural deafness type DFNA
90635	Autosomal dominant isolated sensorineural hearing loss type DFNA
93325	Autosomal dominant Kenny-Caffey syndrome
2334	Autosomal dominant keratitis
293936	Autosomal dominant keratoconus with early-onset anterior polar cataracts
503	Autosomal dominant Larsen syndrome
67042	Autosomal dominant late-onset retinal degeneration
101046	Autosomal dominant lateral temporal lobe epilepsy
313808	Autosomal dominant leukoencephalopathy with neuroaxonal spheroids
266	Autosomal dominant limb-girdle muscular dystrophy type 1A
264	Autosomal dominant limb-girdle muscular dystrophy type 1B
265	Autosomal dominant limb-girdle muscular dystrophy type 1C
34516	Autosomal dominant limb-girdle muscular dystrophy type 1D
34517	Autosomal dominant limb-girdle muscular dystrophy type 1E
55595	Autosomal dominant limb-girdle muscular dystrophy type 1F
55596	Autosomal dominant limb-girdle muscular dystrophy type 1G
238755	Autosomal dominant limb-girdle muscular dystrophy type 1H
140957	Autosomal dominant macrothrombocytopenia
88950	Autosomal dominant medullary cystic kidney disease with hyperuricemia
34149	Autosomal dominant medullary cystic kidney disease with or without hyperuricemia
88949	Autosomal dominant medullary cystic kidney disease without hyperuricemia
319581	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency
319589	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency
319581	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 1 deficiency
319589	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 2 deficiency
330041	Autosomal dominant methemoglobinemia
2514	Autosomal dominant microcephaly
319581	Autosomal dominant MSMD due to partial IFNgammaR1 deficiency
319589	Autosomal dominant MSMD due to partial IFNgammaR2 deficiency
319581	Autosomal dominant MSMD due to partial interferon gamma receptor 1 deficiency
319589	Autosomal dominant MSMD due to partial interferon gamma receptor 2 deficiency
65743	Autosomal dominant multiple pterygium syndrome
99846	Autosomal dominant myoglobinuria
79153	Autosomal dominant nail dysplasia
329211	Autosomal dominant neovascular inflammatory vitreoretinopathy
34149	Autosomal dominant nephronophthisis
98784	Autosomal dominant nocturnal frontal lobe epilepsy
178469	Autosomal dominant nonsyndromic intellectual deficit
90635	Autosomal dominant nonsyndromic neurosensory deafness type DFNA
90635	Autosomal dominant nonsyndromic neurosensory hearing loss type DFNA
90635	Autosomal dominant nonsyndromic sensorineural deafness type DFNA
90635	Autosomal dominant nonsyndromic sensorineural hearing loss type DFNA
93328	Autosomal dominant omodysplasia
306588	Autosomal dominant Opitz BBB/G syndrome
306588	Autosomal dominant Opitz G/BBB syndrome
306588	Autosomal dominant Opitz syndrome
67036	Autosomal dominant optic atrophy and cataract
3212	Autosomal dominant optic atrophy and congenital deafness
255117	Autosomal dominant optic atrophy and late-onset deafness
250932	Autosomal dominant optic atrophy and peripheral neuropathy
1215	Autosomal dominant optic atrophy plus syndrome
67036	Autosomal dominant optic atrophy type 3
98673	Autosomal dominant optic atrophy, classic type
98673	Autosomal dominant optic atrophy, Kjer type
2783	Autosomal dominant osteopetrosis type 1
1798	Autosomal dominant osteosclerosis, Stanescu type
2790	Autosomal dominant osteosclerosis, Worth type
1010	Autosomal dominant palmoplantar hyperkeratosis and congenital alopecia
1010	Autosomal dominant palmoplantar keratoderma and congenital alopecia
32960	Autosomal dominant periodic fever
88924	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis
2924	Autosomal dominant polycystic liver disease
1300	Autosomal dominant popliteal pterygium syndrome
34528	Autosomal dominant primary hypomagnesemia with hypocalcuria
2964	Autosomal dominant prognathism

ORPHA Number	Disease name
254892	Autosomal dominant progressive external ophthalmoplegia
88659	Autosomal dominant progressive nephropathy with hypertension
314889	Autosomal dominant proximal renal tubular acidosis
171871	Autosomal dominant pseudohypoaldosteronism type 1
209867	Autosomal dominant rhegmatogenous retinal detachment
3107	Autosomal dominant Robinow syndrome
247511	Autosomal dominant secondary erythrocytosis
247511	Autosomal dominant secondary polycythemia
98808	Autosomal dominant Segawa syndrome
486	Autosomal dominant severe congenital neutropenia
140481	Autosomal dominant slowed nerve conduction velocity
251282	Autosomal dominant spastic ataxia 1
100984	Autosomal dominant spastic paraplegia type 3
100985	Autosomal dominant spastic paraplegia type 4
100988	Autosomal dominant spastic paraplegia type 6
100989	Autosomal dominant spastic paraplegia type 8
100990	Autosomal dominant spastic paraplegia type 9
100991	Autosomal dominant spastic paraplegia type 10
100993	Autosomal dominant spastic paraplegia type 12
100994	Autosomal dominant spastic paraplegia type 13
100998	Autosomal dominant spastic paraplegia type 17
100999	Autosomal dominant spastic paraplegia type 19
101009	Autosomal dominant spastic paraplegia type 29
101011	Autosomal dominant spastic paraplegia type 31
320365	Autosomal dominant spastic paraplegia type 36
171612	Autosomal dominant spastic paraplegia type 37
171617	Autosomal dominant spastic paraplegia type 38
320355	Autosomal dominant spastic paraplegia type 41
171863	Autosomal dominant spastic paraplegia type 42
94147	Autosomal dominant spinocerebellar ataxia type 7
1797	Autosomal dominant spondylocostal dysostosis

ORPHA Number	Disease name
1797	Autosomal dominant spondylocostal dysplasia
228169	Autosomal dominant striatal neurodegeneration
98757	Autosomal dominant striatonigral degeneration
3086	Autosomal dominant vitreoretinochoroidopathy
88919	Autosomal recessive Alport syndrome
1027	Autosomal recessive amelia
248	Autosomal recessive anhidrotic ectodermal dysplasia
1116	Autosomal recessive aplasia cutis
139485	Autosomal recessive ataxia due to coenzyme Q10 deficiency
247815	Autosomal recessive ataxia due to PEX10 deficiency
139485	Autosomal recessive ataxia due to ubiquinone deficiency
88644	Autosomal recessive ataxia, Beauce type
91024	Autosomal recessive axonal Charcot-Marie-Tooth disease type 2
101101	Autosomal recessive axonal Charcot-Marie-Tooth disease type 2B2
101097	Autosomal recessive axonal Charcot-Marie-Tooth disease type 2K
91024	Autosomal recessive axonal Charcot-Marie-Tooth disease type 4C
98856	Autosomal recessive axonal CMT4C1
101102	Autosomal recessive axonal CMT4C2
101101	Autosomal recessive axonal CMT4C3
101097	Autosomal recessive axonal CMT4C4
324442	Autosomal recessive axonal neuropathy with neuromyotonia
139455	Autosomal recessive bestrophinopathy
169186	Autosomal recessive centronuclear myopathy
95433	Autosomal recessive cerebellar ataxia - blindness - deafness
352403	Autosomal recessive cerebellar ataxia - cognitive defect
284282	Autosomal recessive cerebellar ataxia - epilepsy - intellectual deficit
284271	Autosomal recessive cerebellar ataxia - psychomotor retardation
95434	Autosomal recessive cerebellar ataxia - saccadic intrusion
352641	Autosomal recessive cerebellar ataxia due to GBA2 deficiency
88644	Autosomal recessive cerebellar ataxia type 1
352641	Autosomal recessive cerebellar ataxia with late-onset spasticity
1170	Autosomal recessive cerebelloparenchymal disorder type 3
324442	Autosomal recessive Charcot-Marie-Tooth disease type 2 with neuromyotonia

ORPHA Number	Disease name
98856	Autosomal recessive Charcot-Marie-Tooth disease type 2B1
101097	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness
90118	Autosomal recessive Charcot-Marie-Tooth disease, Ouvrier type
293603	Autosomal recessive CHED
217046	Autosomal recessive childhood-onset cortical cataract
2518	Autosomal recessive chorioretinopathy-microcephaly
100981	Autosomal recessive complex HSP
100981	Autosomal recessive complex spastic paraplegia
100981	Autosomal recessive complex SPG
100981	Autosomal recessive complicated HSP
100981	Autosomal recessive complicated spastic paraplegia
100981	Autosomal recessive complicated SPG
324262	Autosomal recessive congenital cerebellar ataxia due to metabotropic glutamate receptor 1 deficiency
324262	Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency
293603	Autosomal recessive congenital hereditary endothelial dystrophy
99951	Autosomal recessive congenital hypomyelinating neuropathy
260305	Autosomal recessive congenital sideroblastic anemia
90349	Autosomal recessive cutis laxa type 1
90350	Autosomal recessive cutis laxa type 2
357074	Autosomal recessive cutis laxa type 2, classic type
357074	Autosomal recessive cutis laxa type 2, Debré type
357064	Autosomal recessive cutis laxa type 2, progeroid type
357058	Autosomal recessive cutis laxa type 2A
357064	Autosomal recessive cutis laxa type 2B
90349	Autosomal recessive cutis laxa with severe systemic involvement
90349	Autosomal recessive cutis laxa, pulmonary emphysema type
79500	Autosomal recessive deafness-onychodystrophy syndrome
64749	Autosomal recessive demyelinating Charcot-Marie-Tooth
2776	Autosomal recessive distal osteolysis syndrome
93611	Autosomal recessive distal renal tubular acidosis with deafness
93611	Autosomal recessive distal renal tubular acidosis with hearing loss
93609	Autosomal recessive distal renal tubular acidosis without deafness
93609	Autosomal recessive distal renal tubular acidosis without hearing loss

ORPHA Number	Disease name
98920	Autosomal recessive distal spinal muscular atrophy type 1
139552	Autosomal recessive distal spinal muscular atrophy type 2
139547	Autosomal recessive distal spinal muscular atrophy type 3
206580	Autosomal recessive distal spinal muscular atrophy type 4
314485	Autosomal recessive distal spinal muscular atrophy type 5
101150	Autosomal recessive dopa-responsive dystonia
79408	Autosomal recessive dystrophic epidermolysis bullosa generalisata gravis
89842	Autosomal recessive dystrophic epidermolysis bullosa generalisata mitis
89842	Autosomal recessive dystrophic epidermolysis bullosa, generalized other
79408	Autosomal recessive dystrophic epidermolysis bullosa, Hallopeau-Siemens type
238569	Autosomal recessive early-onset IBD
238569	Autosomal recessive early-onset inflammatory bowel disease
98855	Autosomal recessive Emery-Dreifuss muscular dystrophy
89838	Autosomal recessive epidermolysis bullosa simplex
289586	Autosomal recessive exfoliative ichthyosis
1974	Autosomal recessive facio-digito-genital syndrome
329329	Autosomal recessive frontotemporal pachygyria
169446	Autosomal recessive HIES
169446	Autosomal recessive hyper IgE syndrome
331226	Autosomal recessive hyper IgE syndrome due to TYK2 deficiency
79644	Autosomal recessive hyperinsulinemic hypoglycemia due to Kir6.2 deficiency
79643	Autosomal recessive hyperinsulinemic hypoglycemia due to SUR1 deficiency
79644	Autosomal recessive hyperinsulinism due to Kir6.2 deficiency
79643	Autosomal recessive hyperinsulinism due to SUR1 deficiency
248	Autosomal recessive hypohidrotic ectodermal dysplasia
289176	Autosomal recessive hypophosphatemic rickets
300547	Autosomal recessive infantile hypercalcemia
352530	Autosomal recessive intellectual deficit due to TRAPPC9 deficiency
217055	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A
254334	Autosomal recessive intermediate Charcot-Marie-Tooth disease type B
210110	Autosomal recessive intermediate osteopetrosis
90636	Autosomal recessive isolated neurosensory deafness type DFNB
90636	Autosomal recessive isolated sensorineural deafness type DFNB
93324	Autosomal recessive Kenny-Caffey syndrome
263463	Autosomal recessive Larsen syndrome
33108	Autosomal recessive lethal multiple pterygium syndrome
314572	Autosomal recessive leukoencephalopathy with ischemic stroke-retinitis pigmentosa syndrome
352479	Autosomal recessive LGMD due to ISPD deficiency
352482	Autosomal recessive LGMD with cerebellar involvement
267	Autosomal recessive limb girdle muscular dystrophy type 2A
280333	Autosomal recessive limb-girdle muscular dystrophy - dystroglycanopathy type C7
352479	Autosomal recessive limb-girdle muscular dystrophy due to ISPD deficiency
254361	Autosomal recessive limb-girdle muscular dystrophy due to plectin deficiency
268	Autosomal recessive limb-girdle muscular dystrophy type 2B
353	Autosomal recessive limb-girdle muscular dystrophy type 2C
62	Autosomal recessive limb-girdle muscular dystrophy type 2D
119	Autosomal recessive limb-girdle muscular dystrophy type 2E
219	Autosomal recessive limb-girdle muscular dystrophy type 2F
34514	Autosomal recessive limb-girdle muscular dystrophy type 2G
1878	Autosomal recessive limb-girdle muscular dystrophy type 2H
34515	Autosomal recessive limb-girdle muscular dystrophy type 2I
140922	Autosomal recessive limb-girdle muscular dystrophy type 2J
86812	Autosomal recessive limb-girdle muscular dystrophy type 2K
206549	Autosomal recessive limb-girdle muscular dystrophy type 2L
206554	Autosomal recessive limb-girdle muscular dystrophy type 2M
206559	Autosomal recessive limb-girdle muscular dystrophy type 2N
206564	Autosomal recessive limb-girdle muscular dystrophy type 2O
254361	Autosomal recessive limb-girdle muscular dystrophy type 2Q
352482	Autosomal recessive limb-girdle muscular dystrophy with cerebellar involvement
206580	Autosomal recessive lower motor neuron disease with childhood onset
238505	Autosomal recessive lymphoproliferative disease
667	Autosomal recessive malignant osteopetrosis
655	Autosomal recessive medullary cystic kidney disease
319569	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency
319574	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR2 deficiency
319569	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 1 deficiency
319574	Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial interferon gamma receptor 2 deficiency
175	Autosomal recessive metaphyseal chondrodysplasia
319569	Autosomal recessive MSMD due to partial IFNgammaR1 deficiency
319574	Autosomal recessive MSMD due to partial IFNgammaR2 deficiency
319569	Autosomal recessive MSMD due to partial interferon gamma receptor 1 deficiency
319574	Autosomal recessive MSMD due to partial interferon gamma receptor 2 deficiency
2990	Autosomal recessive multiple pterygium syndrome
319332	Autosomal recessive myogenic AMC
319332	Autosomal recessive myogenic arthrogryposis multiplex congenita
280654	Autosomal recessive nail dysplasia
655	Autosomal recessive nephronophthisis
2990	Autosomal recessive non-lethal multiple pterygium syndrome
88616	Autosomal recessive nonsyndromic intellectual deficit
90636	Autosomal recessive nonsyndromic neurosensory deafness type DFNB
90636	Autosomal recessive nonsyndromic sensorineural deafness type DFNB
93329	Autosomal recessive omodysplasia
67047	Autosomal recessive optic atrophy type 3
99012	Autosomal recessive optic atrophy, OPA6 type
227976	Autosomal recessive optic atrophy, OPA7 type
178389	Autosomal recessive osteoclast-poor osteopetrosis with hypogammaglobulinemia
178389	Autosomal recessive osteopetrosis type 7
1366	Autosomal recessive palmoplantar hyperkeratosis and congenital alopecia
1366	Autosomal recessive palmoplantar keratoderma and congenital alopecia

ORPHA Number	Disease name
731	Autosomal recessive polycystic kidney disease
1234	Autosomal recessive popliteal pterygium syndrome
88628	Autosomal recessive posterior column ataxia and retinitis pigmentosa
30924	Autosomal recessive primary hypomagnesemia with normocalcuria and hypocalcemia
2512	Autosomal recessive primary microcephaly
254886	Autosomal recessive progressive external ophthalmoplegia
93607	Autosomal recessive proximal renal tubular acidosis
171876	Autosomal recessive pseudohypoaldosteronism type 1
100982	Autosomal recessive pure HSP
100982	Autosomal recessive pure spastic paraplegia
100982	Autosomal recessive pure SPG
255132	Autosomal recessive pyridoxine-refractory sideroblastic anemia
1507	Autosomal recessive Robinow syndrome
247378	Autosomal recessive secondary erythrocytosis not associated with VHL gene
247378	Autosomal recessive secondary erythrocytosis, non Chuvash type
247378	Autosomal recessive secondary polycythemia not associated with VHL gene
247378	Autosomal recessive secondary polycythemia, non Chuvash type
101150	Autosomal recessive Segawa syndrome
970	Autosomal recessive sensory radicular neuropathy
331176	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency
300345	Autosomal recessive SLE
254343	Autosomal recessive spastic ataxia - optic atrophy - dysarthria
98	Autosomal recessive spastic ataxia of Charlevoix-Saguenay
314603	Autosomal recessive spastic ataxia type 3
254343	Autosomal recessive spastic ataxia type 4
313772	Autosomal recessive spastic ataxia type 5
314603	Autosomal recessive spastic ataxia with leukoencephalopathy
101005	Autosomal recessive spastic paraplegia - disc herniation
100986	Autosomal recessive spastic paraplegia type 5A
99013	Autosomal recessive spastic paraplegia type 7
2822	Autosomal recessive spastic paraplegia type 11
100995	Autosomal recessive spastic paraplegia type 14
100996	Autosomal recessive spastic paraplegia type 15
209951	Autosomal recessive spastic paraplegia type 18
101000	Autosomal recessive spastic paraplegia type 20
101001	Autosomal recessive spastic paraplegia type 21
101003	Autosomal recessive spastic paraplegia type 23
101004	Autosomal recessive spastic paraplegia type 24
101005	Autosomal recessive spastic paraplegia type 25
101006	Autosomal recessive spastic paraplegia type 26
101007	Autosomal recessive spastic paraplegia type 27
101008	Autosomal recessive spastic paraplegia type 28
101010	Autosomal recessive spastic paraplegia type 30
171622	Autosomal recessive spastic paraplegia type 32
171629	Autosomal recessive spastic paraplegia type 35
139480	Autosomal recessive spastic paraplegia type 39
320370	Autosomal recessive spastic paraplegia type 43
320401	Autosomal recessive spastic paraplegia type 44
320396	Autosomal recessive spastic paraplegia type 45
320391	Autosomal recessive spastic paraplegia type 46
306511	Autosomal recessive spastic paraplegia type 48
320385	Autosomal recessive spastic paraplegia type 49
319199	Autosomal recessive spastic paraplegia type 53
320380	Autosomal recessive spastic paraplegia type 54
320375	Autosomal recessive spastic paraplegia type 55
320411	Autosomal recessive spastic paraplegia type 56
284332	Autosomal recessive spinocerebellar ataxia-6
284324	Autosomal recessive spinocerebellar ataxia-7
139485	Autosomal recessive spinocerebellar ataxia-9
284289	Autosomal recessive spinocerebellar ataxia-10
284271	Autosomal recessive spinocerebellar ataxia-11
284282	Autosomal recessive spinocerebellar ataxia-12
2311	Autosomal recessive spondylocostal dysostosis
250984	Autosomal recessive Stickler syndrome
300345	Autosomal recessive systemic lupus erythematosus
100982	Autosomal recessive uncomplicated HSP
100982	Autosomal recessive uncomplicated spastic paraplegia
100982	Autosomal recessive uncomplicated SPG
168629	Autosomal thrombocytopenia with normal platelets
352490	AUTS2 syndrome
96	AVED
98963	Avellino corneal dystrophy
58	AxD
98978	Axenfeld anomaly
782	Axenfeld syndrome
782	Axenfeld-Rieger syndrome
1834	Axial mesodermal dysplasia spectrum
2777	Axial osteosclerosis
168549	Axial spondylometaphyseal dysplasia
90119	Axonal Charcot-Marie-Tooth disease with acrodystrophy
101102	Axonal Charcot-Marie-Tooth disease with pyramidal involvement
209004	Axonal polyneuropathy associated with IgG/IgM/IgA monoclonal gammopathy
1435	Ayazi syndrome
284454	AZOOR
3471	Azoospermia - sinopulmonary infections
217034	Azoospermia due to maturation arrest
217034	Azoospermia due to meiosis defect
98757	Azorean disease of the nervous system
99121	Azygos continuation of the inferior caval vein
99121	Azygos continuation of the inferior vena cava
99121	Azygos continuation of the IVC
79332	B4GALT1-CDG syndrome
75496	B4GALT7-CDG syndrome
108	Babesiosis
36234	Bacterial toxic-shock syndrome
36234	Bacterial TSS
86814	BAFME
2819	Bahemuka-Brown syndrome
352577	Bainbridge-Roppers syndrome
1658	Baird syndrome
139471	Bakrania-Ragge syndrome
1223	Balantidiasis
1223	Balantidiosis
139450	Balikova-Vermeesch syndrome
99860	B-ALL
93395	Ballard syndrome

ORPHA Number	Disease name
1225	Baller-Gerold syndrome
66529	Ballooning cardiomyopathy
228165	Baló concentric sclerosis
634	Bamboo hair syndrome
1226	Bamforth syndrome
1226	Bamforth-Lazarus syndrome
98955	Band-shaped and whorled microcystic dystrophy of the corneal epithelium
1227	Bangstad syndrome
130	Bangungut
1228	Banki syndrome
109	Bannayan-Riley-Ruvalcaba syndrome
139507	Bantu siderosis
289539	BAP1-related tumor predisposition syndrome
1229	Baraitser-Brett-Piesowicz syndrome
2753	Baraitser-Burn syndrome
1229	Baraitser-Reardon syndrome
2995	Baraitser-Winter syndrome
2237	Barakat syndrome
1231	Barber-Say syndrome
110	Bardet-Biedl syndrome
34592	Bare lymphocyte syndrome type 1
572	Bare lymphocyte syndrome type 2
3317	Barnes syndrome
79087	Barraquer-Simons syndrome
111	Barth syndrome
64692	Bartonellosis due to Bartonella bacilliformis infection
50839	Bartonellosis due to Bartonella henselae infection
64694	Bartonellosis due to Bartonella quintana infection
2698	Bart-Pumphrey syndrome
1234	Bartsocas-Papas syndrome
112	Bartter syndrome
93605	Bartter syndrome type 3
89938	Bartter syndrome type 4
263417	Bartter syndrome type 5
263417	Bartter syndrome type V
263417	Bartter syndrome with hypocalcemia
377	Basal cell nevus syndrome
268829	Basal encephalocele
50810	Basel-Vanagaite-Sirota syndrome
244283	BASM syndrome
14	Bassen-Kornzweig disease
1875	Bassoe syndrome
100976	Bathing suit ichthyosis
1948	Battaglia-Neri syndrome
79264	Batten disease
1401	Baughman syndrome
166113	Bazex syndrome

ORPHA Number	Disease name
113	Bazex-Dupré-Christol syndrome
1800	Bazopoulou-Kyrkanidou syndrome
65284	BBGD
110	BBS
41751	BCD
67038	B-cell chronic lymphocytic leukemia
67038	B-cell chronic lymphoid leukemia
86852	B-cell prolymphocytic leukemia
312	BCIE
511	BCKD deficiency
511	BCKDH deficiency
67038	B-CLL
1236	Bd syndrome
247203	BDC
113	BDCS
115	Beals syndrome
115	Beals-Hecht syndrome
1059	Bean syndrome
1555	Beare-Stevenson cutis gyrata syndrome
98895	Becker dystrophinopathy
98895	Becker muscular dystrophy
64755	Becker nevus syndrome
116	Beckwith-Wiedemann syndrome
231127	Beckwith-Wiedemann syndrome due to 11p15 microdeletion
96076	Beckwith-Wiedemann syndrome due to 11p15 microduplication
231130	Beckwith-Wiedemann syndrome due to 11p15 translocation/inversion
231120	Beckwith-Wiedemann syndrome due to CDKN1C mutation
231117	Beckwith-Wiedemann syndrome due to imprinting defect of 11p15
238613	Beckwith-Wiedemann syndrome due to NSD1 mutation
96193	Beckwith-Wiedemann syndrome due to paternal uniparental disomy of chromosome 11
2572	Bedouin spastic ataxia syndrome
322	BEEC
1237	Beemer-Ertbruggen syndrome
275864	Behavioral variant of frontotemporal dementia
117	Behçet disease
1239	Behr syndrome
2705	Behrens-Baumann-Vogel syndrome
2810	Bell palsy
247203	Bellini carcinoma
247203	Bellini duct carcinoma
1240	Bellini syndrome
1492	Ben Ari-Shuper-Mimouni syndrome
100978	Benallegue-Lacete syndrome
1241	Bencze syndrome
86814	Benign adult familial myoclonic epilepsy

ORPHA Number	Disease name
610	Benign autosomal dominant myopathy
157997	Benign cephalic histiocytosis
98816	Benign childhood occipital epilepsy, Gastaut type
98815	Benign childhood occipital epilepsy, Panayiotopoulos type
2841	Benign chronic familial pemphigus of Hailey-Hailey
251287	Benign concentric annular macular dystrophy
254864	Benign COX deficiency
93955	Benign essential blepharospasm
71269	Benign exophthalmos syndrome
1429	Benign familial chorea
1945	Benign familial epilepsy of childhood with rolandic spikes
306	Benign familial infantile convulsions
306	Benign familial infantile epilepsy
306	Benign familial infantile seizures
163717	Benign familial mesial temporal lobe epilepsy
1949	Benign familial neonatal convulsions
1949	Benign familial neonatal epilepsy
1949	Benign familial neonatal seizures
140927	Benign familial neonatal-infantile seizures
209973	Benign familial nocturnal alternating hemiplegia in childhood
209973	Benign familial nocturnal alternating hemiplegia of childhood
65684	Benign focal amyotrophy
64545	Benign idiopathic neonatal seizures
166308	Benign infantile focal epilepsy with midline spikes and wave during sleep
166305	Benign infantile seizures associated to mild gastroenteritis
238624	Benign intracranial hypertension
285	Benign joint hypermobility syndrome
168816	Benign multicystic peritoneal mesothelioma
86909	Benign myoclonic epilepsy of infancy
140927	Benign neonatal-infantile epilepsy
166295	Benign non-familial infantile seizures
25968	Benign occipital epilepsy
342	Benign paroxysmal peritonitis
1179	Benign paroxysmal tonic upgaze of childhood with ataxia
71518	Benign paroxysmal torticollis of infancy
166299	Benign partial epilepsy of infancy with complex partial seizures
166302	Benign partial epilepsy with secondarily generalized seizures in infancy
166311	Benign partial infantile seizures
252131	Benign peripheral nerve sheath tumor
65682	Benign recurrent intrahepatic cholestasis

ORPHA Number	Disease name
99960	Benign recurrent intrahepatic cholestasis type 1
99961	Benign recurrent intrahepatic cholestasis type 2
342	Benign recurrent polyserositis
324581	Benign Samaritan congenital myopathy
252164	Benign schwannoma
180237	Benign tumor of fallopian tube
2198	Bennion-Patterson syndrome
54247	Benson syndrome
528	Beradinelli-Seip syndrome
171839	Berant syndrome
528	Berardinelli-Seip congenital lipodystrophy
2241	Berdon syndrome
647	Berlin breakage syndrome
274	Bernard-Soulier syndrome
133	Berylliosis
133	Beryllium granulomatosis
133	Beryllium pneumonosis
71269	BES
797	Besnier-Boeck-Schaumann disease
321	Bessel-Hagen disease
1243	Best disease
79332	Beta-1,4-galactosyltransferase deficiency
85446	Beta2-microglobulinic amyloidosis
65287	Beta-alanine synthase deficiency
309310	Beta-D-galactosidase deficiency
354	Beta-galactosidase-1 deficiency
584	Beta-glucuronidase deficiency
118	Beta-mannosidase deficiency
118	Beta-mannosidosis
329284	Beta-propeller protein-associated neurodegeneration
119	Beta-sarcoglycanopathy
848	Beta-thalassemia
231256	Beta-thalassemia - trichothiodystrophy
231393	Beta-thalassemia - X-linked thrombocytopenia
231230	Beta-thalassemia associated with another Hb anomaly
231230	Beta-thalassemia associated with another hemoglobin anomaly
231222	Beta-thalassemia intermedia
231214	Beta-thalassemia major
231386	Beta-thalassemia with other manifestations
65287	Beta-ureidopropionase deficiency
610	Bethlem myopathy
2114	Beukes familial hip dysplasia
2114	BFHD
306	BFIE
306	BFIS
127	BFLS

ORPHA Number	Disease name
140927	BFNIS
293284	BH4-responsive HPA/PKU
293284	BH4-responsive hyperphenylalaninemia/ phenylketonuria
98964	Biber-Haab-Dimmer dystrophy
180086	Bicervical bicornuate uterus
180106	Bicervical bicornuate uterus and blind hemi-vagina
180106	Bicervical bicornuate uterus one-eyed hemi-vagina
180111	Bicervical bicornuate uterus with patent cervix and vagina
2088	Bickel-Fanconi glycogenosis
2182	Bickers-Adams syndrome
79138	Bickerstaff brainstem encephalitis
3286	Bidirectional tachycardia
3286	Bidirectional tachycardia induced by catecholamine
1245	BIDS syndrome
1246	Biemond syndrome
141333	Biemond syndrome type 2
41751	Bietti crystalline corneoretinal dystrophy
41751	Bietti crystalline dystrophy
41751	Bietti crystalline retinopathy
1986	Bifid femur - monodactylous ectrodactyly
295006	Bifid great toes
295177	Bifid great toes, bilateral
295175	Bifid great toes, unilateral
295006	Bifid halluces
295177	Bifid halluces, bilateral
295006	Bifid hallux
295177	Bifid hallux, bilateral
295175	Bifid hallux, unilateral
2695	Bifid nose
217266	Bifid nose with or without anorectal and renal anomalies
99771	Bifid uvula
99771	Bifidity of the uvula
300	Bifunctional enzyme deficiency
319205	Bilateral adrenal hemorrhage
325124	Bilateral anorchia
2048	Bilateral anterior opercular syndrome
137920	Bilateral choanal atresia
208444	Bilateral frontal polymicrogyria
101070	Bilateral frontoparietal polymicrogyria
208447	Bilateral generalized polymicrogyria
319205	Bilateral massive adrenal hemorrhage
140963	Bilateral microtia - deafness - cleft palate
97364	Bilateral multicystic renal dysplasia
208441	Bilateral parasagittal parieto-occipital polymicrogyria
98889	Bilateral perisylvian polymicrogyria
268940	Bilateral polymicrogyria

ORPHA Number	Disease name
295150	Bilateral PPD2
1848	Bilateral renal agenesis
93173	Bilateral renal dysplasia
97362	Bilateral renal hypoplasia
357027	Bilateral retinoblastoma
1980	Bilateral striopallidodentate calcinosis
276066	Bile acid CoA ligase deficiency and defective amidation
70567	Bile duct cancer
30391	Biliary atresia
244283	Biliary atresia with splenic malformation syndrome
98836	Bilineal acute leukemia
205	Bilirubin uridinediphosphate glucuronosyltransferase deficiency
79234	Bilirubin uridinediphosphate glucuronosyltransferase deficiency type 1
79235	Bilirubin uridinediphosphate glucuronosyltransferase deficiency type 2
205	Bilirubin-UGT deficiency
79234	Bilirubin-UGT deficiency type 1
79235	Bilirubin-UGT deficiency type 2
1799	Billard-Toutain-Maheut syndrome
1248	Binder syndrome
3304	Bindewald-Ulmer-Müller syndrome
1249	Binswanger disease
79241	Biotinidase deficiency
65284	Biotin-responsive basal ganglia disease
65284	Biotin-thiamine-responsive basal ganglia disease
54247	Biparietal Alzheimer disease
99908	Bird fancier lung
2617	Bird headed-dwarfism, Montreal type
122	Birt-Hogg-Dube syndrome
79133	Bitemporal aplasia cutis congenital
2213	Bixler-Christian-Gorlin syndrome
285	BJHS
123	Björnstad syndrome
124	Blackfan-Diamond anemia
93930	Bladder exstrophy
322	Bladder exstrophy-epispadias-cloacal extrophy complex
37202	Bladder pain syndrome
1250	Blaichman syndrome
98922	Blake pouch cyst
254379	Blaschkoid lichen planus
254379	Blaschkoid LP
86870	Blastic NK-cell lymphoma
86870	Blastic plasmacytoid dendritic cell neoplasm
1834	Blastogenesis defect
90340	Blau syndrome
50945	BLC

ORPHA Number	Disease name
73271	Bleeding diathesis due to a collagen receptor defect
98885	Bleeding diathesis due to glycoprotein VI deficiency
98886	Bleeding diathesis due to integrin alpha2-beta1 deficiency
220443	Bleeding diathesis due to thromboxane synthesis deficiency
1253	Blepharochalasis - double lip
1997	Blepharo-cheilo-odontic syndrome
2353	Blepharo-facio-skeletal syndrome
1252	Blepharonasofacial malformation syndrome
126	Blepharophimosis - epicanthus inversus - ptosis
261559	Blepharophimosis - epicanthus inversus - ptosis due to 3q23 microdeletion
261572	Blepharophimosis - epicanthus inversus - ptosis due to a point mutation
261559	Blepharophimosis - epicanthus inversus - ptosis due to del(3)(q23)
261559	Blepharophimosis - epicanthus inversus - ptosis due to monosomy 3q23
261579	Blepharophimosis - epicanthus inversus - ptosis due to polyA expansion
2057	Blepharophimosis - ptosis - esotropia - syndactyly - short stature
1256	Blepharophimosis - radioulnar synostosis
1968	Blepharophimosis - telecanthus - microstomia
2728	Blepharophimosis syndrome, Ohdo type
126	Blepharophimosis types 1 and 2
261559	Blepharophimosis types 1 and 2 due to 3q23 microdeletion
261572	Blepharophimosis types 1 and 2 due to a point mutation
261559	Blepharophimosis types 1 and 2 due to del(3)(q23)
261559	Blepharophimosis types 1 and 2 due to monosomy 3q23
261579	Blepharophimosis types 1 and 2 due to polyA expansion
329255	Blepharophimosis-intellectual deficit syndrome due to UBE3B deficiency
293725	Blepharophimosis-intellectual deficit syndrome type V
293707	Blepharophimosis-intellectual deficit syndrome, Maat-Kievit-Brunner type
293707	Blepharophimosis-intellectual deficit syndrome, MKB type
2728	Blepharophimosis-intellectual deficit syndrome, Ohdo type
2728	Blepharophimosis-intellectual deficit syndrome, Say-Barber/ Biesecker/Young-Simpson type
3047	Blepharophimosis-intellectual deficit syndrome, SBBYS type
293725	Blepharophimosis-intellectual deficit syndrome, Verloes type
1258	Blepharoptosis - cleft palate - ectrodactyly - dental anomalies
1259	Blepharoptosis - myopia - ectopia lentis
93964	Blepharospasm - oromandibular dystonia
2626	Blethen-Wenick-Hawkins syndrome
171844	Blindness - scoliosis - arachnodactyly
464	Bloch-Siemens syndrome
464	Bloch-Sulzberger syndrome
50945	Blomstrand lethal chondrodysplasia
125	Bloom syndrome
2768	Blount disease
88629	Blue colour blindness
16	Blue cone monochromacy
16	Blue cone monochromatism
94086	Blue diaper syndrome
1059	Blue rubber bleb nevus
98989	Blue-dot cataract
319205	BMAH
98895	BMD
293725	BMRS type V
293707	BMRS, Maat-Kievit-Brunner type
293707	BMRS, MKB type
2728	BMRS, Ohdo type
293725	BMRS, Verloes type
217266	BNAR syndrome
217008	Bockenheimer syndrome
1292	BOD syndrome
2724	Boder syndrome
48686	Body cavity-based lymphoma
91135	Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency
797	Boeck sarcoid
797	Boeck's sarcoid
1297	BOFS
97297	Bohring syndrome
97297	Bohring-Opitz syndrome
84081	Boichis disease
319229	Bolivian hemorrhagic fever
85182	Bone dysplasia - medullary fibrosarcoma
1844	Bone dysplasia, Azouz type
2050	Bone fragility - craniosynostosis - proptosis - hydrocephalus
300284	Bone fragility-contractures-arterial rupture-deafness syndrome
88	Bone marrow failure
2934	Bonneau syndrome
163	Bonneau-Beaumont syndrome
2941	Bonnemann-Meinecke syndrome
1261	Bonnemann-Meinecke-Reich syndrome
53719	Bonnet-Dechaume-Blanc syndrome
1262	Book syndrome
1263	Boomerang dysplasia
1303	BOOP
1933	Booth-Haworth-Dilling syndrome
107	BOR syndrome
206473	Borderline ovarian epithelial tumor
127	Borjeson-Forssman-Lehmann syndrome
1264	Bork syndrome
90001	Bornholm eye disease
36273	Borrmann gastric cancer type 4
97297	BOS syndrome
69737	Bosley-Salih-Alorainy syndrome
2250	Bosma-Henkin-Christiansen syndrome
85128	Bothnia retinal dystrophy
128	Bothriocephalosis
1267	Botulism
1180	Boucher-Neuhäuser syndrome
805	Bourneville syndrome
83313	Boutonneuse fever
3331	Bowed tibiae - radial anomalies - osteopenia - fractures
1270	Bowen syndrome, Hutterite type
1270	Bowen-Conradi syndrome
97353	Boxer's dementia
50814	Boyadjiev-Jabs syndrome
2680	Boylan-Dew syndrome
329284	BPAN
70589	BPD
86870	BPDCN
86852	B-PLL
252131	BPNST
97342	Braak disease
2901	Brachial plexus neuritis
199	Brachmann-de Lange syndrome
1519	Brachycephalofrontonasal dysplasia
1272	Brachycephaly - deafness - cataract - intellectual deficit
2619	Brachydactylous dwarfism, Mseleni type
1276	Brachydactyly - arterial hypertension
1275	Brachydactyly - elbow wrist dysplasia
1275	Brachydactyly - joint dysplasia
2946	Brachydactyly - long thumb
1277	Brachydactyly - mesomelia - intellectual deficit - heart defects
1246	Brachydactyly - nystagmus - cerebellar ataxia
1278	Brachydactyly - preaxial hallux varus
2956	Brachydactyly - scoliosis - carpal fusion
294996	Brachydactyly of fingers
295130	Brachydactyly of fingers, bilateral
295128	Brachydactyly of fingers, unilateral
294998	Brachydactyly of toes
295134	Brachydactyly of toes, bilateral
295132	Brachydactyly of toes, unilateral

ORPHA Number	Disease name
93388	Brachydactyly type A1
93396	Brachydactyly type A2
93394	Brachydactyly type A4
93389	Brachydactyly type A5
93382	Brachydactyly type A6
93397	Brachydactyly type A7
93383	Brachydactyly type B
140908	Brachydactyly type B2
93384	Brachydactyly type C
93387	Brachydactyly type E
93395	Brachydactyly types B and E combined
93388	Brachydactyly, Farabee type
2946	Brachydactyly, long thumb type
93396	Brachydactyly, Mohr-Wriedt type
93397	Brachydactyly, Smorgasbord type
93394	Brachydactyly, Temtamy type
1001	Brachydactyly-intellectual deficit
3168	Brachydactyly-symphalangism syndrome
93409	Brachydactyly-syndactyly, Zhao type
93394	Brachymesophalangy II and V
1292	Brachymorphism - onychodysplasia - dysphalangism
1293	Brachyolmia
2899	Brachyolmia - amelogenesis imperfecta
93301	Brachyolmia type 1, Hobaek type
93303	Brachyolmia type 1, Toledo type
93302	Brachyolmia type 2
93304	Brachyolmia type 3
93302	Brachyolmia, Maroteaux type
79345	Brachytelephalangic chondrodysplasia punctata
1295	Brachytelephalangy - dysmorphism - Kallmann syndrome
441	Bradbury-Eggleston syndrome
52047	Braddock syndrome
3323	Braddock-Carey syndrome
1538	Braddock-Jones-Superneau syndrome
75374	Bradyopsia
178506	Brain calcification, Rajab type
168598	Brain demyelination due to methionine adenosyltransferase deficiency
352649	Brain dopamine-serotonin vesicular transport disease
75389	Brain malformation - congenital heart disease - postaxial polydactyly
36383	Brain small vessel disease with hemorrhage
36414	Brain stem tumor
209905	Brain-lung-thyroid syndrome
255182	Branched chain alpha-ketoacid dehydrogenase complex deficiency
511	Branched-chain ketoacid dehydrogenase deficiency
511	Branched-chain ketoaciduria
1296	Branchial dysplasia - intellectual deficit - inguinal hernia
50815	Branchiogenic deafness syndrome
1297	Branchio-oculo-facial syndrome
52429	Branchio-otic syndrome
107	Branchiootorenal syndrome
1299	Branchio-skeleto-genital syndrome
79133	Brauer syndrome
2669	Braun-Bayer syndrome
319239	Brazilian hemorrhagic fever
85284	BRESEK syndrome
85284	BRESHECK syndrome
65682	BRIC
99960	BRIC type 1
99961	BRIC type 2
99960	BRIC1
99961	BRIC2
99990	Brill-Zinsser disease
666	Brittle bone disease
90354	Brittle cornea syndrome
412	Broad-betalipoproteinemia
53347	Brody myopathy
97287	Bronchial carcinoid tumor
97287	Bronchial endocrine tumor
97287	Bronchial neuroendocrine tumor
1302	Bronchiolitis obliterans organizing pneumonia
1303	Bronchiolitis obliterans with obstructive pulmonary disease
2357	Bronchogenic cyst
70589	Bronchopulmonary dysplasia
1116	Bronspiegel-Zelnick syndrome
99829	Bronze John
79493	Brooke-Spiegler syndrome
97229	Brown-Vialetto-van Laere syndrome
109	BRRS
2353	BRSS
1304	Brucellosis
2771	Bruck syndrome
130	Brugada syndrome
1305	Brunner-Winter syndrome
528	Brunzell syndrome
47	Bruton type agammaglobulinemia
528	BSCL
79304	BSEP deficiency
100976	BSI
46489	BSLE
1980	BSPDC
65284	BTBGD
79241	BTD deficiency
111	BTHS
47	BTK-deficiency
2314	Buckley syndrome
131	Budd-Chiari syndrome
36258	Buerger disease
481	Bulbospinal muscular atrophy
2285	Bull-Nixon syndrome
312	Bullous congenital ichthyosiform erythroderma
312	Bullous congenital ichthyosiform erythroderma of Brock
280785	Bullous DCM
280785	Bullous diffuse cutaneous mastocytosis
1867	Bullous dystrophy, macular type
312	Bullous ichthyosis
36237	Bullous impetigo
33408	Bullous lichen planus
703	Bullous pemphigoid
46489	Bullous systemic lupus erythematosus
543	Burkitt lymphoma
1200	Burn-McKeown syndrome
800	Burton disease
800	Burton skeletal dysplasia
800	Burton syndrome
352763	Buschke scleredema
79501	Buschke-Fischer-Brauer syndrome
1306	Buschke-Ollendorff syndrome
99001	Butterfly-shaped pigment dystrophy
1307	Buttiens-Fryns syndrome
132	Butyrylcholinesterase deficiency
275864	bv-FTD
116	BWS
79306	Byler disease
231242	C- beta-thalassemia
1308	C syndrome
280133	C3 deficiency
93559	C3 deposition glomerulonephritis without proliferation
329931	C3 glomerulonephritis
329918	C3 glomerulopathy
85293	Cabezas syndrome
1309	Cacchi-Ricci disease
75377	CACD
135	CACH syndrome
2848	CACP syndrome
56425	CAD
136	CADASIL
1310	Caffey disease
418	CAH
90795	CAH due to 11-beta-hydroxylase deficiency
90793	CAH due to 17-alpha-hydroxylase deficiency
90791	CAH due to 3-beta-hydroxysteroid dehydrogenase deficiency

ORPHA Number	Disease name
1375	CAHMR syndrome
99429	CAIS
199260	Calcified aponeurotic fibroma
90290	Calcinosis - Raynaud phenomenon - esophageal involvement - sclerodactyly - telangiectasia
280065	Calciphylaxis cutis
1416	Calcium pyrophosphate deposition disease
1416	Calcium pyrophosphate dihydrate crystal deposition disease
300865	C-ALCL
1408	Calderon-Gonzalez Cantu syndrome
228123	California disease
83483	California encephalitis
85192	Calvarial doughnut lesions - bone fragility
3003	Camera syndrome
2163	Camero-Lituania-Cohen syndrome
79395	Camisa disease
83472	CAMOS syndrome
1318	Campomelia, Cumming type
140	Campomelic dwarfism
140	Campomelic dysplasia
1319	Camptobrachydactyly
1320	Camptocormia
1320	Camptocormism
376	Camptodactyly - cleft palate- clubfoot
1321	Camptodactyly - fibrous tissue hyperplasia - skeletal dysplasia
1323	Camptodactyly - joint contractures - facial skeletal defects
3447	Camptodactyly - overgrowth - unusual facies
85164	Camptodactyly - tall stature - scoliosis - hearing loss
1325	Camptodactyly - taurinuria
295016	Camptodactyly of fingers
1327	Camptodactyly syndrome, Guadalajara type 1
1326	Camptodactyly syndrome, Guadalajara type 2
2848	Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome
1766	CAMRQ syndrome
141194	CAMS 1
53719	CAMS 2
141199	CAMS 3
1328	Camurati-Engelmann disease
3261	Canale-Smith syndrome
141	Canavan disease
289385	Cancer diagnosed during pregnancy
180242	Cancer of fallopian tube
71505	Cancer-associated retinopathy
2700	Cancrum oris
325004	CANDLE syndrome
71279	CANOMAD syndrome
2233	Cantalamessa-Baldini-Ambrosi syndrome
1335	Cantrell pentalogy
1517	Cantu syndrome
171881	Cap disease
171881	Cap myopathy
160148	Cap polyposis
85199	CAP syndrome
166260	Capdepont teeth
75327	CAPE dystrophy
75327	CAPED
188	Capillary hyperpermeability syndrome
188	Capillary leak syndrome
79490	Capillary lymphangioma
79490	Capillary lymphatic malformation
137667	Capillary malformation - arteriovenous malformation
171839	Capra-DeMarco syndrome
71505	CAR syndrome
199354	CARASIL
147	Carbamoyl phosphate synthetase 1 deficiency
147	Carbamoylphosphate synthetase deficiency
79324	Carbohydrate deficient glycoprotein syndrome Ig
79325	Carbohydrate deficient glycoprotein syndrome Ih
79326	Carbohydrate deficient glycoprotein syndrome Ii
79318	Carbohydrate deficient glycoprotein syndrome type Ia
79319	Carbohydrate deficient glycoprotein syndrome type Ib
79320	Carbohydrate deficient glycoprotein syndrome type Ic
79321	Carbohydrate deficient glycoprotein syndrome type Id
79322	Carbohydrate deficient glycoprotein syndrome type Ie
79323	Carbohydrate deficient glycoprotein syndrome type If
79329	Carbohydrate deficient glycoprotein syndrome type IIa
79330	Carbohydrate deficient glycoprotein syndrome type IIb
79332	Carbohydrate deficient glycoprotein syndrome type IId
79333	Carbohydrate deficient glycoprotein syndrome type IIe
238459	Carbohydrate deficient glycoprotein syndrome type IIf
263508	Carbohydrate deficient glycoprotein syndrome type IIg
95428	Carbohydrate deficient glycoprotein syndrome type IIh
263487	Carbohydrate deficient glycoprotein syndrome type IIi
263501	Carbohydrate deficient glycoprotein syndrome type IIj
86309	Carbohydrate deficient glycoprotein syndrome type Ij
79327	Carbohydrate deficient glycoprotein syndrome type Ik
79328	Carbohydrate deficient glycoprotein syndrome type IL
91131	Carbohydrate deficient glycoprotein syndrome type Im
244310	Carbohydrate deficient glycoprotein syndrome type In
263494	Carbohydrate deficient glycoprotein syndrome type Io
280071	Carbohydrate deficient glycoprotein syndrome type Ip
300536	Carbohydrate deficient glycoprotein syndrome type Ir
329178	Carbohydrate deficient glycoprotein syndrome type Iu
306686	Carbon monoxide-induced parkinsonism
2785	Carbonic anhydrase 2 deficiency
213605	Carcinofibroma of the corpus uteri
100093	Carcinoid tumor and carcinoid syndrome
97289	Carcinoid tumor of the thymus
319308	Carcinoma associated with MITF/TFE translocation
300557	Carcinoma of the ampulla of Vater
56044	Carcinoma of the gallbladder
137628	Cardiac anomalies - heterotaxy
168796	Cardiac conduction disease - dilated cardiomyopathy - brachydactyly
1686	Cardiac diverticulum
208600	Cardiac papillary fibroelastoma
875	Cardiac tumor of the child
2872	Cardiocranial syndrome, Pfeiffer type
37553	Cardiodysrhythmic potassium-sensitive periodic paralysis
1340	Cardiofaciocutaneous syndrome
97292	Cardiogenic shock
2229	Cardiogenital syndrome
1342	Cardiomelic syndrome type 3
500	Cardiomyopathic lentiginosis
1345	Cardiomyopathy - cataract - hip spine disease
91130	Cardiomyopathy - hypotonia - lactic acidosis
90022	Cardiomyopathy - renal anomalies
70474	Cardiomyopathy with hypotonia due to cytochrome C oxidase deficiency
70474	Cardiomyopathy with myopathy due to COX deficiency

ORPHA Number	Disease name
111	Cardioskeletal myopathy with neutropenia and abnormal mitochondria
111	Cardioskeletal myopathy-neutropenia
3238	Cardiospondylocarpofacial syndrome
2072	Cardiovascular Gaucher disease
1358	Carey-Fineman-Ziter syndrome
79403	Carmi syndrome
2947	Carnevale-Hernandez-del Castillo syndrome
1359	Carney complex
319340	Carney complex variant
319340	Carney complex-trismus-pseudocamptodactyly syndrome
97286	Carney dyad
1359	Carney syndrome
139411	Carney triad
97286	Carney-Stratakis dyad
97286	Carney-Stratakis syndrome
158	Carnitine brain transporter deficiency
156	Carnitine palmitoyl transferase 1A deficiency
228302	Carnitine palmitoyl transferase deficiency type 2, adult-onset form
228305	Carnitine palmitoyl transferase deficiency type 2, hepatocardiomuscular form
228308	Carnitine palmitoyl transferase deficiency type 2, lethal systemic form
228302	Carnitine palmitoyl transferase deficiency type 2, myopathic form
228308	Carnitine palmitoyl transferase deficiency type 2, neonatal form
228305	Carnitine palmitoyl transferase deficiency type 2, severe infantile form
156	Carnitine palmitoyl transferase IA deficiency
157	Carnitine palmitoyl transferase II deficiency
228302	Carnitine palmitoyl transferase II deficiency, adult-onset form
228305	Carnitine palmitoyl transferase II deficiency, hepatocardiomuscular form
228308	Carnitine palmitoyl transferase II deficiency, lethal systemic form
228302	Carnitine palmitoyl transferase II deficiency, myopathic form
228308	Carnitine palmitoyl transferase II deficiency, neonatal form
228305	Carnitine palmitoyl transferase II deficiency, severe infantile form
157	Carnitine palmitoyltransferase deficiency type 2
158	Carnitine uptake deficiency
159	Carnitine-acylcarnitine translocase deficiency
1361	Carnosinase deficiency
1361	Carnosinemia
53035	Caroli disease
65759	Carpenter syndrome
93973	Carpenter-Waziri syndrome
2767	Carpotarsal osteochondromatosis
64692	Carrion disease
175	Cartilage-hair hypoplasia
65282	Carvajal syndrome
209908	CAS*
56425	CAS*
94095	Casamassima-Morton-Nance syndrome
1101	Cassia Stocco dos Santos syndrome
160	Castleman disease
2513	Castro Gago-Pombo-Novo syndrome
926	Catalase deficiency
1373	Cataract - aberrant oral frenula - growth delay
1366	Cataract - alopecia - sclerodactyly
1368	Cataract - ataxia - deafness
1383	Cataract - deafness - hypogonadism
1375	Cataract - hypertrichosis - intellectual deficit
1381	Cataract - intellectual deficit - anal atresia - urinary defects
1387	Cataract - intellectual deficit - hypogonadism
2712	Cataract - microphthalmia - radiculomegaly - septal heart defect
1380	Cataract - nephropathy - encephalopathy
98985	Cataract with Y-shaped suture opacities
98986	Cataract, Coppock-like
98987	Cataract, Hutterite type
314993	Cataract-congenital heart disease-neural tube defect syndrome
162	Cataract-glaucoma
1377	Cataract-microcornea syndrome
100990	Cataracts motor neuropathy - short stature - skeletal anomalies
567	CATCH 22
3286	Catecholaminergic polymorphic ventricular tachycardia
800	Catel-Hempel syndrome
1388	Catel-Manzke syndrome
195	Cat-eye syndrome
228337	Cathepsin D deficiency
60015	Catlin marks
50839	Cat-scratch disease
85164	CATSHL syndrome
276234	CATSPER1-related non syndromic male infertility
1123	Caudal appendage - deafness
1756	Caudal duplication
3027	Caudal dysplasia
3027	Caudal regression sequence
99994	Causalgia
1329	CAVC
99068	CAVC - Fallot tetralogy
99066	CAVC - left heart obstruction
99067	CAVC - ventricle hypoplasia
99066	CAVC type A
99067	CAVC type B
99068	CAVC type C
2124	Cavernous hemangiomas of face - supraumbilical midline raphe
79489	Cavernous lymphangioma
79489	Cavernous lymphatic malformation
165958	Cavitary myiasis
567	Cayler cardiofacial syndrome
94122	Cayman ataxia
70567	CCA
115	CCA syndrome
99781	CCAL1
99782	CCAL2
2444	CCAM
280832	CCAM type I
280840	CCAM type II
280847	CCAM type III
98972	CCDF
48431	CCFDN
2008	CCGE syndrome
99827	CCHF
661	CCHS
289499	CCMCO
319276	CCRCC
2505	CCSF
280779	CCV
86870	CD4+/CD56+ hematodermic neoplasm
85	CDA
293825	CDA due to KLF1 mutation
98869	CDA type 1
98873	CDA type 2
98870	CDA type 3
85199	CDAGS syndrome
98869	CDAI
98873	CDAII
98870	CDAIII
247203	CDC
91131	CDG syndrome type 1m
280071	CDG syndrome type 1p
95428	CDG syndrome type 2h
314667	CDG syndrome type 2k
356961	CDG syndrome type 2m
79318	CDG syndrome type Ia
79319	CDG syndrome type Ib
79320	CDG syndrome type Ic
79321	CDG syndrome type Id
79322	CDG syndrome type Ie

*Caution: one same acronym may correspond to different diseases in medical terms. Please refer to the full name of the disease to get the correct Orpha code.

ORPHA Number	Disease name
79323	CDG syndrome type If
79324	CDG syndrome type Ig
79325	CDG syndrome type Ih
79326	CDG syndrome type Ii
79329	CDG syndrome type IIa
79330	CDG syndrome type IIb
99843	CDG syndrome type IIc
79332	CDG syndrome type IId
79333	CDG syndrome type IIe
238459	CDG syndrome type IIf
263508	CDG syndrome type IIg
95428	CDG syndrome type IIh
263487	CDG syndrome type IIi
263501	CDG syndrome type IIj
314667	CDG syndrome type IIk
356961	CDG syndrome type IIm
86309	CDG syndrome type Ij
79327	CDG syndrome type Ik
79328	CDG syndrome type IL
91131	CDG syndrome type Im
244310	CDG syndrome type In
263494	CDG syndrome type Io
300536	CDG syndrome type Ir
324422	CDG syndrome type Is
329178	CDG syndrome type Iu
79318	CDG1A
79319	CDG1B
79320	CDG1C
79321	CDG1D
79322	CDG1E
79323	CDG1F
79324	CDG1G
79325	CDG1H
79326	CDG1I
86309	CDG1J
79327	CDG1K
79328	CDG1L
91131	CDG1M
244310	CDG1N
263494	CDG1O
280071	CDG1P
324737	CDG1Q
300536	CDG1R
324422	CDG1S
329178	CDG1U
79329	CDG2A
79330	CDG2B
99843	CDG2C
79332	CDG2D
79333	CDG2E

ORPHA Number	Disease name
238459	CDG2F
263508	CDG2G
95428	CDG2H
263487	CDG2I
263501	CDG2J
314667	CDG2K
356961	CDG2M
79318	CDG-Ia
79319	CDG-Ib
79320	CDG-Ic
79321	CDG-Id
79322	CDG-Ie
79323	CDG-If
79324	CDG-Ig
79325	CDG-Ih
79326	CDG-Ii
79329	CDG-IIa
79330	CDG-IIb
99843	CDG-IIc
79332	CDG-IId
79333	CDG-IIe
238459	CDG-IIf
263508	CDG-IIg
95428	CDG-IIh
263487	CDG-IIi
263501	CDG-IIj
314667	CDG-IIk
356961	CDG-IIm
86309	CDG-Ij
79327	CDG-Ik
79328	CDG-IL
91131	CDG-Im
244310	CDG-In
263494	CDG-Io
280071	CDG-Ip
324737	CDG-Iq
300536	CDG-Ir
324422	CDG-Is
319646	CDGIt
329178	CDG-Iu
2140	CDH
1529	CDHS
178029	CDI
1490	CDPD
35173	CDPX2
1459	CEC
2718	Cecato de Lima-Pinheiro syndrome
1515	CED
66631	CEDNIK syndrome
1459	Celiac disease, epilepsy and cerebral calcification syndrome

ORPHA Number	Disease name
293208	Celiac trunk compression syndrome
93942	Celosomia
3258	Cenani syndactyly
3258	Cenani-Lenz syndactyly
3258	Cenani-Lenz syndrome
75377	Central areolar choroidal dystrophy
75377	Central areolar choroidal sclerosis
75327	Central areolar pigment epithelial dystrophy
2431	Central bilateral macrogyria
98972	Central cloudy corneal dystrophy of Francois
98972	Central cloudy dystrophy of Francois
661	Central congenital hypoventilation syndrome
597	Central core disease
178029	Central diabetes insipidus
99832	Central hypothyroidism due to TRH receptor deficiency
3240	Central nervous system calcification - deafness - tubular acidosis - anemia
73256	Central neurocytoma
2398	Central non-encapsulated lipomatosis
295004	Central polydactyly of fingers
295173	Central polydactyly of fingers, bilateral
295171	Central polydactyly of fingers, unilateral
295010	Central polydactyly of foot
295004	Central polydactyly of hand
295010	Central polydactyly of toes
295185	Central polydactyly of toes, bilateral
295183	Central polydactyly of toes, unilateral
759	Central precocious puberty
75327	Central retinal pigment epithelial dystrophy
90156	Centrifugal lipodystrophy
89841	Centripetal dystrophic epidermolysis bullosa
89841	Centripetal recessive dystrophic epidermolysis bullosa
89841	Centripetalis recessive dystrophic epidermolysis bullosa
319160	Centronuclear myopathy type 4
1945	Centrotemporal epilepsy
79277	CEP
79506	CEPT deficiency
333	Ceramidase deficiency
1174	Cerebellar ataxia - ectodermal dysplasia
1173	Cerebellar ataxia - hypogonadism
1766	Cerebellar ataxia - intellectual deficit - dysequilibrium syndrome
83472	Cerebellar ataxia - intellectual deficit - optic atrophy - skin abnormalities
276183	Cerebellar ataxia with azospermia and intellectual deficiency

ORPHA Number	Disease name
94122	Cerebellar ataxia, Cayman type
314404	Cerebellar ataxia-deafness-narcolepsy syndrome
97249	Cerebellar atrophy with progressive microcephaly
2246	Cerebellar hypoplasia - tapetoretinal degeneration
251931	Cerebellar liponeurocytoma
251858	Cerebellar neuroblastoma
94145	Cerebellar plus syndrome
94147	Cerebellar syndrome - pigmentary maculopathy
1454	Cerebellar vermis hypoplasia - oligophrenia - congenital ataxia - coloboma - hepatic fibrosis
2318	Cerebello-oculo-renal syndrome
475	Cerebelloparenchymal disorder IV
1532	Cerebellotrigeminal - dermal dysplasia
1397	Cerebellum agenesis - hydrocephaly
46724	Cerebral arteriovenous fistula
46724	Cerebral arteriovenous malformation
46724	Cerebral arteriovenous shunt
136	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
199354	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy
382	Cerebral creatine deficiency
66631	Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma syndrome
821	Cerebral gigantism
2081	Cerebral gigantism - jaw cysts
2691	Cerebral gigantism, Nevo type
77261	Cerebral juvenile and adult form of Gaucher disease
221126	Cerebral proliferative glomeruloid vasculopathy
329217	Cerebral sinovenous thrombosis
1393	Cerebro-costo-mandibular syndrome
141194	Cerebrofacial arteriovenous metameric syndrome type 1
53719	Cerebrofacial arteriovenous metameric syndrome type 2
141199	Cerebrofacial arteriovenous metameric syndrome type 3
314679	Cerebro-facio-articular syndrome
1394	Cerebro-facio-thoracic dysplasia
2995	Cerebrofrontofacial syndrome type 3
912	Cerebrohepatorenal syndrome
2406	Cerebromedullospinal disconnection
1458	Cerebro-oculo-dento-auriculo-skeletal syndrome
94084	Cerebro-oculo-facial-lymphatic syndrome
1466	Cerebrooculofacioskeletal syndrome
66625	Cerebro-oculo-nasal syndrome
1396	Cerebro-reno-digital syndrome
313838	Cerebroretinal microangiopathy with calcifications and cysts
3421	Cerebroretinal vasculopathy
909	Cerebrotendinous xanthomatosis
1980	Cerebrovascular ferrocalcinosis
169079	Cernunnos deficiency
169079	Cernunnos XLFD
169079	Cernunnos-XLF deficiency
98989	Cerulean cataract
213772	Cervical adenocarcinoma
213828	Cervical adenoid basal carcinoma
213823	Cervical adenoid cystic carcinoma
213792	Cervical adenosarcoma
99079	Cervical aortic arch
141046	Cervical dermoid cyst
93962	Cervical dystonia
213837	Cervical germ cell cancer
2218	Cervical hypertrichosis - peripheral neuropathy
213807	Cervical leiomyosarcoma
213837	Cervical malignant germ cell tumor
213787	Cervical malignant müllerian mixed tumor
213812	Cervical malignant peripheral neuroectodermal tumor
213817	Cervical papillary carcinoma
213812	Cervical peripheral neuroectodermal cancer
213802	Cervical rhabdomyosarcoma
268392	Cervical spina bifida aperta
268762	Cervical spina bifida cystica
141067	Cervicofacial enchondroma
141067	Cervicofacial fibrochondroma
137923	Cervicofacial lymphatic malformation
3456	Cervico-oculo-acoustic syndrome
268397	Cervicothoracic spina bifida aperta
268766	Cervicothoracic spina bifida cystica
586	CF
1340	CFC syndrome
1520	CFND
1520	CFNS
2020	CFTDM
2388	ChAc
307766	CHAC syndrome
307766	CHACS
3386	Chagas disease
1401	CHAND syndrome
98979	Chandler syndrome
1401	CHANDS
2235	Chang-Davidson-Carlson syndrome
88642	Channelopathy-associated congenital insensitivity to pain
3282	Chaotic atrial tachycardia
319244	Chapare hemorrhagic fever
46627	Char syndrome
803	Charcot disease
90658	Charcot-Marie-Tooth disease - deafness
90103	Charcot-Marie-Tooth disease - deafness - intellectual deficit
93114	Charcot-Marie-Tooth disease - nephropathy
64751	Charcot-Marie-Tooth disease - pyramidal features
65753	Charcot-Marie-Tooth disease type 1
101081	Charcot-Marie-Tooth disease type 1A
101082	Charcot-Marie-Tooth disease type 1B
101083	Charcot-Marie-Tooth disease type 1C
101084	Charcot-Marie-Tooth disease type 1D
90658	Charcot-Marie-Tooth disease type 1E
101085	Charcot-Marie-Tooth disease type 1F
98856	Charcot-Marie-Tooth disease type 2B1
101101	Charcot-Marie-Tooth disease type 2B2
101102	Charcot-Marie-Tooth disease type 2H
64748	Charcot-Marie-Tooth disease type 3
64749	Charcot-Marie-Tooth disease type 4
99948	Charcot-Marie-Tooth disease type 4A
99955	Charcot-Marie-Tooth disease type 4B1
99956	Charcot-Marie-Tooth disease type 4B2
99949	Charcot-Marie-Tooth disease type 4C
99950	Charcot-Marie-Tooth disease type 4D
99951	Charcot-Marie-Tooth disease type 4E
99952	Charcot-Marie-Tooth disease type 4F
99953	Charcot-Marie-Tooth disease type 4G
99954	Charcot-Marie-Tooth disease type 4H
139515	Charcot-Marie-Tooth disease type 4J
90120	Charcot-Marie-Tooth disease type 6
91024	Charcot-Marie-Tooth disease, axonal, autosomal recessive type 4C
1964	Char-Douglas-Dungan syndrome
138	CHARGE association
138	CHARGE syndrome
1496	Charlevoix disease
1406	Charlie M syndrome
168577	CHC type 2
98975	CHED1
293603	CHED2
98975	CHEDI
381	Chediak-Higashi like syndrome
167	Chédiak-Higashi syndrome
293603	CHEDII
1221	Cheilitis glandularis
99647	Cheiro-spondylo-enchondromatosis

ORPHA Number	Disease name
812	Cherry-red spot-myoclonus syndrome
184	Cherubism
3019	Cherubism - gingival fibromatosis - intellectual deficit
268882	Chiari malformation type 1
1136	Chiari malformation type 2
268882	Chiari malformation type I
1136	Chiari malformation type II
324625	Chikungunya
90280	Chilblain lupus
139	CHILD nevus
139	CHILD syndrome
64280	Childhood absence epilepsy
209908	Childhood apraxia of speech
135	Childhood ataxia with diffuse central nervous system hypomyelination
168782	Childhood disintegrative disorder
293955	Childhood encephalopathy due to thiamine pyrophosphokinase deficiency
284324	Childhood-onset autosomal recessive slowly progressive spinocerebellar ataxia
247667	Childhood-onset hypophosphatasia
171439	Childhood-onset nemaline myopathy
247667	Childhood-onset phosphoethanolaminuria
209341	Childhood-onset proximal spinal muscular atrophy, autosomal dominant
247667	Childhood-onset Rathburn disease
101000	Childhood-onset spastic paraparesis - distal muscle wasting
3474	CHIME syndrome
2888	Chitayat-Meunier-Hodgkinson syndrome
3218	Chitty-Hall-Baraitser syndrome
3331	Chitty-Hall-Webb syndrome
757	Chloride shunt syndrome
86850	Chloroma
180	CHM
137914	Choanal atresia
1200	Choanal atresia - deafness - cardiac defects - dysmorphism
70567	Cholangiocarcinoma
69663	Cholelithiasis with ABCB4 gene mutation
173	Cholera
1414	Cholestasis - lymphedema
1415	Cholestasis - pigmentary retinopathy - cleft palate
79303	Cholestasis, with delta(4)-3-oxosteroid 5-beta-reductase deficiency
102069	Cholestatic hepatic amyloidosis
75234	Cholesterol ester storage disease
79506	Cholesterol-ester transfer protein deficiency
75234	Cholesteryl ester storage disease
166272	Chondrodysplasia - dentinogenesis imperfecta - joint laxity
1422	Chondrodysplasia - disorder of sex development
1422	Chondrodysplasia - pseudohermaphrodism
79344	Chondrodysplasia punctata, Sheffield type
79346	Chondrodysplasia punctata, tibial-metacarpal type
79347	Chondrodysplasia punctata, Toriello type
263463	Chondrodysplasia with congenital joint dislocations, CHST3 type
280586	Chondrodysplasia with joint dislocations, gPAPP type
3144	Chondrodysplasia with snail-like pelvis
50945	Chondrodysplasia, Blomstrand type
2098	Chondrodysplasia, Grebe type
35173	Chondrodystrophia calcificans congenita
289	Chondroectodermal dysplasia
319195	Chondroectodermal dysplasia with night blindness
55880	Chondrosarcoma
251674	Chordoid glioma
178	Chordoma
2388	Chorea-acanthocytosis
2388	Choreoacanthocytosis
209905	Choreoathetosis - hypothyroidism - neonatal respiratory distress
252015	Choriocarcinoma of the central nervous system
179	Chorioretinopathy, Birdshot type
91353	Choristoma
251899	Choroid plexus carcinoma
1433	Choroidal atrophy - alopecia
39044	Choroidal melanoma
180	Choroideremia
1435	Choroideremia - deafness - obesity
1434	Choroideremia - hypopituitarism
94087	CHP
1436	Christian syndrome
2621	Christian-Rosenberg syndrome
85278	Christianson syndrome
1808	Christianson-Fourie syndrome
1201	Christmas tree syndrome
181	Christ-Siemens-Touraine syndrome
325004	Chromic atypical neutrophilic dermatosis-lipodystrophy-elevated temperature syndrome
182	Chromoblastomycosis
182	Chromomycosis
319303	Chromophobe renal cell adenocarcinoma
319303	Chromophobe renal cell carcinoma
3380	Chromosome 18 duplication
195	Chromosome 22 inversion/duplication
330064	Chronic actinic dermatitis
314928	Chronic adult hydrocephalus
99871	Chronic and localized Langerhans cell histiocytosis
99873	Chronic and multifocal Langerhans cell histiocytosis
137817	Chronic arachnoiditis
71279	Chronic ataxic neuropathy - ophthalmoplegia - IgM paraprotein - cold agglutinins - disialosyl antibodies
2137	Chronic autoimmune hepatitis
133	Chronic berylliosis
133	Chronic beryllium disease
133	Chronic beryllium lung disease
56425	Chronic cold agglutinin disease
79078	Chronic dacryoadenitis and sialoadenitis
103907	Chronic diarrhea due to glucoamylase deficiency
314373	Chronic diarrhea due to guanylate cyclase 2C overactivity
1670	Chronic diarrhea with villous atrophy
168940	Chronic eosinophilic leukemia
2902	Chronic eosinophilic pneumonia
99921	Chronic graft versus host disease
521	Chronic granulocytic leukemia
379	Chronic granulomatous disease
95161	Chronic hepatic porphyria
396	Chronic hiccough
396	Chronic hiccup
1451	Chronic infantile neurological cutaneous articular syndrome
83418	Chronic infantile spinal muscular atrophy
2932	Chronic inflammatory demyelinating polyneuropathy
2932	Chronic inflammatory demyelinating polyradiculoneuropathy
294422	Chronic intestinal failure
2978	Chronic intestinal pseudo-obstruction
284448	Chronic lymphocytic inflammation with pontine perivascular enhancement responsive to steroids
1334	Chronic mucocutaneous candidiasis
1334	Chronic mucocutaneous candidosis
521	Chronic myelogenous leukemia
521	Chronic myeloid leukemia
98823	Chronic myelomonocytic leukemia
77261	Chronic neuronopathic Gaucher disease
86829	Chronic neutrophilic leukemia
95426	Chronic pain requiring intraspinal analgesia
91359	Chronic pneumonitis of infancy
97557	Chronic proteinuria with focal and segmental hyalinosis
324964	Chronic recurrent multifocal osteomyelitis
77297	Chronic recurrent multifocal osteomyelitis - congenital dyserythropoietic anemia - neutrophilic dermatosis

ORPHA Number	Disease name
217566	Chronic respiratory distress with surfactant metabolism deficiency
71279	Chronic sensory ataxic neuropathy with anti-dyalosyl IgM antibodies
379	Chronic septic granulomatosis
83418	Chronic spinal muscular atrophy
70591	Chronic thromboembolic pulmonary hypertension
97353	Chronic traumatic encephalopathy
37748	Chronic urticaria with gammapathy
37748	Chronic urticaria with macroglobulinemia
2953	CHST14-related EDS
2953	CHST14-related Ehlers-Danlos syndrome
263463	CHST3-related skeletal dysplasia
93971	Chudley-Lowry syndrome
93971	Chudley-Lowry-Hoar syndrome
314597	Chudley-McCullough syndrome
3068	Chudley-Rozdilsky syndrome
183	Churg-Strauss syndrome
238557	Chuvash erythrocytosis
238557	Chuvash polycythemia
71	Chylomicron retention disease
1160	Chylous ascites
46486	Cicatricial pemphigoid
217390	CID due to DOCK8 deficiency
317473	CID due to ikaros deficiency
317476	CID due to MAGT1 deficiency
317428	CID due to ORAI1 deficiency
157949	CID due to RAG 1/2 deficiency
317430	CID due to STIM1 deficiency
314689	CID due to STK4 deficiency
231154	CID T+ B+ due to partial RAG1 deficiency
231154	CID with expansion of gamma delta T cells
2932	CIDP
79394	CIE
294422	CIF
1223	Ciliary dysentery
2114	Cilliers-Beighton syndrome
1451	CINCA syndrome
93365	CINCA syndrome with CIAS1 mutations
93367	CINCA syndrome without CIAS1 mutations
2978	CIPO
1114	Circumscribed cutaneous aplasia of the vertex
217410	Circumscribed lymphangioma
217410	Circumscribed lymphatic malformation
69744	Circumscribed palmoplantar hypokeratosis
309854	Cirrhosis-dystonia-polycythemia-hypermanganesemia syndrome
57777	Cirrhotic cardiomyopathy
157820	CISS
187	Citrullinemia
247525	Citrullinemia type 1
247585	Citrullinemia type 2
247525	Citrullinemia type I
247585	Citrullinemia type II
251383	CK syndrome
90790	CLAH
97249	CLAM
168984	CLAPO syndrome
188	Clarkson disease
315306	Classic 21-OHD CAH, salt wasting form
315311	Classic 21-OHD CAH, simple virilizing form
85138	Classic Addison's disease
329977	Classic appendiceal endocrine tumor
329977	Classic appendix endocrine tumor
93605	Classic Bartter syndrome
268145	Classic BCKD deficiency
268145	Classic branched-chain ketoacid dehydrogenase deficiency
268145	Classic branched-chain ketoaciduria
247525	Classic citrullinemia
247546	Classic citrullinemia type 1
247546	Classic citrullinemia type I
325524	Classic CLAH
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form
315311	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form
325524	Classic congenital lipoid adrenal hyperplasia due to STAR deficency
329977	Classic endocrine tumor of the appendix
93930	Classic exstrophy of the bladder
79239	Classic galactosemia
98962	Classic GCD
289857	Classic glycine encephalopathy
98962	Classic granular corneal dystrophy
475	Classic Joubert syndrome
313	Classic lamellar ichthyosis
98964	Classic lattice corneal dystrophy
268145	Classic leucinosis
268145	Classic maple syrup urine disease
158796	Classic mast cell leukemia
247775	Classic Mayer-Rokitansky-Küster-Hauser syndrome
251867	Classic medulloblastoma
324604	Classic MmD
247775	Classic MRKH syndrome
268145	Classic MSUD
324604	Classic multiminicore disease
324604	Classic multiminicore myopathy
216866	Classic pantothenate kinase associated neurodegeneration
163898	Classic paraneoplastic limbic encephalitis
163898	Classic paraneoplastic limbic encephalitis, with or without intracellular antigens
93258	Classic Pfeiffer syndrome
79254	Classic phenylketonuria
79254	Classic PKU
280219	classic PMD
247775	Classic Rokitansky syndrome
99864	Classic seminoma
391	Classical Hodgkin disease
391	Classical Hodgkin's disease
391	Classical Hodgkin's lymphoma
394	Classical homocystinuria
2584	Classical mycosis fungoides
79254	Classical phenylketonuria
79254	Classical PKU
240071	Classical progressive supranuclear palsy
240071	Classical PSP
3467	Classical xanthinuria
2272	Clayton Smith-Donnai syndrome
319276	Clear cell adenocarcinoma
319276	Clear cell renal carcinoma
319276	Clear cell renal cell adenocarcinoma
319276	Clear cell renal cell carcinoma
97338	Clear cell sarcoma of soft tissue
97338	Clear cell sarcoma of the tendons and aponeuroses
3429	Cleft - limb-heart malformation syndrome
101023	Cleft hard palate
1995	Cleft lip - retinopathy
888	Cleft lip and/or palate with mucous cysts of lower lip
2319	Cleft lip/palate - abnormal thumbs - microcephaly
2003	Cleft lip/palate - deafness - sacral lipoma
2328	Cleft lip/palate - facial, eye, heart and intestinal anomalies
2001	Cleft lip/palate - intestinal malrotation - cardiopathy
95465	Cleft mitral valve
141242	Cleft nose
2014	Cleft palate
2008	Cleft palate - cardiac defect - genital anomalies - ectrodactyly
921	Cleft palate - coloboma - deafness
2013	Cleft palate - large ears - small head
2015	Cleft palate - short stature - vertebral anomalies
2010	Cleft palate - stapes fixation - oligodontia
2016	Cleft palate-lateral synechia syndrome
2017	Cleft sternum

ORPHA Number	Disease name
99772	Cleft velum
99772	Cleft velum palatinum
1997	Clefting - ectropion - conical teeth
1453	Cleido rhizomelic syndrome
1452	Cleidocranial dysostosis
1452	Cleidocranial dysplasia
3472	Cleidocranial dysplasia - micrognathia - absent thumbs
97297	C-like syndrome
2542	Clinical anophthalmia
284448	CLIPPERS
228329	CLN1 disease
228349	CLN2 disease
228346	CLN3 disease
228340	CLN4A disease
228343	CLN4B disease
228360	CLN5 disease
228363	CLN6 disease
228366	CLN7 disease
228354	CLN8 disease
1947	CLN8 disease, Northern epilepsy variant
228357	CLN9 disease
228337	CLN10 disease
314629	CLN11 disease
314632	CLN12 disease
352709	CLN13 disease
93929	Cloacal exstrophy
314950	Clonal hypereosinophilic syndrome
221083	Clonic hemifacial spasm
268366	Closed iniencephaly
189	Clouston syndrome
140944	CLOVE syndrome
100978	Cloverleaf skull - asphyxiating thoracic dysplasia
93274	Cloverleaf skull - micromelic bone dysplasia
93267	Cloverleaf skull - multiple congenital anomalies
3253	CLPED1
192	CLS
85136	CLWM
289504	CMAMMA
137667	CM-AVM
1334	CMC
1522	CMD
252202	CMMR-D syndrome
99763	CMO I
99763	CMO II
86830	CMPD-U
238459	CMP-sialic acid transporter deficiency
71	CMRD
324611	CMT due to KIF5A mutation

ORPHA Number	Disease name
65753	CMT1
101081	CMT1A
101082	CMT1B
101083	CMT1C
101084	CMT1D
90658	CMT1E
101085	CMT1F
101075	CMT1X
64746	CMT2
99946	CMT2A1
99947	CMT2A2
99936	CMT2B
99937	CMT2C
99938	CMT2D
99939	CMT2E
99940	CMT2F
99941	CMT2G
99942	CMT2I
99943	CMT2J
99944	CMT2K
99945	CMT2L
228179	CMT2M
228174	CMT2N
284232	CMT2O
300319	CMT2P
329258	CMT2Q
101076	CMT2X
101077	CMT3X
64749	CMT4
99948	CMT4A
99955	CMT4B1
99956	CMT4B2
99949	CMT4C
99950	CMT4D
99951	CMT4E
99952	CMT4F
99953	CMT4G
99954	CMT4H
139515	CMT4J
101078	CMT4X
99014	CMT5X
90120	CMT6
352675	CMT6X
1556	CMTC
90114	CMTDI
100043	CMTDIA
100044	CMTDIB
100045	CMTDIC
100046	CMTDID
93114	CMTDIE
352670	CMTDIF

ORPHA Number	Disease name
64747	CMTX
101075	CMTX1
101076	CMTX2
101077	CMTX3
101078	CMTX4
99014	CMTX5
352675	CMTX6
294	CMV antenatal infection
137698	CMV disease in patients with impaired cell mediated immunity deemed at risk
319160	CNM4
1454	COACH syndrome
1456	Coarctation of the abdominal aorta
190	Coats disease
313838	Coats plus syndrome
53721	Cobb syndrome
352704	Cobblestone lissencephaly type B
352682	Cobblestone lissencephaly without muscular or eye involvement
352682	Cobblestone lissencephaly without muscular or ocular involvement
1911	Cocaine embryofetopathy
90068	Cocaine poisoning
228123	Coccidioides infection
228123	Coccidioidomycosis
3233	Cochleosaccular degeneration - cataract
191	Cockayne syndrome
90321	Cockayne syndrome type 1
90322	Cockayne syndrome type 2
90324	Cockayne syndrome type 3
1458	CODAS syndrome
35656	Coenzyme Q 10 deficiency
192	Coffin-Lowry syndrome
1465	Coffin-Siris syndrome
1466	COFS syndrome
263508	COG1-CDG syndrome
263501	COG4-CDG syndrome
263487	COG5-CDG syndrome
79333	COG7-CDG syndrome
95428	COG8-CDG syndrome
1467	Cogan syndrome
98980	Cogan-Reese syndrome
193	Cohen syndrome
2969	Cohen-Hayden syndrome
79144	COIF
79144	COIF syndrome
306686	CO-induced parkinsonism
31824	Colchicine poisoning
56425	Cold agglutinin disease
56425	Cold agglutinin syndrome
157820	Cold-induced sweating syndrome
324561	Cole disease

ORPHA Number	Disease name
2050	Cole-Carpenter syndrome
84087	Collagen type III glomerulopathy
36205	Collagenous colitis
247203	Collecting duct carcinoma
2412	Collins-Pope syndrome
3474	Coloboma - congenital heart disease - ichthyosiform dermatosis-intellectual deficit - ear anomalies syndrome
168	Coloboma - hair abnormality
138	Coloboma - heart defects - atresia choanae - retardation of growth and development - genitourinary problems - ear abnormalities
98942	Coloboma of choroid and retina
98943	Coloboma of eye lens
98946	Coloboma of eyelid
98944	Coloboma of iris
98945	Coloboma of macula
1471	Coloboma of macula - brachydactyly type B
1475	Coloboma of optic nerve with renal disease
98947	Coloboma of optic papilla
98938	Colobomatous microphthalmia
100080	Colon endocrine tumor
1198	Colonic atresia
16	Color blindness, blue monocone monochromatic type
83595	Colorado tick encephalitis
83595	Colorado tick fever
83595	Colorado tick-borne disease
733	Colorectal adenomatous polyposis
261584	Colorectal adenomatous polyposis due to monosomy 5q22.2
356978	Combined D-2-hydroxyglutaric acidemia and L-2-hydroxyglutaric acidemia
356978	Combined D-2-hydroxyglutaric aciduria and L-2-hydroxyglutaric aciduria
26	Combined defect in adenosylcobalamin and methylcobalamin synthesis
79282	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cblC
79283	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cblD
79284	Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cblF
35909	Combined deficiency of factor V and factor VIII
99732	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase
308386	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase type A
308393	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase type B
308400	Combined deficiency of sulfite oxidase, xanthine dehydrogenase and aldehyde oxidase type C
221078	Combined hyperactive dysfunction syndrome of the cranial nerves
169079	Combined immunodeficiency - microcephaly - growth retardation - sensitivity to ionizing radiation
169082	Combined immunodeficiency due to CD3gamma deficiency
169090	Combined immunodeficiency due to CRAC channel dysfunction
217390	Combined immunodeficiency due to dedicator of cytokinesis 8 protein deficiency
217390	Combined immunodeficiency due to DOCK8 deficiency
317473	Combined immunodeficiency due to ikaros deficiency
317476	Combined immunodeficiency due to MAGT1 deficiency
317428	Combined immunodeficiency due to ORAI1 deficiency
157949	Combined immunodeficiency due to RAG 1/2 deficiency
317430	Combined immunodeficiency due to STIM1 deficiency
314689	Combined immunodeficiency due to STK4 deficiency
911	Combined immunodeficiency due to ZAP70 deficiency
231154	Combined immunodeficiency T+ B+ due to partial RAG1 deficiency
221139	Combined immunodeficiency with facio-oculo-skeletal anomalies
39041	Combined immunodeficiency with hypereosinophilia
157949	Combined immunodeficiency with skin granulomas
228423	Combined immunodeficiency with susceptibility to mycobacterial, viral and fungal infections
1979	Combined insulin, insulin-like growth factor 1 (IGF1) and epidermal growth factor (EGF) deficiency
289504	Combined malonic and methylmalonic acidemia
289504	Combined malonic and methylmalonic aciduria
254920	Combined oxidative phosphorylation defect type 2
254925	Combined oxidative phosphorylation defect type 4
137908	Combined oxidative phosphorylation defect type 5
254930	Combined oxidative phosphorylation defect type 7
319504	Combined oxidative phosphorylation defect type 8
319509	Combined oxidative phosphorylation defect type 9
314637	Combined OXPHOS defect type 10
314637	Combined oxidative phosphorylation defect type 10
324535	Combined oxidative phosphorylation defect type 11
319514	Combined oxidative phosphorylation defect type 13
319519	Combined oxidative phosphorylation defect type 14
319524	Combined oxidative phosphorylation defect type 15
352563	Combined oxidative phosphorylation defect type 16
314637	Combined OXPHOS deficiency type 10
309111	Combined pancreatic lipase-colipase deficiency
95494	Combined pituitary hormone deficiencies, genetic forms
139406	Combined prosaposin deficiency
300564	Combined pulmonary fibrosis-emphysema syndrome
166286	Comedo nevus of the palm
634	Comèl-Netherton syndrome
141276	Commissural facial cleft
141061	Commissural lip fistula
3384	Common aortico-pulmonary trunk
3384	Common arterial trunk
1329	Common atrioventricular canal
98864	Common hereditary elliptocytosis
620	Common mesentery
1572	Common variable immunodeficiency
77303	Common variable immunodeficiency due to an intrinsic B cell defect
99831	Common variable immunodeficiency due to an intrinsic T cell defect
231205	Common variable immunodeficiency without known genetic defect
280821	Communicating congenital bronchopulmonary-foregut malformation
280133	Complement component 3 deficiency
99429	Complete androgen insensitivity syndrome
99429	Complete androgen resistance syndrome
1329	Complete atrioventricular canal
99068	Complete atrioventricular canal - Fallot tetralogy
99066	Complete atrioventricular canal - left heart obstruction
99067	Complete atrioventricular canal - ventricle hypoplasia
99066	Complete atrioventricular canal type A
99067	Complete atrioventricular canal type B
99068	Complete atrioventricular canal type C
1329	Complete atrioventricular septal defect
98949	Complete cryptophthalmia
289916	Complete deficiency of methylmalonyl-CoA mutase

ORPHA Number	Disease name
633	Complete growth hormone insensitivity
254688	Complete hydatidiform mole
79293	Complete LCAT deficiency
29	Complete mevalonate kinase deficiency
254688	Complete molar pregnancy
49382	Complete or incomplete color blindness
101063	Complete situs inversus
101063	Complete situs inversus viscerum
180074	Complete unilateral aplasia of the Müllerian duct
83452	Complex regional pain syndrome
99995	Complex regional pain syndrome type 1
99994	Complex regional pain syndrome type 2
98888	Complex X-linked HSP
98888	Complex X-linked SPG
98888	Complicated X-linked HSP
98888	Complicated X-linked SPG
268316	Complication in hemodialysis
168966	Composite Hodgkin and non-Hodgkin lymphoma
168966	Composite lymphoma
228165	Concentric demyelination
3216	Conductive deafness - malformed external ear
3236	Conductive deafness - ptosis - skeletal anomalies
383	Conductive deafness with stapes fixation
1871	Cone dystrophy
209932	Cone dystrophy with supernormal rod electroretinogram
209932	Cone dystrophy with supernormal rod ERG
209932	Cone dystrophy with supernormal rod response
209932	Cone dystrophy with supernormal scotopic electroretinogram
1872	Cone rod dystrophy
1873	Cone rod dystrophy - amelogenesis imperfecta
221142	Confetti-like macular atrophy
3091	Congenital abnormal systemic venous return
294979	Congenital absence of both forearm and hand
295095	Congenital absence of both forearm and hand, bilateral
295093	Congenital absence of both forearm and hand, unilateral
294981	Congenital absence of both lower leg and foot
295099	Congenital absence of both lower leg and foot, bilateral
295097	Congenital absence of both lower leg and foot, unilateral
86815	Congenital absence of lacrimal puncta and salivary glands
217399	Congenital absence of pain with hyperhidrosis
982	Congenital absence of the pulmonary valve
294977	Congenital absence of thigh and lower leg with foot present
295091	Congenital absence of thigh and lower leg with foot present, bilateral
295089	Congenital absence of thigh and lower leg with foot present, unilateral
294975	Congenital absence of upper arm and forearm with hand present
295087	Congenital absence of upper arm and forearm with hand present, bilateral
295085	Congenital absence of upper arm and forearm with hand present, unilateral
96269	Congenital absence of vagina
294990	Congenital absence/hypoplasia of fingers excluding thumb
295114	Congenital absence/hypoplasia of fingers excluding thumb, bilateral
973	Congenital absence/hypoplasia of fingers excluding thumb, unilateral
294988	Congenital absence/hypoplasia of thumb
295112	Congenital absence/hypoplasia of thumb, bilateral
295110	Congenital absence/hypoplasia of thumb, unilateral
324353	Congenital achiasma
418	Congenital adrenal hyperplasia
90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency
90793	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency
90791	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency
95699	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency
95699	Congenital adrenal hyperplasia due to cytochrome POR deficiency
95701	Congenital adrenal hypoplasia of maternal cause
33355	Congenital aleukocytosis
79	Congenital alpha2 antiplasmin deficiency
210122	Congenital alveolar capillary dysplasia
3319	Congenital amegakaryocytic thrombocytopenia
86816	Congenital analbuminemia
217399	Congenital analgesia with hyperhidrosis
95507	Congenital anomaly of hepatic vein
95498	Congenital anomaly of superior caval vein
95498	Congenital anomaly of superior vena cava
95500	Congenital anomaly of the coronary sinus
95499	Congenital anomaly of the inferior caval vein
95499	Congenital anomaly of the inferior vena cava
95499	Congenital anomaly of the IVC
95498	Congenital anomaly of the SVC
79143	Congenital anonychia
91489	Congenital anterior megalophthalmia
95449	Congenital aortic valve insufficiency
3093	Congenital aortic valve stenosis
93322	Congenital aplasia and dysplasia of the tibia with intact fibula
353334	Congenital arteriovenous anastomoses of the retina
353334	Congenital arteriovenous communication of the retina
1195	Congenital atransferrinemia
60041	Congenital atrioventricular block
162526	Congenital auditory ossicle malformation without external ear abnormality
1216	Congenital benign spinal muscular atrophy with contractures
48	Congenital bilateral absence of vas deferens
48	Congenital bilateral agenesis of vas deferens
48	Congenital bilateral aplasia of vas deferens
93177	Congenital bilateral megacalycosis
79301	Congenital bile acid synthesis defect type 1
79303	Congenital bile acid synthesis defect type 2
79302	Congenital bile acid synthesis defect type 3
79095	Congenital bile acid synthesis defect type 4
300337	Congenital blindness due to retinal non-attachment
2292	Congenital bowing of long bones
71278	Congenital brain dysgenesis due to glutamine synthetase deficiency
2040	Congenital bronchobiliary fistula
3161	Congenital bronchopulmonary sequestration
1369	Congenital cataract - hypertrophic cardiomyopathy - mitochondrial myopathy
1376	Congenital cataract - ichthyosis
2543	Congenital cataract - microphthalmia
330054	Congenital cataract - progressive muscular hypotonia - deafness - developmental delay
330054	Congenital cataract - progressive muscular hypotonia - hearing loss - developmental delay
289499	Congenital cataract microcornea with corneal opacity
98983	Congenital cataract, Volkmann type
300313	Congenital cataract-deafness-severe developmental delay syndrome

ORPHA Number	Disease name
300313	Congenital cataract-hearing loss-severe developmental delay syndrome
48431	Congenital cataracts - facial dysmorphism - neuropathy
99803	Congenital central alveolar hypoventilation - Hirschsprung disease
661	Congenital central alveolar hypoventilation syndrome
95501	Congenital central diabetes insipidus
2345	Congenital cervical vertebral fusion
53689	Congenital chloride diarrhea
329242	Congenital chronic diarrhea with exudative enteropathy
329242	Congenital chronic diarrhea with protein-losing enteropathy
264688	Congenital chylothorax
2505	Congenital circumferential skin folds
91413	Congenital Claude-Bernard-Horner syndrome
467	Congenital combined pituitary hormone deficiency
269505	Congenital communicating hydrocephalus
99129	Congenital complete agenesis of pericardium
115	Congenital contractural arachnodactyly
178382	Congenital convex foot
178382	Congenital convex pes valgus
53691	Congenital cornea plana
95491	Congenital coronary artery aneurysm
168612	Congenital deficiency in alpha-fetoprotein
859	Congenital deficiency of transcobalamin
2140	Congenital diaphragmatic hernia
79324	Congenital disorder of glycosylation 1g
79325	Congenital disorder of glycosylation 1h
79326	Congenital disorder of glycosylation 1i
324737	Congenital disorder of glycosylation due to steroid 5alpha-reductase type 3 deficiency
79324	Congenital disorder of glycosylation Ig
79325	Congenital disorder of glycosylation Ih
79326	Congenital disorder of glycosylation Ii
79318	Congenital disorder of glycosylation type 1a
79319	Congenital disorder of glycosylation type 1b
79320	Congenital disorder of glycosylation type 1c
79321	Congenital disorder of glycosylation type 1d
79322	Congenital disorder of glycosylation type 1e
79323	Congenital disorder of glycosylation type 1f
79327	Congenital disorder of glycosylation type 1k
79328	Congenital disorder of glycosylation type 1L
91131	Congenital disorder of glycosylation type 1m
244310	Congenital disorder of glycosylation type 1n
280071	Congenital disorder of glycosylation type 1p
300536	Congenital disorder of glycosylation type 1r
324422	Congenital disorder of glycosylation type 1s
79329	Congenital disorder of glycosylation type 2a
79330	Congenital disorder of glycosylation type 2b
79332	Congenital disorder of glycosylation type 2d
79333	Congenital disorder of glycosylation type 2e
238459	Congenital disorder of glycosylation type 2f
95428	Congenital disorder of glycosylation type 2h
86309	Congenital disorder of glycosylation type 2j
356961	Congenital disorder of glycosylation type 2m
79318	Congenital disorder of glycosylation type Ia
79319	Congenital disorder of glycosylation type Ib
79320	Congenital disorder of glycosylation type Ic
79321	Congenital disorder of glycosylation type Id
79322	Congenital disorder of glycosylation type Ie
79323	Congenital disorder of glycosylation type If
79329	Congenital disorder of glycosylation type IIa
79330	Congenital disorder of glycosylation type IIb
79332	Congenital disorder of glycosylation type IId
79333	Congenital disorder of glycosylation type IIe
238459	Congenital disorder of glycosylation type IIf
263508	Congenital disorder of glycosylation type IIg
95428	Congenital disorder of glycosylation type IIh
263487	Congenital disorder of glycosylation type IIi
263501	Congenital disorder of glycosylation type IIj
356961	Congenital disorder of glycosylation type IIm
86309	Congenital disorder of glycosylation type Ij
79327	Congenital disorder of glycosylation type Ik
79328	Congenital disorder of glycosylation type IL
91131	Congenital disorder of glycosylation type Im
244310	Congenital disorder of glycosylation type In
263494	Congenital disorder of glycosylation type Io
280071	Congenital disorder of glycosylation type Ip
300536	Congenital disorder of glycosylation type Ir
324422	Congenital disorder of glycosylation type Is
329178	Congenital disorder of glycosylation type Iu
85	Congenital dyserythropoietic anemia
293825	Congenital dyserythropoietic anemia due to KLF1 mutation
98869	Congenital dyserythropoietic anemia type 1
98873	Congenital dyserythropoietic anemia type 2
98870	Congenital dyserythropoietic anemia type 3
67044	Congenital dyserythropoietic anemia with thrombocytopenia, X-linked
91491	Congenital ectropion uveae
295032	Congenital elbow dislocation
295227	Congenital elbow dislocation, bilateral
295225	Congenital elbow dislocation, unilateral
103910	Congenital enterocyte heparan sulfate deficiency
168601	Congenital enterokinase deficiency
168601	Congenital enteropathy due to enteropeptidase deficiency
292	Congenital enterovirus infection
70596	Congenital Epstein-Barr virus infection
157826	Congenital epulis
231573	Congenital erosive and vesicular dermatosis
90042	Congenital erythrocytosis due to erythropoietin receptor mutation
79277	Congenital erythropoietic porphyria
91358	Congenital esophageal diverticulum
215	Congenital essential nyctalopia
91	Congenital estrogen deficiency
280811	Congenital extrapulmonary sequestration
99176	Congenital eyelid retraction
570	Congenital facial diplegia
325	Congenital factor II deficiency
326	Congenital factor V deficiency
327	Congenital factor VII deficiency
328	Congenital factor X deficiency

ORPHA Number	Disease name
329	Congenital factor XI deficiency
330	Congenital factor XII deficiency
331	Congenital factor XIII deficiency
92050	Congenital familial intractable diarrhea with epithelial or epithelium abnormalities
2020	Congenital fiber-type disproportion myopathy
335	Congenital fibrinogen deficiency
45358	Congenital fibrosis of extraocular muscles
90045	Congenital folate malabsorption
2345	Congenital fused cervical segments
1023	Congenital generalized hypertrichosis, Ambras type
79495	Congenital generalized hypertrichosis, Macias-Flores type
295232	Congenital genu flexum
295229	Congenital genu recurvatum
157826	Congenital gingival cell tumor
98976	Congenital glaucoma
157826	Congenital granular cell tumor
330	Congenital Hageman factor deficiency
60041	Congenital heart block
139	Congenital hemidysplasia with ichthyosiform nevus and limbs defects
238691	Congenital hepatic hemangioma
98975	Congenital hereditary endothelial dystrophy type 1
293603	Congenital hereditary endothelial dystrophy type 2
98975	Congenital hereditary endothelial dystrophy type I
293603	Congenital hereditary endothelial dystrophy type II
306530	Congenital hereditary facial palsy with variable deafness
306530	Congenital hereditary facial palsy with variable hearing loss
306530	Congenital hereditary facial paralysis with variable deafness
306530	Congenital hereditary facial paralysis with variable hearing loss
101068	Congenital hereditary stromal dystrophy
293	Congenital herpes virus infection
483	Congenital high-molecular-weight kininogen deficiency
91413	Congenital Horner syndrome
2185	Congenital hydrocephalus
268874	Congenital hydromyelia
2190	Congenital hydronephrosis
478	Congenital hypogonadotropic hypogonadism with anosmia
467	Congenital hypopituitarism
124	Congenital hypoplastic anemia, Blackfan-Diamond type
442	Congenital hypothyroidism
226313	Congenital hypothyroidism due to maternal intake of antithyroid drugs
95715	Congenital hypothyroidism due to transplacental passage of maternal TSH-binding inhibitory antibodies
1195	Congenital hypotransferrinemia
79458	Congenital hypotrichosis - milia
352333	Congenital ichthyosis - intellectual deficit - spastic quadriplegia
352333	Congenital ichthyosis - intellectual deficit - spastic tetraplegia
2271	Congenital ichthyosis - microcephalus - quadriplegia
2271	Congenital ichthyosis - microcephalus - tetraplegia
88621	Congenital ichthyosis type 4
631	Congenital IGHD
231662	Congenital IGHD type IA
231671	Congenital IGHD type IB
231679	Congenital IGHD type II
231692	Congenital IGHD type III
217399	Congenital indifference to pain with hyperhidrosis
64752	Congenital insensitivity to pain and thermal analgesia
217399	Congenital insensitivity to pain with hyperhidrosis
388	Congenital intestinal aganglionosis
280802	Congenital intrapulmonary sequestration
1229	Congenital intrauterine infection-like syndrome
332	Congenital intrinsic factor deficiency
199296	Congenital isolated ACTH deficiency
631	Congenital isolated GH deficiency
231662	Congenital isolated GH deficiency type IA
231671	Congenital isolated GH deficiency type IB
231679	Congenital isolated GH deficiency type II
231692	Congenital isolated GH deficiency type III
631	Congenital isolated growth hormone deficiency
231662	Congenital isolated growth hormone deficiency type IA
231671	Congenital isolated growth hormone deficiency type IB
231679	Congenital isolated growth hormone deficiency type II
231692	Congenital isolated growth hormone deficiency type III
209893	Congenital isolated TBG deficiency
209893	Congenital isolated thyroxine-binding globulin deficiency
295034	Congenital knee dislocation
53690	Congenital lactase deficiency
70472	Congenital lactic acidosis, Saguenay-Lac-St. Jean type
313	Congenital lamellar ichthyosis
99872	Congenital Langerhans cell histiocytosis
141124	Congenital laryngeal cyst
137932	Congenital laryngeal palsy
2374	Congenital laryngeal web
2373	Congenital laryngomalacia
1954	Congenital lethal erythroderma
210163	Congenital lethal myopathy, Compton-North type
93937	Congenital limb amputation
90790	Congenital lipoid adrenal hyperplasia due to STAR deficency
811	Congenital lipomatosis of pancreas
140944	Congenital lipomatous overgrowth - vascular malformation - epidermal nevi
238691	Congenital liver hemangioma
1928	Congenital lobar emphysema
768	Congenital long QT syndrome
93323	Congenital longitudinal deficiency of the fibula
93321	Congenital longitudinal deficiency of the radius
93322	Congenital longitudinal deficiency of the tibia
93320	Congenital longitudinal deficiency of the ulna
2430	Congenital macroglossia
95430	Congenital major airway collapse
83620	Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells
141214	Congenital maxillomandibular fusion
93109	Congenital megacalycosis
280671	Congenital megaconial myopathy
2665	Congenital mesoblastic nephroma
566	Congenital microcoria
199293	Congenital microgastria
2290	Congenital microvillous atrophy
99057	Congenital mitral stenosis
98905	Congenital multicore myopathy with external ophthalmoplegia
97242	Congenital muscular dystrophy
1875	Congenital muscular dystrophy - infantile cataract - hypogonadism
329206	Congenital muscular dystrophy - muscle hypertrophy - intellectual deficit due to POMT1
329206	Congenital muscular dystrophy - muscle hypertrophy - severe intellectual deficit
258	Congenital muscular dystrophy due to laminin alpha2 deficiency
157973	Congenital muscular dystrophy due to LMNA mutation
280671	Congenital muscular dystrophy due to phosphatidylcholine biosynthesis defect
258	Congenital muscular dystrophy type 1A
98893	Congenital muscular dystrophy type 1B
52428	Congenital muscular dystrophy type 1C

ORPHA Number	Disease name
98894	Congenital muscular dystrophy type 1D
34520	Congenital muscular dystrophy with integrin deficiency
280671	Congenital muscular dystrophy with mitochondrial structural abnormalities
75840	Congenital muscular dystrophy, Ullrich type
590	Congenital myasthenic syndromes
353327	Congenital myasthenic syndromes with glycosylation defect
97245	Congenital myopathy
168572	Congenital myopathy - cleft palate - malignant hyperthermia
98904	Congenital myopathy with excess of thin filaments
319160	Congenital myopathy with internal nuclei and atypical cores
199329	Congenital myopathy, Paradas type
289380	Congenital myosclerosis, Löwenthal type
831	Congenital narrowing of cervical spinal canal
162521	Congenital nasal pyriform aperture stenosis with holoprosencephaly
141083	Congenital nasolacrimal duct obstruction
141083	Congenital nasolacrimal mucocele
168486	Congenital NCL
839	Congenital nephrotic syndrome, Finnish type
306504	Congenital nephrotic syndrome-interstitial lung disease-epidermolysis bullosa syndrome
168486	Congenital neuronal ceroid lipofuscinosis
79394	Congenital non-bullous ichthyosiform erythroderma
269510	Congenital non-communicating hydrocephalus
269505	Congenital non-obstructive hydrocephalus
1216	Congenital nonprogressive spinal muscular atrophy
208513	Congenital nonprogressive spinocerebellar ataxia
269510	Congenital obstructive hydrocephalus
79144	Congenital onychodysplasia
79144	Congenital onychodysplasia of the index fingers
99012	Congenital or early infantile optic atrophy
2772	Congenital osteogenesis imperfecta - microcephaly - cataracts
465	Congenital PAI-1 deficiency
2805	Congenital pancreatic agenesis
313906	Congenital pancreatic cyst
139414	Congenital panfollicular nevus
264675	Congenital PAP
99130	Congenital partial agenesis of pericardium
99124	Congenital partial pulmonary venous return anomaly
295036	Congenital patella dislocation
295237	Congenital patella dislocation, bilateral
295234	Congenital patella dislocation, unilateral
99072	Congenital patent ductus arteriosus aneurysm
2846	Congenital pericardium anomaly
332	Congenital pernicious anemia
626	Congenital pigmented nevus
465	Congenital plasminogen activator inhibitor type 1 deficiency
2907	Congenital poikiloderma with bullae, Weary type
90042	Congenital polycythemia due to erythropoietin receptor mutation
124	Congenital PRCA
749	Congenital prekallikrein deficiency
83461	Congenital primary aphakia
2416	Congenital primary lymphedema
617	Congenital primary megalo-ureter
617	Congenital primary megaureter
238654	Congenital primary megaureter, nonrefluxing and unobstructed form
238646	Congenital primary megaureter, obstructed form
238650	Congenital primary megaureter, refluxing form
327	Congenital proconvertin deficiency
66630	Congenital pseudoarthrosis of clavicle
295020	Congenital pseudoarthrosis of the femur
295022	Congenital pseudoarthrosis of the fibula
157808	Congenital pseudoarthrosis of the limbs
295024	Congenital pseudoarthrosis of the radius
295018	Congenital pseudoarthrosis of the tibia
295026	Congenital pseudoarthrosis of the ulna
91411	Congenital ptosis
2444	Congenital pulmonary airway malformation of the lung
280827	Congenital pulmonary airway malformation, type 0
280832	Congenital pulmonary airway malformation, type 1
280840	Congenital pulmonary airway malformation, type 2
280847	Congenital pulmonary airway malformation, type 3
280854	Congenital pulmonary airway malformation, type 4
264675	Congenital pulmonary alveolar proteinosis
2414	Congenital pulmonary lymphangiectasia
3161	Congenital pulmonary sequestration
3189	Congenital pulmonary valve stenosis
3188	Congenital pulmonary veins atresia or stenosis
185	Congenital pulmonary venolobar syndrome
3090	Congenital pulmonary venous connection anomaly
3090	Congenital pulmonary venous return anomaly
124	Congenital pure red cell aplasia
295032	Congenital radial head dislocation
97598	Congenital renal artery stenosis
97598	Congenital renovascular hypoplasia
281190	Congenital reticular ichthyosiform erythroderma
353334	Congenital retinal arteriovenous anastomoses
353334	Congenital retinal arteriovenous communication
300337	Congenital retinal detachment
190	Congenital retinal telangiectasia
178382	Congenital rocker-bottom foot
290	Congenital rubella syndrome
974	Congenital scalp defects with distal limb anomalies
974	Congenital scalp defects with distal limb reduction anomalies
238536	Congenital secondary erythrocytosis
238536	Congenital secondary polycythemia
2301	Congenital short bowel syndrome
1987	Congenital short femur
295030	Congenital shoulder dislocation
93400	Congenital sialidosis type 2
263435	Congenital smooth muscle hamartoma
103908	Congenital sodium diarrhea
94068	Congenital spondyloepiphyseal dysplasia
215	Congenital stationary night blindness
75382	Congenital stationary night blindness, Oguchi type
99122	Congenital stenosis of the inferior caval vein
99122	Congenital stenosis of the inferior vena cava
99122	Congenital stenosis of the IVC
3197	Congenital stiff man syndrome
101068	Congenital stromal corneal dystrophy
328	Congenital Stuart factor deficiency
141121	Congenital subglottic stenosis
35122	Congenital sucrase-isomaltase deficiency
306446	Congenital sucrase-isomaltase deficiency with minimal starch tolerance
306474	Congenital sucrase-isomaltase deficiency with starch and lactose intolerance
306436	Congenital sucrase-isomaltase deficiency with starch intolerance
306462	Congenital sucrase-isomaltase deficiency without starch intolerance
306486	Congenital sucrase-isomaltase deficiency without sucrose intolerance
35122	Congenital sucrase-isomaltose malabsorption

ORPHA Number	Disease name
306446	Congenital sucrase-isomaltose malabsorption with minimal starch tolerance
306474	Congenital sucrase-isomaltose malabsorption with starch and lactose intolerance
306436	Congenital sucrase-isomaltose malabsorption with starch intolerance
306462	Congenital sucrase-isomaltose malabsorption without starch intolerance
306486	Congenital sucrase-isomaltose malabsorption without sucrose intolerance
35122	Congenital sucrose intolerance
306446	Congenital sucrose intolerance with minimal starch tolerance
306474	Congenital sucrose intolerance with starch and lactose intolerance
306436	Congenital sucrose intolerance with starch intolerance
306462	Congenital sucrose intolerance without starch intolerance
306486	Congenital sucrose intolerance without sucrose intolerance
3465	Congenital suprabulbar paresis
99059	Congenital supravalvular mitral ring
98948	Congenital symblepharon
141214	Congenital syngnathia
210576	Congenital temporomandibular joint ankylosis
93583	Congenital thrombotic thrombocytopenic purpura due to ADAMTS-13 deficiency
99125	Congenital total pulmonary venous return anomaly
858	Congenital toxoplasmosis
141127	Congenital tracheal stenosis
95430	Congenital tracheomalacia
95459	Congenital tricuspid stenosis
231013	Congenital trigeminal anesthesia
210576	Congenital trismus
88629	Congenital tritanopia
98686	Congenital trochlear nerve palsy
141099	Congenital tubular nose
99060	Congenital unguarded mitral orifice
95457	Congenital unguarded tricuspid orifice
1166	Congenital unilateral hypoplasia of depressor anguli oris
2258	Congenital unilateral pulmonary hypoplasia
1864	Congenital valvular dysplasia
2291	Congenital velopharyngeal incompetence
178382	Congenital vertical talus
295203	Congenital vertical talus, bilateral
295201	Congenital vertical talus, unilateral
137932	Congenital vocal cord paralysis

ORPHA Number	Disease name
216694	Congenitally corrected transposition of the great arteries
216694	Congenitally corrected transposition of the great vessels
2391	Congenitally short costocoracoid ligament
216729	Congenitally uncorrected transposition of the great arteries with cardiac malformation
99042	Congenitally uncorrected transposition of the great arteries with coarctation
216729	Congenitally uncorrected transposition of the great vessels with cardiac malformation
99042	Congenitally uncorrected transposition of the great vessels with coarctation
99827	Congo fever
99827	Congo hemorrhagic fever
97231	Conjunctivitis lignosa
280210	Connatal PMD
300284	Connective tissue disorder due to LH3 deficiency
300284	Connective tissue disorder due to lysyl hydroxylase-3 deficiency
140969	Conorenal syndrome
567	Conotruncal anomaly face syndrome
2445	Conotruncal heart malformations
35173	Conradi-Hünermann-Happle syndrome
293830	Constitutional dyserythropoietic anemia
319651	Constitutional megaloblastic anemia with severe neurologic disease
252202	Constitutional mismatch repair deficiency syndrome
295000	Constriction rings syndrome
1303	Constrictive bronchiolitis
84142	Continuous muscle fiber activity syndrome
725	Continuous spike-wave during slow sleep syndrome
1484	Contractures - ectodermal dysplasia - cleft lip/palate
3454	Contractures of feet-muscle atrophy-oculomotor apraxia
314002	Contractures-webbed neck-micrognathia-hypoplastic nipples syndrome
1487	Cooks syndrome
231214	Cooley anemia
1488	Cooper-Jabs syndrome
2062	Copenhagen syndrome
35656	CoQ10 deficiency
1463	Cor triatriatum
99098	Cor triatriatum dexter
99098	Cor triatriatum dextrum
99099	Cor triatriatum sinister
99099	Cor triatriatum sinistrum
98990	Coralliform cataract
180118	Cordiform uterus

ORPHA Number	Disease name
366	Cori disease
366	Cori-Forbes disease
1051	Corneal anesthesia - deafness - intellectual deficit
1490	Corneal dystrophy - perceptive deafness
1661	Corneal dystrophy epithelial - short stature
98962	Corneal dystrophy Groenouw type I
98969	Corneal dystrophy Groenouw type II
98961	Corneal dystrophy of Bowman layer type I
98960	Corneal dystrophy of Bowman layer type II
1490	Corneal dystrophy with progressive deafness
352662	Corneal intraepithelial dyskeratosis with palmoplantar hyperkeratosis and laryngeal dyskeratosis
171673	Corneal lesions with associated corneal stem cell deficiency due to ocular burns
3177	Corneal-cerebellar syndrome
199	Cornelia de Lange syndrome
96095	Cornelia de Lange-like syndrome
3194	Corneodermatoosseous syndrome
2041	Coronaro-cardiac fistula
2041	Coronary arterial fistulas
2041	Coronary arterial malformations
1081	Coronary artery congenital malformation
94062	Coronary artery disease - hyperlipidemia - hypertension - diabetes - osteoporosis
99085	Coronary artery intramyocardial course
99118	Coronary sinus atresia
99117	Coronary sinus stenosis
3338	Corpus callosum agenesis - blepharophimosis - Robin sequence
1493	Corpus callosum agenesis - cataract - immunodeficiency
1492	Corpus callosum agenesis - double urinary collecting system
1496	Corpus callosum agenesis - neuronopathy
1553	Corpus callosum agenesis - polysyndactyly
50	Corpus callosum agenesis of with chorioretinal abnormality
275543	Corpus callosum hypoplasia-retardation-adducted thumbs-spasticity-hydrocephalus syndrome
2318	CORS
1499	Cortada-Koussef-Matsumoto syndrome
1389	Cortical blindness - intellectual deficit - polydactyly
300570	Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation
163681	Cortical dysplasia - focal epilepsy syndrome
65683	Cortical dysplasia, Taylor type
3152	Cortical hyperostosis - syndactyly
278	Corticobasal degeneration

ORPHA Number	Disease name
199247	Corticosteroid-binding globulin deficiency
54251	Corticosteroid-sensitive aseptic abscesses
99763	Corticosterone methyloxidase deficiency type I
96253	Corticotroph pituitary adenoma
189427	Corticotropin-independent macronodular adrenal hyperplasia
141163	Cosack syndrome
67047	Costeff optic atrophy syndrome
67047	Costeff syndrome
3071	Costello syndrome
1507	Costovertebral segmentation defect - mesomelia
1914	Coumarin embryopathy
93333	Cousin syndrome
1507	COVESDEM syndrome
101078	Cowchock syndrome
201	Cowden disease
201	Cowden syndrome
99932	Cow's milk hypersensitivity
1508	Coxoauricular syndrome
1509	Coxo-podo-patellar syndrome
254920	COXPD2
254925	COXPD4
137908	COXPD5
254930	COXPD7
319504	COXPD8
319509	COXPD9
314637	COXPD10
324535	COXPD11
319514	COXPD13
319519	COXPD14
319524	COXPD15
352563	COXPD16
280827	CPAM type 0
280832	CPAM type 1
280840	CPAM type 2
280847	CPAM type 3
280854	CPAM type 4
475	CPD IV
300564	CPFE
91359	CPI
1416	CPPD
1416	CPPDD
147	CPS1 deficiency
156	CPT1A deficiency
157	CPT2
228302	CPT2, adult-onset form
228305	CPT2, hepatocardiomuscular form
228308	CPT2, lethal systemic form
228302	CPT2, myopathic form
228308	CPT2, neonatal form
228305	CPT2, severe infantile form
157	CPTII
228302	CPTII, adult-onset form
228305	CPTII, hepatocardiomuscular form
228308	CPTII, lethal systemic form
228302	CPTII, myopathic form
228308	CPTII, neonatal form
228305	CPTII, severe infantile form
3286	CPVT
2081	Cramer-Niederdellmann syndrome
202	Crandall syndrome
1512	Crane-Heise syndrome
97339	Cranial dural arteriovenous fistula
97339	Cranial dural arteriovenous malformations
268820	Cranial meningocele
98919	Cranial variant of GBS
98919	Cranial variant of Guillain-Barré syndrome
1339	Cranioacrofacial syndrome
2053	Craniocarpotarsal dysplasia
2053	Craniocarpotarsal dystrophy
7	Craniocerebellocardiac dysplasia
1513	Craniodiaphyseal dysplasia
1514	Craniodigital syndrome - intellectual deficit
1515	Cranioectodermal dysplasia
2099	Craniofacial and osseous defects - intellectual deficit
85168	Craniofacial conodysplasia
1777	Craniofacial dysmorphism - coloboma - corpus callosum agenesis
1789	Craniofacial dysostosis - arthrogryposis - progeroid appearance
1798	Craniofacial dysostosis - diaphyseal hyperplasia
2095	Craniofacial dysostosis - genital, dental, cardiac anomalies
314555	Craniofacial dysplasia-osteopenia syndrome
1516	Craniofacial dyssynostosis
374	Craniofacial microsomia
1529	Craniofacial-deafness-hand syndrome
293843	Craniofacial-ulnar-renal syndrome
1517	Craniofaciocardioskeletal syndrome
1800	Craniofaciocervical osteoglyphic dysplasia
2115	Cranio-facio-digito-genital syndrome
1520	Craniofrontonasal dysplasia
1521	Craniofrontonasal dysplasia - Poland anomaly
228390	Craniofrontonasal dysplasia with alopecia and hypogonadism
1519	Craniofrontonasal dysplasia, Teebi type
1520	Craniofrontonasal syndrome
50814	Craniolenticulosutural dysplasia
85184	Craniometadiaphyseal dysplasia, wormian bone type
1522	Craniometaphyseal dysplasia
1524	Craniomicromelic syndrome
1525	Cranio-osteoarthropathy
54595	Craniopharyngioma
63260	Craniorachischisis
157832	Craniorhiny
1532	Craniosynostosis - alopecia - brain defect
85199	Craniosynostosis - anal anomalies - porokeratosis
1530	Craniosynostosis - cataract
2872	Craniosynostosis - congenital heart disease - intellectual deficit
1538	Craniosynostosis - Dandy-Walker - hydrocephalus
1535	Craniosynostosis - dysmorphism - brachydactyly
1533	Craniosynostosis - fibular aplasia
171839	Craniosynostosis - hydrocephalus - Chiari I malformation - radioulnar synostosis
52054	Craniosynostosis - intracranial calcifications
1540	Craniosynostosis - midfacial hypoplasia - foot abnormalities
1526	Craniosynostosis - synostoses - hypertensive nephropathy
284149	Craniosynostosis and dental anomalies
1541	Craniosynostosis, Boston type
2145	Craniosynostosis, Herrmann-Opitz type
1527	Craniosynostosis, Philadelphia type
1541	Craniosynostosis, Warman type
1534	Craniosynostosis-radial aplasia, Imaizumi type
1528	Craniotelencephalic dysplasia
75373	CRAPB
275543	CRASH syndrome
71	CRD
99854	Cree leukoencephalopathy
504	Creeping myiasis
280569	Crescentic glomerulonephritis
90290	CREST syndrome
204	Creutzfeldt-Jakob disease
281	Cri du chat syndrome
281190	CRIE
205	Crigler-Najjar syndrome
79234	Crigler-Najjar syndrome type 1
79235	Crigler-Najjar syndrome type 2
99827	Crimean hemorrhagic fever
99827	Crimean-Congo hemorrhagic fever
1545	Crisponi syndrome
1461	Criss-cross heart
891	Criswick-Schepens syndrome

ORPHA Number	Disease name
313838	CRMCC
324964	CRMO
1380	Crome syndrome
2930	Cronkhite-Canada syndrome
2719	Cross syndrome
2935	Crossed polydactyly
2935	Crossed polysyndactyly
207	Crouzon craniofacial dysostosis
207	Crouzon disease
93262	Crouzon syndrome - acanthosis nigricans
93262	Crouzono-dermoskeletal syndrome
2905	Crow-Fukase syndrome
52503	CRTR-D
3421	CRV
98910	CRYAB-related myofobrillar myopathy
91139	Cryoglobulinemia type 1
91138	Cryoglobulinemic vascularitis
91138	Cryoglobulinemic vasculitis
1546	Cryptococcosis
163708	Cryptogenic late-onset epileptic spasms
1302	Cryptogenic organizing pneumonia
1547	Cryptomicrotia - brachydactyly - excess fingertip arch
2052	Cryptophthalmos-syndactyly syndrome
1548	Cryptorchidism - arachnodactyly - intellectual deficit
1549	Cryptosporidiosis
357329	Cryptosporidiosis - chronic cholangitis - liver disease
98967	Crystalline stromal dystrophy
101068	CSCD
35122	CSID
306446	CSID with minimal starch tolerance
306474	CSID with starch and lactose intolerance
306436	CSID with starch intolerance
306462	CSID without starch intolerance
306486	CSID without sucrose intolerance
329217	CSVT
725	CSWSS syndrome
70591	CTEPH
247525	CTLN1
247585	CTLN2
909	CTX
3207	Curatolo-Cilio-Pessagno syndrome
98960	Curly fiber corneal dystrophy
1401	Curly hair - ankyloblepharon - nail dysplasia syndrome
307766	Curly hair-acral keratoderma-caries syndrome
1525	Currarino disease
1525	Currarino idiopathic osteoarthropathy
1552	Currarino syndrome
1552	Currarino triad
640	Current pressure-sensitive neuropathy
952	Curry-Hall syndrome
1553	Curry-Jones syndrome
96253	Cushing disease
553	Cushing syndrome
99889	Cushing syndrome due to ectopic ACTH secretion
553	Cushing's syndrome
53721	Cutaneomeningospinal angiomatosis
2451	Cutaneous and mucosal venous malformation
280779	Cutaneous collagenous vasculopathy
329324	Cutaneous hemangioma with muscle or bone atrophy
889	Cutaneous hypersensitivity vasculitis
178475	Cutaneous infectious botulism
504	Cutaneous larva migrans
889	Cutaneous leukocytoclastic angiitis
79455	Cutaneous local mastocytoma
535	Cutaneous lupus erythematosus
79490	Cutaneous lymphangioma circumscriptum
79455	Cutaneous mastocytoma
66646	Cutaneous mastocytosis
90395	Cutaneous mucinosis of infancy
79140	Cutaneous neuroendocrine carcinoma
2881	Cutaneous photosensitivity - lethal colitis
889	Cutaneous small vessel vasculitis
178475	Cutaneous toxin-mediated botulism
1555	Cutis gyrata - acanthosis nigricans - craniosynostosis
209	Cutis laxa
2962	Cutis laxa - corneal clouding - intellectual deficit
228285	Cutis laxa acquisita
90350	Cutis laxa with joint laxity and developmental delay
221145	Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies
171719	Cutis laxa-Marfanoid syndrome
1556	Cutis marmorata telangiectatica congenita
3327	Cutler-Bass-Romshe syndrome
1572	CVID
77303	CVID due to an intrinsic B cell defect
99831	CVID due to an intrinsic T cell defect
306692	Cyanide-induced parkinsonism
2686	Cyclic neutropenia
228379	Cyclosporine-induced folliculodystrofy
210	Cyclosporosis
171886	Cylindrical spirals myopathy
90795	CYP11B1 deficiency
2674	Cyprus facial-neuromusculoskeletal syndrome
212	Cystathionase deficiency
212	Cystathione gamma - lyase deficiency
394	Cystathionine beta-synthase deficiency
212	Cystathioninuria
586	Cystic fibrosis
2575	Cystic fibrosis - gastritis - megaloblastic anemia
2111	Cystic hamartoma of lung and kidney
79486	Cystic hygroma
85136	Cystic leukoencephalopathy without megalencephaly
229	Cystic medial necrosis of aorta
1560	Cysticercosis
213	Cystinosis
214	Cystinuria
214	Cystinuria - lysinuria
93612	Cystinuria type A
93613	Cystinuria type B
75381	Cystoid macular dystrophy
180261	Cystosarcoma phyllode
180261	Cystosarcoma phylloide
95702	Cytomegalic congenital adrenal hypoplasia
294	Cytomegalovirus antenatal infection
137698	Cytomegalovirus disease in patients with impaired cell mediated immunity deemed at risk
94087	Cytophagic histiocytic panniculitis
137678	Czech dysplasia, metatarsal type
2736	Czeizel syndrome
2917	Czeizel-Brooser syndrome
2437	Czeizel-Losonci syndrome
356978	D,L-2-HGA
356978	D,L-2-hydroxyglutaric acidemia
356978	D,L-2-hydroxyglutaric aciduria
90038	D+HUS
79315	D-2-HGA
79315	D-2-hydroxyglutaric acidemia
79315	D-2-hydroxyglutaric aciduria
2953	D4ST1-deficient EDS
2953	D4ST1-deficient Ehlers-Danlos syndrome
1495	Da Silva syndrome
1146	DA1
1146	DA1A
329457	DA5D
251515	DA10
1562	Dacryocystitis - osteopoikilosis
141083	Dacryocystocele
2186	Daentl-Townsend-Siegel syndrome
1563	Dahlberg-Borer-Newcomer syndrome
2181	Daish-Hardman-Lamont syndrome
275523	DALD

ORPHA Number	Disease name
1183	Dancing eye syndrome
1564	Dandy Walker - facial hemangioma
1566	Dandy-Walker malformation - postaxial polydactyly
2091	Daneman-Davy-Mancer syndrome
34587	Danon disease
99645	Dappled diaphyseal dysplasia
218	Darier disease
316	Darier-Gottron disease
218	Darier-White disease
390	Darling disease
3215	Davenport-Donlan syndrome
293978	DAVID syndrome
75565	Davies disease
3046	Davis-Lafer syndrome
2806	Dawson's encephalitis
2143	DBS/FOAR syndrome
79456	DCM
66634	DCMA syndrome
75381	DCMD
1653	DD
231568	DDEB, generalized
231568	DDEB, Pasini and Cockayne-Touraine types
231568	DDEB-gen
99789	DD-I
99791	DD-II
99970	DDLS
79499	DDOD syndrome
52368	DDON syndrome
300536	DDOST-CDG syndrome
2962	De Barsy syndrome
1130	De Die-Smulders-Vles-Fryns syndrome
1598	De Grouchy syndrome
56304	De la Chapelle dysplasia
393	De la Chapelle syndrome
3157	De Morsier syndrome
1570	De Smet-Fabry-Fryns syndrome
33355	De Vaal disease
71277	De Vivo disease
3214	Deaf blind hypopigmentation syndrome, Yemenite type
3217	Deafness - small bowel diverticulosis - neuropathy
2663	Deafness - cataracts - skeletal anomalies
52368	Deafness - dystonia - optic neuronopathy syndrome
3232	Deafness - ear malformation - facial palsy
3220	Deafness - enamel hypoplasia - nail defects
254898	Deafness - encephaloneuropathy - obesity - valvulopathy
3218	Deafness - epiphyseal dysplasia - short stature
3224	Deafness - genital anomalies - metacarpal and metatarsal synostosis
90646	Deafness - hypogonadism
85321	Deafness - intellectual deficit, Martin-Probst type
3226	Deafness - lymphedema - leukemia
2408	Deafness - nephritis - ano-rectal malformation
3230	Deafness - oligodontia
3231	Deafness - onychodystrophy
79500	Deafness - onychodystrophy - osteodystrophy - intellectual deficit
79499	Deafness - onychodystrophy, autosomal dominant
79500	Deafness - onycho-osteodystrophy - intellectual deficit
3229	Deafness - peripheral neuropathy - arterial disease
123	Deafness - pili torti - hypogonadism
3219	Deafness - skeletal dysplasia - lip granuloma
3237	Deafness - symphalangism syndrome, Hermann type
3221	Deafness - thyroid hormone resistance
3239	Deafness - vitiligo - achalasia
3215	Deafness - white hair - contractures - papillomas
90024	Deafness with labyrinthine aplasia, microtia, and microdontia
3241	Deafness-craniofacial syndrome
94064	Deafness-infertility syndrome
303	DEB
158673	DEB, acral
79411	DEB, bullous dermolysis of the newborn
89843	DEB, pruriginosa
158673	DEB-ac
79411	DEB-BDN
158676	DEB-na
89843	DEB-Pr
79410	DEB-Pt
817	Deciduous skin
99970	Dedifferentiated liposarcoma
75496	Defective biosynthesis of proteodermatan sulfate
293978	Deficiency in anterior pituitary function-variable immunodeficiency syndrome
169150	Deficiency of complement of terminal pathway
247353	Deficiency of the IL-36R antagonist
247353	Deficiency of the IL-36Ra
244310	Déficit en Man5GlcNAc2-PP-Dol flippase
679	Degos disease
315	Degos genodermatosis "en cocardes"
1578	Dehydratase deficiency
3202	Dehydrated hereditary stomatocytosis
64748	Dejerine-Sottas syndrome
2318	Dekaban-Arima syndrome
1606	Del(1)(p36)
250989	Del(1)(q21)
250999	Del(1)(q41q42)
238769	Del(1)(q44)
293948	Del(1)p(21.3)
261349	Del(2)(p15p16.1)
163693	Del(2)(p21)
228402	Del(2)(q23.1)
1617	Del(2)(q24)
251014	Del(2)(q31.1)
251019	Del(2)(q32)
251019	Del(2)(q32q33)
251028	Del(2)(q33.1)
1001	Del(2)(q37)
1621	Del(3)(q13)
356947	Del(3)(q26q27)
65286	Del(3)(q29)
238750	Del(4)(q21)
228384	Del(5)(q14.3)
314655	Del(5)(q31.3)
251046	Del(6)(p22)
171829	Del(6)(q16)
251056	Del(6)(q25)
251061	Del(7)(q31)
251066	Del(8)(p11.2)
251071	Del(8)(p23.1)
284160	Del(8)(q21.11)
2496	Del(8)q(13)
324313	Del(9)(p13)
352665	Del(9)(q21)
284169	Del(10)(p11.21p12.31)
276413	Del(10)(q22.3q23.3)
2308	Del(11)(q23.3)
2308	Del(11)(qter)
313884	Del(12)(p12.1)
280325	Del(12)(p13.33)
94063	Del(12)(q14)
289513	Del(12)(q15)(q21.1)
1587	Del(13)(q14)
96168	Del(13)(q34)
261120	Del(14)(q11.2)
261144	Del(14)(q12)
264200	Del(14)(q22q23)
261183	Del(15)(q11.2)
199318	Del(15)(q13.3)
261190	Del(15)(q14)
94065	Del(15)(q24)
261211	Del(16)(p11.2p12.2)
261236	Del(16)(p13.11)

ORPHA Number	Disease name
352629	Del(16)(q24.1)
261250	Del(16)(q24.3)
97685	Del(17)(q11)
261265	Del(17)(q12)
96169	Del(17)(q21.31)
261279	Del(17)(q23.1q23.2)
254346	Del(19)(p13.12)
357001	Del(19)(p13.13)
217346	Del(19)(q13.11)
261295	Del(20)(p12.3)
313781	Del(20)(p13)
261311	Del(20)(q13.33)
261323	Del(21)(q22.11q22.12)
268261	Del(21)(q22.13q22.2)
261476	Del(X)(p21)
1643	Del(X)(p23)
3034	Delayed membranous cranial ossification
3038	Delayed speech - facial asymmetry - strabismus - ear lobe creases
1606	Deletion 1p36
1606	Deletion 1pter
1001	Deletion 2q37
1001	Deletion 2q37-qter
1625	Deletion 4q
281	Deletion 5p
904	Deletion 7q11.23
284160	Deletion 8q21.11
502	Deletion 8q24.1
284169	Deletion 10p11.21p12.31
276413	Deletion 10q22.3q23.3
77	Deletion 11p
893	Deletion 11p13
94063	Deletion 12q14
289513	Deletion 12q15q21.1
1587	Deletion 13q14 syndrome
1590	Deletion 13q32
1600	Deletion 18q
1611	Deletion 20p
1647	Delleman syndrome
1647	Delleman-Oorthuys syndrome
35664	Delta-1-pyrroline 5-carboxylate synthetase deficiency
79101	Delta1-pyrroline-5-carboxylate dehydrogenase deficiency
231237	Delta-beta thalassemia
219	Delta-sarcoglycanopathy
168782	Dementia Infantilis
97353	Dementia pugilistica
283	Demodicidosis
283	Demodicosis
314451	Demons-Meigs syndrome
79134	DEND syndrome
86903	Dendritic cell sarcoma not otherwise specified
99828	Dengue fever
99828	Dengue hemorrhagic fever
99828	Dengue shock syndrome
99828	Dengue virus infection
1651	Dennis-Cohen syndrome
2109	Dennis-Fairhurst-Moore syndrome
93571	Dense deposit disease
1652	Dent disease
93622	Dent disease type 1
93623	Dent disease type 2
1652	Dent syndrome
1077	Dental ankylosis
101	Dentatorubral-pallidoluysian atrophy
101	Dentatorubropallidoluysian atrophy
1653	Dentin dysplasia
99792	Dentin dysplasia - sclerotic bones
314721	Dentin dysplasia type 1 with microdontia and shape anomalies
99789	Dentin dysplasia type I
99791	Dentin dysplasia type II
49042	Dentinogenesis imperfecta
71267	Dentinogenesis imperfecta - short stature - hearing loss - intellectual deficit
166260	Dentinogenesis imperfecta type 2
166265	Dentinogenesis imperfecta type 3
49042	Dentinogenesis imperfecta without osteogenesis imperfecta
166260	Dentinogenesis imperfecta, Shields type 2
166265	Dentinogenesis imperfecta, Shields type 3
77295	Dentoleukoencephalopathy
228423	Dentritic cell, monocyte, B and NK lymphoid deficiency
220	Denys-Drash syndrome
3177	Der Kaloustian-Jarudi-Khoury syndrome
3270	Der Kaloustian-McIntosh-Silver syndrome
96170	Der(22)t(11;22) syndrome
2861	D'Ercole syndrome
36397	Dercum's disease
297	Dermacentor-borne necrosis - erythema - lymphadenopathy
1656	Dermatitis herpetiformis
1266	Dermato-cardio-skeletal syndrome, Borrone type
31112	Dermatofibrosarcoma protuberans
1659	Dermatoleukodystrophy
221	Dermatomyositis
1657	Dermatoosteolysis, Kirghizian type
86920	Dermatopathia pigmentosa reticularis
36426	Dermatostomatitis, Stevens Johnson type
79149	Dermochondrocorneal dystrophy
141051	Dermoid cyst of the face
141046	Dermoid cyst of the neck
303	Dermolytic epidermolysis bullosa
1660	Dermo-odonto dysplasia
99688	Dermotrichic syndrome
1916	DES embryofetopathy
1916	DES syndrome
1425	Desbuquois dysplasia
1425	Desbuquois syndrome
163703	DESC syndrome
228123	Desert fever
228123	Desert rheumatism
98909	Desminopathy
98909	Desmin-related myofibrillar myopathy
84132	Desmin-related myopathy with Mallory body-like inclusions
873	Desmoid disease
251940	Desmoplastic infantile astrocytoma/ ganglioglioma
83469	Desmoplastic small round cell tumor
251863	Desmoplastic/nodular medulloblastoma
35107	Desmosterolosis
98852	Desquamative interstitial pneumonia
158014	Destombes-Rosaï-Dorfman disease
163703	Devastating epileptic encephalopathy in school-aged children
313892	Developmental and speech delay due to SOX5 deficiency
163988	Developmental delay - deafness, Hildebrand type
79134	Developmental delay - epilepsy - neonatal diabetes
2101	Developmental delay - hypotonia - extremities hypertrophy
79157	Developmental delay due to 2-methylbutyryl-CoA dehydrogenase deficiency
289307	Developmental delay due to ALDH6A1 deficiency
289307	Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency
289307	Developmental delay due to MMSDH deficiency
329195	Developmental delay with ASD and gait instability
329195	Developmental delay with autism spectrum disorder and gait instability
79107	Developmental malformations - deafness - dystonia
209908	Developmental verbal dyspraxia
71211	Devic disease
1014	Devriendt-Vandenberghe-Fryns syndrome
403	Dexamethasone sensitive hypertension
1666	Dextrocardia

ORPHA Number	Disease name
99828	DF
383	DFNX2
31112	DFSP
49042	DGI
49042	DGI without OI
166260	DGI-2
93599	D-glycerate dehydrogenase deficiency
941	D-glycerate kinase deficiency
941	D-glyceric acidemia
941	D-glyceric aciduria
373	DGSX
99828	DHF
319651	DHFR deficiency
53739	dHMN
357043	dHMN with upper motor neuron signs
139518	dHMN1
139525	dHMN2
139547	dHMN3 and dHMN4
139536	dHMN5
100998	dHMN5B
98920	dHMN6
139589	dHMN7
139552	dHMNJ
75376	DHRD
2134	D-HUS
93581	D-HUS with anti-factor H antibodies
93578	D-HUS with B factor anomaly
93575	D-HUS with C3 anomaly
357008	D-HUS with DGKE deficiency
93579	D-HUS with H factor anomaly
93580	D-HUS with I factor anomaly
93576	D-HUS with MCP/CD46 anomaly
217023	D-HUS with thrombomodulin anomaly
49042	DI
166260	DI-2
251940	DIA/DIG
3464	Diabetes - hypogonadism - deafness - intellectual deficit
3463	Diabetes insipidus - diabetes mellitus - optic atrophy - deafness
1926	Diabetic embryopathy
85446	Dialysis-related amyloidosis
85446	Dialysis-related arthropathy
275523	Dianzani autoimmune lymphoproliferative disease
66637	Diaphanospondylodysostosis
255182	Diaphorase deficiency
2140	Diaphragmatic agenesia
2141	Diaphragmatic defect - limb deficiency - skull defect
2059	Diaphragmatic hernia - abnormal face - distal limb anomalies
98920	Diaphragmatic spinal muscular atrophy
1802	Diaphyseal dysplasia - anemia
85182	Diaphyseal medullary stenosis - bone malignancy
85182	Diaphyseal medullary stenosis - malignant fibrous histiocytoma
103909	Diarrhea-vomiting due to trehalase deficiency
97282	Diarrheogenic islet cell tumor
1671	Diastematomyelia
628	Diastrophic dwarfism
628	Diastrophic dysplasia
276603	Diazoxide-resistant focal hyperinsulinism due to Kir6.2 deficiency
276598	Diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency
2195	Dicarboxylic aminoaciduria
284343	DICER1 syndrome
180086	Didelphys uterus
3463	DIDMOAD syndrome
1672	Diencephalic syndrome
319192	Diencephalic-mesencephalic junction dysplasia
1916	Diethylstilbestrol embryofetopathy
1916	Diethylstilbestrol syndrome
90060	Diffuse alveolar hemorrhage
324	Diffuse angiokeratoma
251595	Diffuse astrocytoma
79456	Diffuse cutaneous maculopapulous mastocytosis
79456	Diffuse cutaneous mastocytosis
220393	Diffuse cutaneous systemic scleroderma
220393	Diffuse cutaneous systemic sclerosis
73	Diffuse cystic angiomatosis of bone
2199	Diffuse erythrodermic palmoplantar keratoderma, Voerner type
2199	Diffuse erythrodermic palmoplantar keratoderma, Vörner type
702	Diffuse familial brain sclerosis
3165	Diffuse fasciitis with eosinophilia
544	Diffuse large B-cell lymphoma
300849	Diffuse large B-cell lymphoma of the central nervous system
300888	Diffuse large B-cell lymphoma with chronic inflammation
252031	Diffuse leptomeningeal melanocytosis
141209	Diffuse lymphangioma
141209	Diffuse lymphangiomatosis
141209	Diffuse lymphatic malformation
168811	Diffuse malignant peritoneal mesothelioma
2123	Diffuse neonatal hemangiomatosis
86918	Diffuse palmoplantar hyperkeratosis-acrocyanosis syndrome
86918	Diffuse palmoplantar keratoderma-acrocyanosis syndrome
171700	Diffuse panbronchiolitis
71274	Diffuse peritoneal leiomyomatosis
567	DiGeorge sequence
567	DiGeorge syndrome
238	Digestive duplication
141071	Digestive duplication cyst of the tongue
352487	Digital anomalies - intellectual deficit - short stature
1305	Digital anomalies with short palpebral fissures and atresia of esophagus or duodenum
31828	Digitalis poisoning
1146	Digitotalar dysmorphism
294990	Digits 2-5 hypodactyly
295114	Digits 2-5 hypodactyly, biilateral
294990	Digits 2-5 oligodactyly
295114	Digits 2-5 oligodactyly, biilateral
319651	Dihydrofolate reductase deficiency
79244	Dihydrolipoamide acetyltransferase component of pyruvate dehydrogenase complex deficiency
2394	Dihydrolipoamide dehydrogenase deficiency
255182	Dihydrolipoyl dehydrogenase deficiency
79244	Dihydrolipoyllysine-residue acetyltransferase component of pyruvate dehydrogenase complex deficiency
226	Dihydropteridine reductase deficiency
38874	Dihydropyrimidinase deficiency
1675	Dihydropyrimidine dehydrogenase deficiency
38874	Dihydropyrimidinuria
99102	Dilatation of the left appendage
99102	Dilatation of the left auricle
99101	Dilatation of the right atrial appendage
99101	Dilatation of the right atrial auricle
2229	Dilated cardiomyopathy - hypergonadotropic hypogonadism
66634	Dilated cardiomyopathy with ataxia
231111	DILE
243343	Dimethylglycine dehydrogenase deficiency
314002	Dinno syndrome
1493	Dionisi-Vici-Sabetta-Gambarara syndrome
227	Diphallia
1679	Diphtheria
128	Diphyllobothriasis
1681	Diprosopia
1756	Dipygus
210115	DIRA
166291	Dirofilariasis
94064	DIS
35122	Disaccharide intolerance

ORPHA Number	Disease name
306446	Disaccharide intolerance with minimal starch tolerance
306474	Disaccharide intolerance with starch and lactose intolerance
306436	Disaccharide intolerance with starch intolerance
306462	Disaccharide intolerance without starch intolerance
306486	Disaccharide intolerance without sucrose intolerance
90281	Discoid lupus erythematosus
216694	Discordant ventriculoarterial and atrioventricular connections
99052	Discrete fibromuscular subaortic stenosis
99051	Discrete fixed membranous subaortic stenosis
90394	Discrete papular lichen myxedematosus
139420	Disease-associated transverse myelitis
210272	disembarkment syndrome
2412	Dislocation of the hip - dysmorphism
8	Disomy Y
872	Disorder in the hormonal synthesis with or without goiter
2983	Disorder of sex development - intellectual deficit
345	Dissecting cellulitis of the scalp
54251	Disseminated aseptic abscesses
1306	Disseminated dermatofibrosis with osteopoikilosis
141209	Disseminated lymphangioma
141209	Disseminated lymphangiomatosis
141209	Disseminated lymphatic malformation
228264	Disseminated nevus anelasticus
71274	Disseminated peritoneal leiomyomatosis
79152	Disseminated superficial actinic porokeratosis
1620	Distal 3p deletion
1627	Distal 5q deletion
254351	Distal 7q11.23 microdeletion syndrome
261102	Distal 7q11.23 microduplication syndrome
1580	Distal 10p deletion
1590	Distal 13q deletion
1596	Distal 15q deletion
261222	Distal 16p11.2 microdeletion syndrome
319171	Distal 17p13.1 microdeletion syndrome
261257	Distal 17p13.3 microdeletion syndrome
1597	Distal 17q deletion
261330	Distal 22q11.2 microdeletion syndrome
261337	Distal 22q11.2 microduplication syndrome
63273	Distal ABD-filaminopathy
178400	Distal anterior compartment myopathy
1146	Distal arthrogryposis type 1
2053	Distal arthrogryposis type 2A
1147	Distal Arthrogryposis type 2B

ORPHA Number	Disease name
376	Distal arthrogryposis type 3
65720	Distal arthrogryposis type 4
1154	Distal arthrogryposis type 5
329457	Distal arthrogryposis type 5 without ophthalmoparesis
329457	Distal arthrogryposis type 5 without ophthalmoplegia
329457	Distal arthrogryposis type 5D
1144	Distal arthrogryposis type 6
3377	Distal arthrogryposis type 7
65743	Distal arthrogryposis type 8
115	Distal arthrogryposis type 9
251515	Distal arthrogryposis type 10
376	Distal arthrogryposis type IIA
1154	Distal arthrogryposis type IIB
65720	Distal arthrogryposis type IID
1154	Distal arthrogryposis with ophthalmoplegia
254351	Distal del(7)(q11.23)
261222	Distal del(16)(p11.2)
319171	Distal del(17)(p13.1)
261257	Distal del(17)(p13.3)
261330	Distal del(22)(q11.2)
36367	Distal deletion 1q
280	Distal deletion 4p
96145	Distal deletion 4q
96125	Distal deletion 6p
96126	Distal deletion 7p
1636	Distal deletion 7q36
1642	Distal deletion 9p
96148	Distal deletion 10q
2308	Distal deletion 11q
280325	Distal deletion 12p
96149	Distal deletion 12q
96168	Distal deletion 13q34
96150	Distal deletion 14q
96129	Distal deletion 19p
96152	Distal deletion 20q
261102	Distal dup(7)(q11.23)
261337	Distal dup(22)(q11.2)
293939	Distal dup(X)q(28)
96069	Distal duplication 1p36
96070	Distal duplication 2p
96094	Distal duplication 2q
96071	Distal duplication 3p
96072	Distal duplication 4p
96096	Distal duplication 4q
96097	Distal duplication 5q
1745	Distal duplication 6p
96098	Distal duplication 6q
96074	Distal duplication 7p
96100	Distal duplication 8q

ORPHA Number	Disease name
96101	Distal duplication 9q
96102	Distal duplication 10q
96103	Distal duplication 11q
96105	Distal duplication 13q
1705	Distal duplication 14q
1707	Distal duplication 15q
96078	Distal duplication 16p
96106	Distal duplication 16q
3379	Distal duplication 17q
1716	Distal duplication 18q
1717	Distal duplication 19q
96107	Distal duplication 20q
96109	Distal duplication 22q
1762	Distal duplication Xq
53739	Distal hereditary motor neuropathy
139518	Distal hereditary motor neuropathy type 1
139525	Distal hereditary motor neuropathy type 2
139547	Distal hereditary motor neuropathy type 3 and type 4
139536	Distal hereditary motor neuropathy type 5
100998	Distal hereditary motor neuropathy type 5B
98920	Distal hereditary motor neuropathy type 6
139589	Distal hereditary motor neuropathy type 7
357043	Distal hereditary motor neuropathy with upper motor neuron signs
139552	Distal hereditary motor neuropathy, Jerash type
1307	Distal limb deficiencies - micrognathia syndrome
36367	Distal monosomy 1q
1620	Distal monosomy 3p
280	Distal monosomy 4p
96145	Distal monosomy 4q
1627	Distal monosomy 5q
96125	Distal monosomy 6p
96126	Distal monosomy 7p
254351	Distal monosomy 7q11.23
1636	Distal monosomy 7q36
1642	Distal monosomy 9p
1580	Distal monosomy 10p
96148	Distal monosomy 10q
2308	Distal monosomy 11q
280325	Distal monosomy 12p
96149	Distal monosomy 12q
1590	Distal monosomy 13q
96150	Distal monosomy 14q
1596	Distal monosomy 15q
261222	Distal monosomy 16p11.2
261257	Distal monosomy 17p13.3
1597	Distal monosomy 17q
96129	Distal monosomy 19p13.3

ORPHA Number	Disease name
96152	Distal monosomy 20q
261330	Distal monosomy 22q11.2
59135	Distal myopathy type 1
178400	Distal myopathy with anterior tibial onset
34521	Distal myopathy with early respiratory muscle involvement
63273	Distal myopathy with posterior leg and anterior hand involvement
602	Distal myopathy with rimmed vacuoles
600	Distal myopathy with vocal cord weakness
609	Distal myopathy, Markesbery-Griggs type
602	Distal myopathy, Nonaka type
603	Distal myopathy, Swedish type
609	Distal myopathy, Udd type
603	Distal myopathy, Welander type
2776	Distal osteolysis - short stature - intellectual deficit
18	Distal renal tubular acidosis
93610	Distal renal tubular acidosis with anemia
53739	Distal spinal muscular atrophy
139525	Distal spinal muscular atrophy type 2
139547	Distal spinal muscular atrophy type 3
206580	Distal spinal muscular atrophy type 4
139536	Distal spinal muscular atrophy type 5
139589	Distal spinal muscular atrophy with vocal cord paralysis
3248	Distal symphalangism
314588	Distal tetrasomy 15q
96069	Distal trisomy 1p36
96070	Distal trisomy 2p
96094	Distal trisomy 2q
96071	Distal trisomy 3p
96072	Distal trisomy 4p
96096	Distal trisomy 4q
96097	Distal trisomy 5q
1745	Distal trisomy 6p
96098	Distal trisomy 6q
96074	Distal trisomy 7p
261102	Distal trisomy 7q11.23
96100	Distal trisomy 8q
96101	Distal trisomy 9q
96102	Distal trisomy 10q
96103	Distal trisomy 11q
96105	Distal trisomy 13q
1705	Distal trisomy 14q
1707	Distal trisomy 15q
96078	Distal trisomy 16p
96106	Distal trisomy 16q
3379	Distal trisomy 17q
1716	Distal trisomy 18q
1717	Distal trisomy 19q
96107	Distal trisomy 20q
96109	Distal trisomy 22q
261337	Distal trisomy 22q11.2
293939	Distal trisomy Xq28
293939	Distal Xq28 microduplication syndrome
1683	Distichiasis - congenital heart defects - peripheral vascular anomalies
1916	Distilbene embryofetopathy
1685	Distomatosis
1685	Distomiasis
247353	DITRA
3439	DK phocomelia syndrome
91131	DK1-CDG syndrome
1775	DKC
544	DLBCL
300849	DLBCL of the CNS
300888	DLBCL with chronic inflammation
2394	DLD deficiency
252031	DLM
221	DM
273	DM1
98896	DMD
243343	DMG dehydrogenase deficiency
243343	DMGDH deficiency
99812	DNA ligase IV deficiency
251946	DNET
251975	DNT of the cerebellum
1215	DOA+
217390	DOCK8 immunodeficiency syndrome
91131	Dolichol kinase deficiency
2616	Dolichospondylic dysplasia
86309	Dolichyl-phosphate N-acetylgalactosamine phosphotransferase deficiency
79322	Dol-P-mannosyltransferase deficiency
231226	Dominant beta-thalassemia
75376	Dominant drusen
898	Dominant hyaloideoretinal dystrophy of Wagner
244305	Dominant hypophosphatemia with nephrolithiasis or osteoporosis
75376	Dominant radial drusen
90035	Donath-Landsteiner hemolytic anemia
90035	Donath-Landsteiner syndrome
2143	Donnai-Barrow syndrome
508	Donohue syndrome
79500	DOOR syndrome
1942	Doose syndrome
230	Dopamine beta-hydroxylase deficiency
255	Dopa-responsive dystonia
70594	Dopa-responsive dystonia due to sepiapterin reductase deficiency
98907	Dorfman-Chanarin disease
3426	DORV
99043	DORV, Fallot type
869	Double A syndrome
216694	Double discordance
1464	Double inlet left ventricle
141091	Double nose
3427	Double outlet left ventricle
3426	Double outlet right ventricle
99047	Double outlet right ventricle with doubly committed ventricular septal defect
99046	Double outlet right ventricle with non-committed subpulmonary ventricular septal defect
99044	Double outlet right ventricle with subaortic ventricular septal defect
99045	Double outlet right ventricle with subpulmonary ventricular septal defect
99043	Double outlet right ventricle, Fallot type
3286	Double tachycardia induced by catecholamines
3411	Double uterus - hemivagina - renal agenesis
3411	Double uterus and obstructed hemivagina syndrome
8	Double y
95474	Double-orifice mitral valve
79145	Dowling-Degos disease
870	Down syndrome
75376	Doyne honeycomb retinal dystrophy
86309	DPAGT1-CDG syndrome
314621	DPG-plus syndrome
71274	DPL
79322	DPM1-CDG syndrome
329178	DPM2-CDG syndrome
263494	DPM3-CDG syndrome
231	Dracunculiasis
231	Dracunculosis
220	Drash syndrome
33069	Dravet syndrome
130	Dream disease
139402	DRESS syndrome
101	DRPLA
233	DRS
18	dRTA
93610	dRTA with anemia
139402	Drug rash with eosinophilia and systemic symptoms
139402	Drug reaction eosinophilic systemic syndrome
90037	Drug-induced AIHA
90037	Drug-induced autoimmune hemolytic anemia
90157	Drug-induced localized lipodystrophy
231111	Drug-induced lupus erythematosus
94086	Drummond syndrome

ORPHA Number	Disease name
99887	DS-AMKL
53739	dSMA
98920	dSMA1
139525	dSMA2
139547	dSMA3
206580	dSMA4
314485	dSMA5
83469	DSRCT
99789	DTDP1
99791	DTDP2
2639	Du Pan syndrome
50817	Duane anomaly - myopathy - scoliosis
233	Duane retraction syndrome
233	Duane syndrome
93293	Duane-radial ray syndrome
261647	Duane-radial ray syndrome due to a point mutation
261638	Duane-radial ray syndrome due to monosomy 20q13
234	Dubin-Johnson syndrome
234	Dubin-Sprinz disease
235	Dubowitz syndrome
262	Duchenne and Becker muscular dystrophy
98896	Duchenne muscular dystrophy
280315	Duct-centric pancreatitis
2442	Duncan disease
2348	Dunnigan syndrome
1203	Duodenal atresia
100076	Duodenal endocrine tumor
250994	Dup(1)(q21.1)
313947	Dup(2)(q23.1)
294026	Dup(2)(q31.1)
96095	Dup(3)(q26)
329802	Dup(5)(p13)
228415	Dup(5)(q35)
314034	Dup(7)(p22.1)
96121	Dup(7)(q11.23)
251076	Dup(8)(p23.1)
228399	Dup(8)(q12)
276422	Dup(10)(q22.3q23.3)
300305	Dup(11)p(15.4)
261229	Dup(14)(q11.2)
238446	Dup(15)(q11q13)
261204	Dup(16)(p11.2p12.2)
261243	Dup(16)(p13.11)
96078	Dup(16)(p13.3)
217385	Dup(17)(p13.3)
139474	Dup(17)(q11.2)
261272	Dup(17)(q12)
217340	Dup(17)(q21.31)
261290	Dup(17p)
261318	Dup(20p)
1727	Dup(22)(q11)
284180	Dup(X)(p22)
284180	Dup(X)(p22.13p22.2)
314389	Dup(X)(q12-q13.3)
261483	Dup(X)(q27.3q28)
261344	Duplication 1q
1738	Duplication 4p
1739	Duplication 4q
1742	Duplication 5p
264450	Duplication 8p
1752	Duplication 8q
96167	Duplication 8q/deletion 8p
236	Duplication 9p
1699	Duplication 12p
1715	Duplication 18p
1727	Duplication 22q11.2
261318	Duplication of 20p
91357	Duplication of the esophagus
314621	Duplication of the pituitary gland
314621	Duplication of the pituitary gland-plus syndrome
237	Duplication of urethra
284180	Duplication Xp22
3306	Duplication/inversion 15q11
97339	Dural sinus malformation
1656	Durhing-Brocq disease
98984	Dusty cataract
3377	Dutch-Kentucky syndrome
2650	Dwarfism - intellectual deficit - eye abnormality
2569	Dwarfism - stiff joint - ocular abnormalities
239	Dyggve-Melchior-Clausen disease
2274	Dykes-Markes-Harper syndrome
1765	Dyschondrosteosis - nephritis
41	Dyschromatosis symmetrica hereditaria
241	Dyschromatosis universalis
251946	Dysembryoplastic neuroepithelial tumor
251975	Dysembryoplastic neuroepithelial tumor of the cerebellum
1766	Dysequilibrium syndrome
842	Dysgerminoma
99912	Dysgerminomatous germ cell cancer
3010	Dysharmonic skeletal maturation - muscular fibre disproportion
1775	Dyskeratosis congenita
1779	Dysmorphism - cleft palate - loose skin
289553	Dysmorphism - conductive hearing loss - heart defect
1780	Dysmorphism - multiple structural anomalies
2104	Dysmorphism - pectus carinatum - joint laxity
2282	Dysmorphism - short stature - deafness - disorder of sex development
2282	Dysmorphism - short stature - deafness - pseudohermaphroditism
1782	Dysosteosclerosis
800	Dysostosis enchondralis metaepiphysaria, Catel-Hempel type
1798	Dysostosis, Stanescu type
99082	Dysphagia lusoria
1822	Dysplasia epiphysealis hemimelica
168621	Dysplasia of head of femur, Meyer type
2204	Dysplastic cortical hyperostosis
65285	Dysplastic gangliocytoma of the cerebellum
325	Dysprothrombinemia
2476	Dysraphism - cleft lip/palate - limb reduction defects
1804	Dyssegmental dysplasia - glaucoma
156731	Dyssegmental dysplasia, Rolland-Desbuquois type
1865	Dyssegmental dysplasia, Silverman-Handmaker type
85198	Dysspondyloenchondromatosis
71517	Dystonia 12
210571	Dystonia 16
98811	Dystonia 18
256	Dystonia musculorum deformans
199351	Dystonia-parkinsonism, Paisan-Ruiz type
293381	Dystrophia Helsinglandica
293381	Dystrophia Smolandiensis
303	Dystrophic epidermolysis bullosa
79409	Dystrophic epidermolysis bullosa inversa
89843	Dystrophic epidermolysis bullosa pruriginosa
158676	Dystrophic epidermolysis bullosa, nails only
256	DYT1
53351	DYT3
98805	DYT4
255	DYT5
98808	DYT5a
98806	DYT6
93963	DYT7
53583	DYT9
36899	DYT11
71517	DYT12
98807	DYT13
210566	DYT15
210571	DYT16
98811	DYT18
99657	DYT2
306734	DYT21

ORPHA Number	Disease name
2394	E3-deficient maple syrup urine disease
2970	Eagle-Barret syndrome
40923	Eales disease
1934	Early infantile epileptic encephalopathy
1934	Early infantile epileptic encephalopathy with suppression-bursts
1935	Early myoclonic encephalopathy
1935	Early myoclonic encephalopathy with suppression-bursts
157941	Early onset prion disease with prominent psychiatric features
1020	Early-onset autosomal dominant Alzheimer disease
98815	Early-onset benign childhood occipital epilepsy
1177	Early-onset cerebellar ataxia with retained tendon reflex
84132	Early-onset desmin-related myopathy
1667	Early-onset diabetes mellitus with multiple epiphyseal dysplasia
289266	Early-onset epileptic encephalopathy and intellectual deficit due to GRIN2A mutation
1020	Early-onset familial autosomal dominant Alzheimer disease
256	Early-onset generalized torsion dystonia
88660	Early-onset hypertension with exacerbation in pregnancy
324290	Early-onset Lafora body disease
79242	Early-onset multiple carboxylase deficiency
289377	Early-onset myopathy with fatal cardiomyopathy
2828	Early-onset Parkinson disease
2379	Early-onset parkinsonism - intellectual deficit
256	Early-onset primary dystonia
352654	Early-onset progressive neurodegeneration - blindness - ataxia - spasticity
96369	Early-onset schizophrenia
313772	Early-onset spastic ataxia-neuropathy syndrome
256	Early-onset torsion dystonia
98890	Early-onset X-linked optic atrophy
2554	Ear-patella-short stature syndrome
199343	EAST syndrome
83594	Eastern equine encephalitis
83594	Eastern equine encephalomyelitis
1973	Eastman-Bixler syndrome
166418	Eating seizures
86880	EATL
79406	EB progressive
231249	E-beta-thalassemia
305	EBJ
79405	EBJ-I

ORPHA Number	Disease name
319218	Ebola fever
319218	Ebola hemorrhagic fever
319218	Ebola virus disease
304	EBS
89838	EBS-AR
79400	EBS-loc
257	EBS-MD
158681	EBS-migr
79397	EBS-MP
79401	EBS-O
158684	EBS-PA
89839	EBSS
1880	Ebstein anomaly of the tricuspid valve
1880	Ebstein malformation
313920	EBVaGC
313920	EBV-associated gastric carcinoma
289661	EBV-positive DLBCL of the elderly
199332	ECO syndrome
99102	Ectasia of the left appendage
99102	Ectasia of the left auricle
99101	Ectasia of the right atrial appendage
99101	Ectasia of the right atrial auricle
35737	Ectasic coloboma
140936	Ectodermal dysplasia - acanthosis nigricans
1806	Ectodermal dysplasia - blindness
3354	Ectodermal dysplasia - cataracts - kyphoscoliosis
247827	Ectodermal dysplasia - cutaneous syndactyly syndrome
1897	Ectodermal dysplasia - ectrodactyly - macular dystrophy
1812	Ectodermal dysplasia - intellectual deficit - central nervous system malformation
1883	Ectodermal dysplasia - sensorineural deafness
158668	Ectodermal dysplasia - skin fragility syndrome
247820	Ectodermal dysplasia - syndactyly syndrome
69083	Ectodermal dysplasia with natal teeth, Turnpenny type
1816	Ectodermal dysplasia, Berlin type
1818	Ectodermal dysplasia, tricho-odonto-onychial type
1884	Ectopia lentis - chorioretinal dystrophy - myopia
1885	Ectopia lentis syndrome
99889	Ectopic ACTH secreting tumor
231632	Ectopic aldosterone-producing tumor
99889	Ectopic Cushing syndrome
95496	Ectopic neurohypophysis
2440	Ectrodactyly

ORPHA Number	Disease name
1896	Ectrodactyly - ectodermal dysplasia - cleft lip/palate
1892	Ectrodactyly - polydactyly
1894	Ectrodactyly - spina bifida - cardiopathy
1997	Ectropion inferior - cleft lip and or palate
906	Eczema-thrombocytopenia-immunodeficiency syndrome
98813	EDA-ID
247827	EDCS
293936	EDICT syndrome
1895	Edinburgh malformation syndrome
93308	EDM1
93307	EDM4
93311	EDM5
261	EDMD
98863	EDMD1
98855	EDMD3
90309	EDS I
90318	EDS II
285	EDS III
286	EDS IV
198	EDS IX
286	EDS type 4
75497	EDS V
1900	EDS VIA
1899	EDS VII
99875	EDS VIIA
99876	EDS VIIB
1901	EDS VIIC
75392	EDS VIII
82004	EDS with periventricular heterotopia
300179	EDS with progressive kyphoscoliosis, myopathy, and deafness
300179	EDS with progressive kyphoscoliosis, myopathy, and hearing loss
75501	EDS X
2295	EDS XI
2953	EDS, arthrogryposic type
230851	EDS, cardiac valvular type
287	EDS, classic type
230839	EDS, classic-like type
2953	EDS, Kosho type
300179	EDS, kyphoscoliotic and hearing loss type
1900	EDS, kyphoscoliotic type
2953	EDS, musculocontractural type
1900	EDS, oculoscoliotic type
75496	EDS, progeroid type
157965	EDS, spondylocheirodysplastic type
230845	EDS, vascular-like type
230857	EDS/OI syndrome
247820	EDSS
247820	EDSS1

ORPHA Number	Disease name
247827	EDSS2
178464	Edström Myopathy
3380	Edwards syndrome
2668	Edwards-Patton-Dilly syndrome
304	EEB
322	EEC
1896	EEC syndrome
1897	EEM syndrome
357131	Effort subclavian vein thrombosis
101039	EFMR
2070	EGE
319218	EHF
312	EHK
230839	Ehlers-Danlos syndrome due to tenascin-X deficiency
90309	Ehlers-Danlos syndrome type 1
75501	Ehlers-Danlos syndrome type 10
2295	Ehlers-Danlos syndrome type 11
90318	Ehlers-Danlos syndrome type 2
285	Ehlers-Danlos syndrome type 3
286	Ehlers-Danlos syndrome type 4
75497	Ehlers-Danlos syndrome type 5
1900	Ehlers-Danlos syndrome type 6A
1899	Ehlers-Danlos syndrome type 7
99875	Ehlers-Danlos syndrome type 7A
99876	Ehlers-Danlos syndrome type 7B
1901	Ehlers-Danlos syndrome type 7C
75392	Ehlers-Danlos syndrome type 8
286	Ehlers-Danlos syndrome type IV
198	Ehlers-Danlos syndrome type IX
82004	Ehlers-Danlos syndrome with periventricular heterotopia
75501	Ehlers-Danlos syndrome with platelet dysfunction from fibronectin abnormality
300179	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and deafness
300179	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss
1899	Ehlers-Danlos syndrome, arthrochalasia type
1899	Ehlers-Danlos syndrome, arthrochalasic type
2953	Ehlers-Danlos syndrome, arthrogryposic type
230851	Ehlers-Danlos syndrome, cardiac valvular type
287	Ehlers-Danlos syndrome, classic type
230839	Ehlers-Danlos syndrome, classic-like type
1901	Ehlers-Danlos syndrome, dermatosparaxis type
75501	Ehlers-Danlos syndrome, fibronectin-deficient
75501	Ehlers-Danlos syndrome, fibronectinemic type
285	Ehlers-Danlos syndrome, hypermobile type
285	Ehlers-Danlos syndrome, hypermobility type
2953	Ehlers-Danlos syndrome, Kosho type
300179	Ehlers-Danlos syndrome, kyphoscoliotic and deafness type
300179	Ehlers-Danlos syndrome, kyphoscoliotic and hearing loss type
1900	Ehlers-Danlos syndrome, kyphoscoliotic type
2953	Ehlers-Danlos syndrome, musculocontractural type
1900	Ehlers-Danlos syndrome, oculoscoliotic type
75392	Ehlers-Danlos syndrome, periodontitis type
75496	Ehlers-Danlos syndrome, progeroid type
157965	Ehlers-Danlos syndrome, spondylocheirodysplastic type
198	Ehlers-Danlos syndrome, type 9
286	Ehlers-Danlos syndrome, vascular type
230845	Ehlers-Danlos syndrome, vascular-like type
230857	Ehlers-Danlos/osteogenesis imperfecta syndrome
1902	Ehrlichiosis
820	Ehrmann-Sneddon syndrome
312	EI
1934	EIEE
165991	EIHI
79106	Eiken syndrome
97214	Eisenmenger syndrome
317	EKV
228240	Elastoderma
228243	Elastofibroma dorsi
228254	Elastoma
79148	Elastosis perforans serpiginosa
228236	Elastotic striae
26791	Electron transfer flavoprotein deficiency
26791	Electron transfer flavoprotein ubiquinone oxidoreductase deficiency
33445	Elejalde disease
221054	Elejalde syndrome
289	Ellis Van Creveld syndrome
2516	Ellis-Yale-Winter syndrome
1997	Elsching syndrome
96170	Emanuel syndrome
3226	Emberger syndrome
1914	Embryofetopathy due to oral anticoagulant therapy
180226	Embryonal carcinoma
99757	Embryonal rhabdomyosarcoma
178315	Embryonal sarcoma of the liver
1664	Embryonary disorganization syndrome
983	Embryonic testicular regression syndrome
139431	EMEA
261	Emerinopathy
261	Emery-Dreifuss muscular dystrophy
1927	Emery-Nelson syndrome
83600	Encephalitis lethargica
221126	Encephaloclastic proliferative vasculopathy
2396	Encephalocraniocutaneous lipomatosis
319678	Encephalopathy - hypertrophic cardiomyopathy - renal tubular disease
1261	Encephalopathy - intracerebral calcification - retinal degeneration
1035	Encephalopathy due to beta-mercaptolactate-cysteine disulfiduria
71277	Encephalopathy due to GLUT1 deficiency
79155	Encephalopathy due to hydroxykynureninuria
139406	Encephalopathy due to prosaposin deficiency
833	Encephalopathy due to sulfite oxidase deficiency
210128	Encephalopathy due to urocanase deficiency
51	Encephalopathy with basal ganglia calcification
51	Encephalopathy with intracranial calcification and chronic lymphocytosis of cerebrospinal fluid
296	Enchondromatosis
99075	Encircling double aortic arch
100079	Endocrine tumor of the appendix
199332	Endocrine-cerebro-osteodysplasia syndrome
876	Endodermal sinus tumor
252006	Endodermal sinus tumor of the central nervous system
98974	Endoepithelial corneal dystrophy
213741	Endometrial adenoid cystic carcinoma
213726	Endometrial capillary carcinoma
213716	Endometrial squamous cell carcinoma
213711	Endometrial stromal sarcoma
213746	Endometrial transitional cell carcinoma
213721	Endometrial undifferentiated carcinoma
2022	Endomyocardial fibroelastosis
199323	Endophthalmitis
209959	Endophthalmitis phacoanaphylactica
2790	Endosteal hyperostosis, Worth type
85186	Endosteal sclerosis - cerebellar hypoplasia
293936	Endothelial dystrophy-iris hypoplasia-congenital cataract-stromal thinning syndrome
137602	Endotheliitis
1937	Eng-Strom syndrome
53540	Enhanced S-cone syndrome

ORPHA Number	Disease name
60015	Enlarged parietal foramina
83620	Enteric anendocrinosis
141071	Enteric duplication cyst of the tongue
99745	Enteric fever
86880	Enteropathy-associated T-cell lymphoma
86880	Enteropathy-type T-cell lymphoma
292	Enterovirus antenatal infection
85438	Enthesitis-related arthritis
1939	Envenomization by Bothrops lanceolatus
1939	Envenomization by the Martinique lancehead viper
1177	EOCARR
1020	EOFAD
168829	EOPPC
901	Eosinophilic cellulitis
75566	Eosinophilic endocarditis
73247	Eosinophilic esophagitis
3165	Eosinophilic fasciitis
2070	Eosinophilic gastroenteritis
99871	Eosinophilic granuloma
482	Eosinophilic lymphogranuloma
256	EOTD
251880	Ependymoblastoma
251636	Ependymoma
99169	Epiblepharon
185	Epibronchial right pulmonary artery syndrome
231742	Epibulbar lipodermoid - preauricular appendage - polythelia
83314	Epidemic typhus
35125	Epidermal hamartoma syndrome
2694	Epidermal nevus - vitamin D resistant rickets
35125	Epidermal nevus syndrome
302	Epidermodysplasia verruciformis
46487	Epidermolysis bullosa acquisita
305	Epidermolysis bullosa atrophicans
303	Epidermolysis bullosa dystrophica
79404	Epidermolysis bullosa letalis
304	Epidermolysis bullosa simplex
158668	Epidermolysis bullosa simplex due to plakophilin deficiency
79400	Epidermolysis bullosa simplex of palms and soles
89839	Epidermolysis bullosa simplex superficialis
2325	Epidermolysis bullosa simplex with anodontia/hypodontia
158681	Epidermolysis bullosa simplex with circinate migratory erythema
79397	Epidermolysis bullosa simplex with mottled pigmentation
257	Epidermolysis bullosa simplex with muscular dystrophy
158684	Epidermolysis bullosa simplex with pyloric atresia
79396	Epidermolysis bullosa simplex, Dowling-Meara type
79396	Epidermolysis bullosa simplex, herpetiformis
79399	Epidermolysis bullosa simplex, Köbner type
79399	Epidermolysis bullosa simplex, Koebner type
79401	Epidermolysis bullosa simplex, Ogna type
79400	Epidermolysis bullosa simplex, Weber-Cockayne type
312	Epidermolytic hyperkeratosis
312	Epidermolytic ichthyosis
2199	Epidermolytic palmoplantar keratoderma
2199	Epidermolytic palmoplantar keratoderma of Voerner
2199	Epidermolytic palmoplantar keratoderma of Vörner
141077	Epignathus
1946	Epilepsy - dementia - amelogenesis imperfecta
1948	Epilepsy - microcephaly - skeletal dysplasia
65683	Epilepsy due to FCD
1951	Epilepsy telangiectasia
86911	Epilepsy with myoclonic absences
1942	Epilepsy with myoclonic-astatic seizures
353217	Epileptic encephalopathy with global cerebral demyelination
79238	Epimerase deficiency galactosemia
1819	Epimetaphyseal skeletal dysplasia
1825	Epiphyseal dysplasia - hearing loss - dysmorphism
1824	Epiphyseal dysplasia - microcephaly - nystagmus
1952	Epiphyseal stippling syndrome - osteoclastic hyperplasia
649	Episkopi blindness
37612	Episodic ataxia type 1
97	Episodic ataxia type 2
79135	Episodic ataxia type 3
79136	Episodic ataxia type 4
211067	Episodic ataxia type 5
209967	Episodic ataxia type 6
209970	Episodic ataxia type 7
37612	Episodic ataxia with myokymia
53583	Episodic choreoathetosis/spasticity
93928	Epispadias
293381	Epithelial recurrent erosion dystrophy
103912	Epithelio-exfoliative colitis - deafness
157791	Epithelioid hemangioendothelioma
293202	Epithelioid sarcoma
254698	Epithelioid trophoblastic tumor
91414	Epithelioma calcificans of Malherbe
79278	EPP
2199	EPPK
313920	Epstein-Barr virus-associated gastric carcinoma
289661	Epstein-Barr Virus-positive diffuse large B cell lymphoma of the elderly
85438	ERA
229	Erdheim disease
35687	Erdheim-Chester disease
293381	ERED
999	Ermine phenotype
160148	Eroded polypoid hyperplasia
222	Erosive pustular dermatosis of the scalp
228264	Eruptive collagenoma
90000	Erythema elevatum diutinum
231031	Erythema palmaris hereditarium
729	Erythremia
308473	Erythrocyte epimerase deficiency galactosemia
308473	Erythrocyte galactose epimerase deficiency
308473	Erythrocyte GALE deficiency
308473	Erythrocyte GALE-D
171690	Erythrocyte lactate transporter defect
308473	Erythrocyte UDP-galactose-4-epimerase deficiency
308473	Erythrocyte uridine diphosphate galactose-4-epimerase deficiency
314	Erythroderma desquamativum
79394	Erythrodermic ichthyosis
247165	Erythroedema polyneuritis
315	Erythrokeratoderma "en cocardes"
1955	Erythrokeratodermia - ataxia
317	Erythrokeratodermia variabilis
171851	Erythrokeratodermia variabilis 3
171851	Erythrokeratodermia variabilis, Kamouraska type
317	Erythrokeratodermia variabilis, Mendes da Costa type
318	Erythroleukemia
1956	Erythromelalgia
79278	Erythropoietic protoporphyria
280379	Erythropoietic uroporphyria associated with myeloid malignancy
2405	Escher-Hirt syndrome
2990	Escobar syndrome
2990	Escobar variant multiple pterygium syndrome
99976	Esophageal adenocarcinoma
1199	Esophageal atresia
70482	Esophageal cancer

ORPHA Number	Disease name
70482	Esophageal carcinoma
100047	Esophageal duplication cyst
99977	Esophageal epidermoid carcinoma
99977	Esophageal squamous cell carcinoma
65748	ESS1
91138	Essential cryoglobulinemia
2056	Essential fructosuria
98981	Essential iris atrophy
91138	Essential mixed cryoglobulinemia
73	Essential osteolysis
2843	Essential pentosuria
98682	Essential strabismus
3318	Essential thrombocythemia
3318	Essential thrombocytosis
1957	Esthesioneuroblastoma
785	Estrogen resistance syndrome
3318	ET
26791	ETFA deficiency
26791	ETFB deficiency
26791	ETFDH deficiency
31826	Ethylene glycol poisoning
51188	Ethylmalonic encephalopathy
983	ETRS
86880	ETTL
2892	Euhidrotic ectodermal dysplasia
99172	Euryblepharon
1959	Evans syndrome
2990	EVMPS
319	Ewing sarcoma
99734	Exercise-induced delayed-onset myotonia
165991	Exercise-induced hyperinsulinemic hypoglycemia
165991	Exercise-induced hyperinsulinism
289586	Exfoliative ichthyosis
116	Exomphalos - macroglossia - gigantism
1962	Exostoses - anetodermia - brachydactyly type E
322	Exstrophy-epispadias complex
321	EXT1/EXT2-CDG syndrome
3294	Extensor tendons of finger anomalies
141074	External auditory canal aplasia/hypoplasia
141074	External auditory canal stenosis/atresia
231632	Extra-adrenal aldosterone-producing tumor
289362	Extracranial embryonal carcinoma
66662	Extracutaneous mastocytoma
280811	Extralobar congenital bronchopulmonary sequestration
280811	Extralobar congenital pulmonary sequestration
2800	Extramammary Paget disease
86850	Extramedullary myeloid tumor
100022	Extramedullary soft tissue plasmacytoma
100002	Extraneural perineurioma
52417	Extranodal marginal zone B-cell lymphoma
86879	Extranodal nasal NK/T cell lymphoma
168829	Extra-ovarian primary peritoneal carcinoma
209916	Extraskeletal myxoid chondrosarcoma
1964	Extrasystoles - short stature - hyperpigmentation - microcephaly
251927	Extraventricular neurocytoma
31740	Extrinsic allergic alveolitis
2725	Eye defects - arachnodactyly - cardiopathy
3172	Eyebrow duplication - syndactyly
2985	Eyebrows and eyelashes absence - intellectual deficit
139431	Eyelid myoclonia with and without absences
957	F syndrome
35909	F5F8D
324	Fabry disease
1969	FACES syndrome
1167	Facial asymmetry - temporal seizures
141051	Facial dermoid cyst
352712	Facial dysmorphism - immunodeficiency - livedo - short stature
2588	Facial dysmorphism - intellectual deficit - short stature - hearing loss
1970	Facial dysmorphism - macrocephaly - myopia - Dandy-Walker malformation
1778	Facial dysmorphism - shawl scrotum - joint laxity
221083	Facial hemispasm
85162	Facial onset sensory and motor neuronopathy
3237	Facio-audio-symphalangism
374	Facioauriculovertebral dysplasia
1973	Faciocardiorenal syndrome
3071	Faciocutaneoskeletal syndrome
915	Faciodigitogenital syndrome
1974	Facio-digito-genital syndrome, Kuwait type
915	Faciogenital dysplasia
1300	Facio-genito-popliteal syndrome
2143	Facio-oculo-acoustico-renal syndrome
2048	Facio-pharyngo-glosso-masticatory diplegia
269	Facioscapulohumeral dystrophy
269	Facioscapulohumeral muscular dystrophy
269	Facioscapulohumeral myopathy
98879	Factor IX deficiency
220436	Factor V Quebec
98878	Factor VIII deficiency
300359	FACU
306550	FADD-related immunodeficiency
994	FADS
882	FAH deficiency
329308	FAHN
1077	Failure of teeth eruption
254707	Faisalabad histiocytosis
3304	Fallot complex - intellectual deficit - growth delay
86814	FAME
86	Familial abdominal aortic aneurysm
637	Familial acoustic neurinoma
637	Familial acoustic neuroma
88619	Familial acute necrotizing encephalopathy
733	Familial adenomatous polyposis
261584	Familial adenomatous polyposis due to 5q22.2 microdeletion
261584	Familial adenomatous polyposis due to del(5)(q22.2)
261584	Familial adenomatous polyposis due to monosomy 5q22.2
404	Familial adrenal adenoma
95700	Familial adrenal hypoplasia with absent pituitary LH
95700	Familial adrenal hypoplasia with absent pituitary luteinizing hormone
95700	Familial adrenal hypoplasia, miniature type
86814	Familial adult myoclonic epilepsy
164736	Familial advanced sleep-phase syndrome
98880	Familial afibrinogenemia
1020	Familial Alzheimer disease
280397	Familial Alzheimer-like prion disease
319465	Familial AML
85450	Familial amyloid nephropathy
93560	Familial amyloid nephropathy due to apolipoprotein AI variant
238269	Familial amyloid nephropathy due to apolipoprotein AII variant
93562	Familial amyloid nephropathy due to fibrinogen A alpha-chain variant
93561	Familial amyloid nephropathy due to lysozyme variant
85447	Familial amyloid polyneuropathy
85448	Familial amyloid polyneuropathy type 4
85448	Familial amyloidosis, Finnish type
228277	Familial anetoderma
199279	Familial angiolipomatosis
91378	Familial angioneurotic edema
229	Familial aortic dissection
309020	Familial apoC-II deficiency
309020	Familial apolipoprotein C-II deficiency
1416	Familial articular chondrocalcinosis
99781	Familial articular chondrocalcinosis type 1
99782	Familial articular chondrocalcinosis type 2
334	Familial atrial fibrillation

ORPHA Number	Disease name
615	Familial atrial myxoma
300359	Familial atypical cold urticaria
86820	Familial avascular necrosis of femoral head
2398	Familial benign cervical lipomatosis
2841	Familial benign chronic pemphigus
1551	Familial benign copper deficiency
405	Familial benign hypercalcemia
405	Familial benign hypocalciuric hypercalcemia
231160	Familial berry aneurysm
227535	Familial breast cancer
227535	Familial breast carcinoma
36382	Familial CAD
2678	Familial café-au-lait spots
91415	Familial capillary hemangioma
1768	Familial caudal dysgenesis
169085	Familial CD8 deficiency
892	Familial cerebelloretinal angiomatosis
221061	Familial cerebral cavernoma
221061	Familial cerebral cavernous malformation
231160	Familial cerebral saccular aneurysm
36382	Familial cervical artery dissections
1428	Familial chondromalacia patellae
238578	Familial clubfoot due to 17q23.1q23.2 microduplication
293144	Familial clubfoot due to 5q31 microdeletion
293150	Familial clubfoot due to PITX1 point mutation
199315	Familial clubfoot with or without associated lower limb anomalies
47045	Familial cold autoinflammatory syndrome
247868	Familial cold autoinflammatory syndrome type 2
47045	Familial cold urticaria
300359	Familial cold urticaria with common variable immunodeficiency
99166	Familial combined hyperlipoproteinemia
238722	Familial congenital controlateral synkinesia
95494	Familial congenital hypopituitarism
238722	Familial congenital mirror movements
91498	Familial congenital palsy of trochlear nerve
86814	Familial cortical myoclonic tremor and epilepsy
319189	Familial cortical myoclonus
85453	Familial cutaneous amyloidosis
53296	Familial cutaneous collagenoma
313846	Familial cutaneous telangiectasia and oropharyngeal predisposition cancer syndrome
211	Familial cylindromatosis
97345	Familial dementia, British type
97346	Familial dementia, Danish type
1799	Familial developmental dysphasia
85169	Familial digital arthropathy-brachydactyly
300751	Familial dilated cardiomyopathy with conduction defect due to LMNA mutation
18	Familial distal primary acidosis
85192	Familial doughnut lesions of skull
75376	Familial drusen
79142	Familial Dupuytren contracture
1764	Familial dysautonomia
314381	Familial dysautonomia with contractures
98881	Familial dysfibrinogenemia
324588	Familial dyskinesia and facial myokymia
1885	Familial ectopia lentis
2762	Familial ectopic ossification
85110	Familial encephalopathy with neuroserpin inclusion bodies
101039	Familial epilepsy and mental retardation limited to females
90042	Familial erythrocytosis
99723	Familial esophageal achalasia
225968	Familial essential thrombocythemia
85195	Familial expansile osteolysis
891	Familial exudative vitreoretinopathy
98820	Familial focal epilepsy with variable foci
314022	Familial fundic gland polyposis with gastric cancer
26106	Familial gastric cancer
231040	Familial generalized lentiginosis
99819	Familial gestational hyperthyroidism
361	Familial glucocorticoid deficiency
3000	Familial gonadotropin-independent male-limited sexual precocity
540	Familial hemophagocytic lymphohistiocytosis
32960	Familial hibernian fever
540	Familial HLH
2604	Familial hollow visceral myopathy
403	Familial hyperaldosteronism type 1
404	Familial hyperaldosteronism type 2
251274	Familial hyperaldosteronism type 3
79506	Familial hyperalphalipoproteinemia
94086	Familial hypercalcemia - nephrocalcinosis - indicanuria
238475	Familial hypercholanemia
411	Familial hyperchylomicronemia
178345	Familial hyperestrogenism
757	Familial hyperkalemic hypertension
682	Familial hyperkalemic periodic paralysis
682	Familial hyperPP
99763	Familial hyperreninemic hypoaldosteronism type 1
99764	Familial hyperreninemic hypoaldosteronism type 2
424	Familial hyperthyroidism due to mutations in TSH receptor
413	Familial hypertriglyceridemia
427	Familial hypoaldosteronism
425	Familial hypoalphalipoproteinemia
405	Familial hypocalciuric hypercalcemia
93372	Familial hypocalciuric hypercalcemia type 1
101049	Familial hypocalciuric hypercalcemia type 2
101050	Familial hypocalciuric hypercalcemia type 3
248408	Familial hypodysfibrinogenemia
101041	Familial hypofibrinogenemia
31043	Familial hypomagnesemia - hypercalciuria - nephrocalcinosis
2196	Familial hypomagnesemia - hypercalciuria - nephrocalcinosis - severe ocular involvement
440	Familial hypospadias
225154	Familial IBSN
1677	Familial idiopathic dilatation of the right atrium
656	Familial idiopathic nephrotic syndrome
656	Familial idiopathic steroid-resistant nephrotic syndrome
93214	Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial proliferation
93217	Familial idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis
93213	Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental glomerulosclerosis
93213	Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis
93216	Familial idiopathic steroid-resistant nephrotic syndrome with minimal changes
225154	Familial infantile bilateral striatal necrosis
300373	Familial infantile gigantism
300547	Familial infantile hypercalcemia with suppressed intact parathyroid hormone
352582	Familial infantile myoclonic epilepsy
225154	Familial infantile striatonigral necrosis
2454	Familial intestinal malrotation - facial anomalies
2300	Familial intestinal polyatresia syndrome
217656	Familial isolated arrhythmogenic right ventricular cardiomyopathy
217656	Familial isolated arrhythmogenic right ventricular dysplasia
217656	Familial isolated arrhythmogenic ventricular cardiomyopathy
293899	Familial isolated arrhythmogenic ventricular cardiomyopathy, biventricular form

ORPHA Number	Disease name
293910	Familial isolated arrhythmogenic ventricular cardiomyopathy, classic form
293888	Familial isolated arrhythmogenic ventricular cardiomyopathy, left dominant form
293910	Familial isolated arrhythmogenic ventricular cardiomyopathy, right dominant form
217656	Familial isolated arrhythmogenic ventricular dysplasia
293899	Familial isolated arrhythmogenic ventricular dysplasia, biventricular form
293910	Familial isolated arrhythmogenic ventricular dysplasia, classic form
293888	Familial isolated arrhythmogenic ventricular dysplasia, left dominant form
293910	Familial isolated arrhythmogenic ventricular dysplasia, right dominant form
217656	Familial isolated ARVC
217656	Familial isolated ARVD
295014	Familial isolated clinodactyly of fingers
101351	Familial isolated congenital asplenia
154	Familial isolated dilated cardiomyopathy
99879	Familial isolated hyperparathyroidism
2238	Familial isolated hypoparathyroidism
2239	Familial isolated hypoparathyroidism due to agenesis of parathyroid gland
189466	Familial isolated hypoparathyroidism due to impaired PTH secretion
314777	Familial isolated pituitary adenoma
75249	Familial isolated restrictive cardiomyopathy
96	Familial isolated vitamin E deficiency
2295	Familial joint instability syndrome
2295	Familial joint laxity
180176	Familial juvenile gigantomastia
209886	Familial juvenile gouty nephropathy
180176	Familial juvenile hypertrophy of the breast
209886	Familial juvenile hyperuricemic nephropathy type 1
217330	Familial juvenile hyperuricemic nephropathy type 2
493	Familial keratoacanthoma
293936	Familial keratoconus with cataract
3267	Familial lambdoid synostosis
79293	Familial LCAT deficiency
523	Familial leiomyomatosis
523	Familial leiomyomatosis with renal carcinoma
523	Familial leiomyomatosis with renal cell cancer
231040	Familial lentigines profusa
309015	Familial lipoprotein lipase deficiency
768	Familial long QT syndrome
75381	Familial macular edema
342	Familial Mediterranean fever
99361	Familial medullary thyroid carcinoma
35858	Familial megaloblastic anemia
618	Familial melanoma
165805	Familial mesial temporal lobe epilepsy with febrile seizures
741	Familial mitral valve prolapse
276399	Familial MNG
99361	Familial MTC
276399	Familial multinodular goiter
35909	Familial multiple coagulation factor deficiency
523	Familial multiple cutaneous leiomyomas
338	Familial multiple fibrofolliculoma
500	Familial multiple lentigines syndrome
231040	Familial multiple lentigines syndrome without systemic involvement
199276	Familial multiple lipomatosis
263662	Familial multiple meningioma
867	Familial multiple trichoepithelioma
922	Familial nasal acilia
209886	Familial nephropathy with gout
424	Familial non-immune hyperthyroidism
144	Familial nonpolyposis colon cancer
144	Familial nonpolyposis colorectal cancer
88632	Familial ocular anterior segment mesenchymal dysgenesis
280403	Familial omphalocele syndrome with facial dysmorphism
154	Familial or idiopathic dilated cardiomyopathy
75249	Familial or idiopathic restrictive cardiomyopathy
569	Familial or sporadic hemiplegic migraine
251262	Familial osteochondritis dissecans
2769	Familial osteodysplasia, Anderson type
2801	Familial osteoectasia
86820	Familial osteonecrosis of the femoral head
1333	Familial pancreatic cancer
1333	Familial pancreatic carcinoma
319487	Familial papillary or follicular thyroid carcinoma
47044	Familial papillary renal cell carcinoma
97290	Familial papillary thyroid carcinoma with renal papillary neoplasia
99877	Familial parathyroid adenoma
99878	Familial parathyroids hyperplasia
2828	Familial Parkinson disease
97	Familial paroxysmal ataxia
98809	Familial paroxysmal kinesigenic dyskinesia
342	Familial paroxysmal polyserositis
228140	Familial paroxysmal ventricular fibrillation, not Brugada type
309	Familial partial epilepsy
98820	Familial partial epilepsy with variable foci
280356	Familial partial lipodystrophy associated with PLIN1 mutations
79083	Familial partial lipodystrophy associated with PPARG mutations
79085	Familial partial lipodystrophy due to AKT2 mutations
79084	Familial partial lipodystrophy type 1
2348	Familial partial lipodystrophy type 2
79083	Familial partial lipodystrophy type 3
2348	Familial partial lipodystrophy, Dunnigan type
79084	Familial partial lipodystrophy, Köbberling type
93333	Familial pelvis-scapular dysplasia
29072	Familial pheochromocytoma-paraganglioma
98809	Familial PKD
71290	Familial platelet disorder with associated myeloid malignancy
71290	Familial platelet syndrome
71290	Familial platelet syndrome with predisposition to acute myelogenous leukemia
330061	Familial polymorphous light eruption of American Indians
733	Familial polyposis coli
261584	Familial polyposis coli due to monosomy 5q22.2
99810	Familial porencephaly
2207	Familial primary hyperparathyroidism
34527	Familial primary hypomagnesemia with normocalcuria and normocalcemia
353220	Familial primary localized cutaneous amyloidosis
2257	Familial primary pulmonary hypoplasia
871	Familial progressive cardiac conduction defect
871	Familial progressive heart block
280628	Familial progressive hyper- and hypopigmentation
79146	Familial progressive hyperpigmentation
1767	Familial progressive vestibulocochlear dysfunction
1331	Familial prostate cancer
90044	Familial pseudohyperkalemia
100039	Familial pseudohyperkalemia type 1
100040	Familial pseudohyperkalemia type 2
100041	Familial pseudohyperkalemia, Cardiff type
275777	Familial pulmonary arterial hypertension
319487	Familial pure nonmedullary thyroid carcinoma
1675	Familial pyrimidinemia
79147	Familial reactive perforating collagenosis
46348	Familial rectal pain
69126	Familial recurrent arthritis

ORPHA Number	Disease name
2809	Familial recurrent peripheral facial palsy
85450	Familial renal amyloidosis
93560	Familial renal amyloidosis due to Apolipoprotein AI variant
238269	Familial renal amyloidosis due to Apolipoprotein AII variant
93562	Familial renal amyloidosis due to fibrinogen A alpha-chain variant
93561	Familial renal amyloidosis due to lysozyme variant
151	Familial renal cell carcinoma
69076	Familial renal glucosuria
99985	Familial restrictive cardiomyopathy type 1
99986	Familial restrictive cardiomyopathy type 2
218432	Familial restrictive cardiomyopathy type 3
284247	Familial retinal arterial macroaneurysm
357027	Familial retinoblastoma
231108	Familial rhabdoid tumor
254712	Familial Rosaï-Dorfman disease
171839	Familial scaphocephaly - radioulnar synostosis
168624	Familial scaphocephaly syndrome, McGillivray type
3135	Familial Scheuermann disease
3135	Familial Scheuermann juvenile kyphosis
79428	Familial segmental neurofibromatosis
254712	Familial SHML
51083	Familial short QT syndrome
254712	Familial sinus histiocytosis with massive lymphadenopathy
300345	Familial SLE
685	Familial spastic paraplegia
79429	Familial spinal neurofibromatosis
3135	Familial spinal osteochondrosis
2903	Familial spontaneous pneumothorax
3197	Familial startle disease
280406	Familial steroid-resistant nephrotic syndrome with sensorineural deafness
26106	Familial stomach cancer
2456	Familial supernumerary nipples
2398	Familial symmetric lipomatosis
300345	Familial systemic lupus erythematosus
91387	Familial TAAD
98819	Familial temporal epilepsy
91387	Familial thoracic aortic aneurysm and aortic dissection
71493	Familial thrombocythemia
71493	Familial thrombocytosis
329319	Familial thrombocytosis with transverse limb defect
3324	Familial thrombomodulin anomalies
93953	Familial thyroglossal duct cyst
95716	Familial thyroid dyshormonogenesis
53372	Familial trembling of the chin
36383	Familial vascular leukoencephalopathy
289365	Familial vesicoureteral reflux
637	Familial vestibular schwannoma
2604	Familial visceral myopathy
2808	Familial vocal cord dysfunction
289365	Familial VUR
170	Familial woolly hair syndrome
170	Familial wooly hair syndrome
84	Fanconi anemia
84	Fanconi pancytopenia
2088	Fanconi-Bickel disease
163654	Fantasy Island syndrome
733	FAP
261584	FAP due to monosomy 5q22.2
2792	Fara-Chlupackova syndrome
333	Farber disease
333	Farber lipogranulomatosis
99906	Farmer's lung disease
3261	FAS deficiency
164736	FASPS
166105	FASTKD2-related infantile mitochondrial encephalomyopathy
466	Fatal familial insomnia
1561	Fatal infantile cardioencephalomyopathy due to cytochrome c oxidase deficiency
1561	Fatal infantile COX deficiency
1561	Fatal infantile cytochrome C oxidase deficiency
166073	Fatal infantile encephalopathy with mitochondrial respiratory chain defects
166063	Fatal infantile encephalopathy with olivopontocerebellar hypoplasia
293838	Fatal infantile encephalopathy-pulmonary hypertension syndrome
289527	Fatal infantile HCM due to mitochondrial complex I deficiency
280553	Fatal infantile hypertonic myofibrillar myopathy
289527	Fatal infantile hypertrophic cardiomyopathy due to mitochondrial complex I deficiency
289527	Fatal infantile hypertrophic cardiomyopathy due to NADH-coenzyme Q reductase deficiency
289527	Fatal infantile hypertrophic cardiomyopathy due to NADH-CoQ reductase deficiency
17	Fatal infantile lactic acidosis with methylmalonic aciduria
168566	Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3
289573	Fatal multiple mitochondrial dysfunction syndrome
816	Fatty acid alcohol oxidoreductase deficiency
329308	Fatty acid hydroxylase-associated neurodegeneration
2064	Faulk-Epstein-Jones syndrome
405	FBH
405	FBHH
47045	FCAS
47045	FCAS1
247868	FCAS2
98970	FCD
268961	FCD type I
268973	FCD type Ia
268980	FCD type Ib
268987	FCD type Ic
268994	FCD type II
269001	FCD type IIa
269008	FCD type IIb
86814	FCMTE
3071	FCS syndrome
47045	FCU
324	FD
324588	FDFM
163703	Febrile infection-related epilepsy syndrome
98974	FECD
79292	FED
247165	Feer disease
98969	Fehr corneal dystrophy
1192	Feigenbaum-Bergeron-Richardson syndrome
1305	Feingold syndrome
53693	Fellman disease
47612	Felty syndrome
2973	Female pseudohermaphrodism - anorectal anomalies
2975	Female pseudohermaphroditism - skeletal anomalies
101039	Female restricted epilepsy with intellectual deficit
1987	Femoral agenesis/hypoplasia
295067	Femoral agenesis/hypoplasia, bilateral
295065	Femoral agenesis/hypoplasia, unilateral
1988	Femoral hypoplasia - unusual facies syndrome
1987	Femoral intercalary meromelia
295067	Femoral intercalary meromelia, bilateral
295065	Femoral intercalary meromelia, unilateral
1863	Femoral trochlear groove insufficiency
1988	Femoral-facial syndrome
294977	Femorotibiofibular intercalary transverse meromelia
295091	Femorotibiofibular intercalary transverse meromelia, bilateral
295089	Femorotibiofibular intercalary transverse meromelia, unilateral

ORPHA Number	Disease name
2019	Femur-fibula-ulna complex
2019	Femur-fibula-ulna dysostosis
2019	Femur-fibula-ulna syndrome
85110	FENIB
1184	Fenton-Wilkinson-Toselano syndrome
45358	FEOM
2180	Ferlini-Ragno-Calzolari syndrome
157846	Ferritin-related neurodegeneration
139491	Ferroportin disease
40366	Fetal acitretin syndrome
994	Fetal akinesia deformation sequence
1915	Fetal alcohol syndrome
1908	Fetal aminopterin syndrome
853	Fetal and neonatal alloimmune thrombocytopenia
1665	Fetal brain disruption sequence
1911	Fetal cocaine syndrome
294	Fetal cytomegalovirus syndrome
1912	Fetal dihydantoin syndrome
1041	Fetal edema
97360	Fetal face syndrome
85212	Fetal Gaucher disease
1912	Fetal hydantoin syndrome
1909	Fetal indomethacin syndrome
1910	Fetal iodine syndrome
1055	Fetal left ventricular aneurysm
284362	Fetal lung interstitial tumor
1917	Fetal methylmercury syndrome
1918	Fetal minoxidil syndrome
295	Fetal parvovirus syndrome
3312	Fetal thalidomide syndrome
1913	Fetal trimethadione syndrome
1906	Fetal valproate syndrome
1906	Fetal valproic acid syndrome
291	Fetal varicella syndrome
1914	Fetal warfarin syndrome
166068	Fetal-onset olivopontocerebellar hypoplasia
95431	Feto-fetal transfusion syndrome
163703	Fever-induced refractory epileptic encephalopathy in school-aged children
891	FEVR
254492	FFA
79133	FFDD type I
98820	FFEVF
1988	FFS
2019	FFU complex
93932	FG syndrome type 1
313855	FGFR2-related bent bone dysplasia
403	FH1
404	FH2
254707	FHC
405	FHH
93372	FHH type 1
101049	FHH type 2
101050	FHH type 3
99763	FHHA1
99764	FHHA2
263479	FHI
1988	FHUFS
251601	Fibrillary astrocytoma
93562	Fibrinogen A alpha-chain amyloidosis
331	Fibrin-stabilizing factor deficiency
99654	Fibrocalculous pancreatopathy
2021	Fibrochondrogenesis
337	Fibrodysplasia ossificans progressiva
122	Fibrofolliculomas with trichodiscomas and acrochordons
336	Fibromuscular dysplasia of arteries
79105	Fibromyxosarcoma
84090	Fibronectin glomerulopathy
2030	Fibrosarcoma
63999	Fibrosing mediastinitis
249	Fibrous dysplasia of bone
2639	Fibular aplasia - complex brachydactyly
1118	Fibular aplasia - ectrodactyly
1757	Fibular dimelia - diplopodia
93323	Fibular hemimelia
295083	Fibular hemimelia, bilateral
295081	Fibular hemimelia, unilateral
2854	Fibular hypoplasia or aplasia - femoral bowing - oligodactyly
295083	Fibular longitudinal meomelia, bilateral
295081	Fibular longitudinal meomelia, unilateral
2256	Fibulo-ulnar hypoplasia - renal anomalies
79306	FIC1 deficiency
29207	Fiessinger-Leroy disease
29207	Fiessinger-Leroy-Reiter syndrome
2756	Figuera syndrome
99879	FIHPT
2034	Filariasis
3255	Filippi syndrome
352712	FILS syndrome
352582	FIME
1272	Fine-Lubinsky syndrome
97232	Fingerprint body myopathy
209335	Finkel disease
2036	Finlay-Marks syndrome
839	Finnish congenital nephrosis
1825	Finucane-Kurtz-Scott syndrome
314777	FIPA
163703	FIRES
141136	First branchial arch syndrome
141013	First branchial cleft anomaly
141013	First branchial cleft cyst
141013	First branchial cleft fistula
98919	Fisher syndrome
79292	Fish-eye disease
35056	Fish-odor syndrome
840	Fistulous vegetative verrucous hydradenoma
2823	Fitzsimmons-Guilbert syndrome
2824	Fitzsimmons-McLachlan-Gilbert syndrome
2820	Fitzsimmons-Walson-Mellor syndrome
293812	Fixed pigmented erythema
3092	Fixed subaortic stenosis
209886	FJHN type 1
217330	FJHN type 2
1968	Flat face - microstomia - ear anomaly
79293	FLD
98970	Fleck corneal dystrophy
409	Flegel disease
284362	FLIT
2044	Floating-Harbor syndrome
83451	Florid cemento-osseous dysplasia
83451	Florid osseous dysplasia
2045	FLOTCH syndrome
99734	Fluctuating myotonia
2047	Flynn-Aird syndrome
342	FMF
276399	FMNG
319487	FNMTC
137675	Foamy myocardial transformation of infancy
2143	FOAR syndrome
308013	Focal acral hyperkeratosis
83451	Focal cemento-osseous dysplasia
2092	Focal dermal hypoplasia
1866	Focal dystonia
352587	Focal epilepsy - intellectual deficit - cerebro-cerebellar malformation
352587	Focal epilepsy - intellectual deficit - dysarthria - ataxia
79133	Focal facial dermal dysplasia
221083	Focal myoclonus of face
48918	Focal myositis
48918	Focal nodular myositis
2200	Focal palmoplantar and gingival hyperkeratosis
2200	Focal palmoplantar and gingival keratoderma
79093	Foix-Alajouanine syndrome
2048	Foix-Chavany-Marie syndrome
79097	Folinic acid-responsive seizures
113	Follicular atrophoderma and basal cell carcinomas

ORPHA Number	Disease name
79459	Follicular atrophoderma-basal cell carcinoma
300552	Follicular cholangitis and pancreatitis
86902	Follicular dendritic cell sarcoma
69745	Follicular dyskeratoma
2112	Follicular hamartoma - alopecia - cystic fibrosis
525	Follicular lichen planus
545	Follicular lymphoma
300552	Follicular pancreatocholangitis
243	Follicular stimulating hormone-resistant ovaries
79100	Folliculitis ulerythematosa reticulate
178512	Folliculotropic mycosis fungoides
228371	Foodborne botulism
337	FOP
60015	Foramina parietalia permagna
366	Forbe disease
141071	Foregut duplication cyst of the tongue
51208	Formiminoglutamic aciduria
51208	Formiminotransferase cyclodeaminase deficiency
3238	Forney syndrome
3238	Forney-Robinson-Pascoe syndrome
178333	Forsius-Eriksson syndrome
178333	Forsius-Eriksson type ocular albinism
85162	FOSMN syndrome
3219	Fountain syndrome
141037	Fourth branchial cleft anomaly
141037	Fourth branchial cleft cyst
141037	Fourth branchial cleft fistula
2253	Foveal hypoplasia - presenile cataract
221126	Fowler syndrome
2795	Fowler-Christmas-Chapple syndrome
1799	FOXP2-associated dysphasia
275777	FPAH
71290	FPD/AML syndrome
280628	FPHH
353220	FPLCA
79084	FPLD1
2348	FPLD2
79083	FPLD3
71290	FPS/AML syndrome
69126	FRA
908	Fragile X syndrome
93256	Fragile X-associated tremor/ataxia syndrome
284247	FRAM
861	Franceschetti-Klein syndrome
2108	François dyscephalic syndrome
79149	François syndrome
98970	François-Neetens speckled corneal dystrophy
2523	Franek-Bocker-Kahlen syndrome
100026	Franklin disease
137834	Frank-Ter Haar syndrome
2052	Fraser syndrome
2051	Fraser-like syndrome
347	Frasier syndrome
908	FraX syndrome
908	FRAXA syndrome
100973	FRAXE intellectual deficit
100974	FRAXF syndrome
834	Free sialic acid storage disease
309324	Free sialic acid storage disease, infantile form
2053	Freeman-Sheldon syndrome
1147	Freeman-Sheldon syndrome variant
2673	Freire Maia-Pinheiro-Opitz syndrome
2723	Freire-Maia syndrome
70472	French-Canadian type COX deficiency
70472	French-Canadian type cytochrome C oxidase deficiency
70472	French-Canadian type Leigh syndrome
2055	Frias syndrome
85335	Fried syndrome
2487	Fried-Goldberg-Mundel syndrome
1969	Friedman-Goodman syndrome
95	Friedreich ataxia
96	Friedreich-like ataxia
99672	Fried's tooth and nail syndrome
1931	Frontal encephalocele
254492	Frontal fibrosing alopecia
1791	Fronto-facio-nasal dysostosis
1791	Fronto-facio-nasal dysplasia
1826	Frontometaphyseal dysplasia
141168	Frontonasal arteriovenous malformation
250	Frontonasal dysplasia
228390	Frontonasal dysplasia with alopecia and genital abnomality
228390	Frontonasal dysplasia with alopecia and genital anomaly
306542	Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome
282	Frontotemporal dementia
275872	Frontotemporal dementia with amyotrophic lateral sclerosis
275872	Frontotemporal dementia with motor neuron disease
293848	Frontotemporal dementia, right temporal atrophy variant
2141	Froster-Huch syndrome
2215	Froster-Iskenius-Waterson syndrome
2056	Fructokinase deficiency
348	Fructose-1,6-bisphosphatase deficiency
2057	Frydman-Cohen-Karmon syndrome
2429	Fryns macrocephaly
1104	Fryns microphthalmia syndrome
2059	Fryns syndrome
94084	Fryns-Aftimos syndrome
2497	Fryns-Hofkens-Fabry syndrome
2058	Fryns-Smeets-Thiry syndrome
269	FSH dystrophy
269	FSHD
243	FSH-RO
51208	FTCD deficiency
282	FTD
275872	FTD-ALS
275872	FTD-MND
247790	FTH1-associated iron overload
247790	FTH1-related iron overload
98974	Fuchs endothelial corneal dystrophy
263479	Fuchs heterochromic iridocyclitis
349	Fucosidosis
2854	Fuhrmann syndrome
2854	Fuhrmann-Rieger-de Sousa syndrome
2060	Fukuda-Miyanomae-Nakata syndrome
551	Fukuhara syndrome
35063	Fulminant viral hepatitis
24	Fumarase deficiency
24	Fumaric aciduria
882	Fumaryl acetoacetase deficiency
882	Fumaryl acetoacetate hydrolase deficiency
622	Functional methionine synthase deficiency
308380	Functional methionine synthase deficiency type cblDv1
2169	Functional methionine synthase deficiency type cblE
2170	Functional methionine synthase deficiency type cblG
91348	Functioning gonadotroph adenoma
91348	Functioning pituitary gonadotrophic adenoma
227796	Fundus albipunctatus
827	Fundus flavimaculatus
99004	Fundus pulverulentus
2579	Furukawa-Takagi-Nakao syndrome
591	Furuncular myiasis
591	Furunculoid myiasis
591	Furunculous myiasis
228119	Fusariosis
228119	Fusarium infection
2287	Fused mandibular incisors
2498	Fusion of metacarpals 4 and 5
35909	FV and FVIII combined deficiency
908	FXS

ORPHA Number	Disease name
93256	FXTAS syndrome
364	G6P deficiency
79258	G6P deficiency type a
79259	G6P deficiency type b
79259	G6P translocase deficiency
79259	G6PT deficiency
25	GA1
2066	GABA transaminase deficiency
79402	GABEB
90041	Gaisböck syndrome
487	Galactocerebrosidase deficiency
79237	Galactokinase deficiency
79237	Galactokinase deficiency galactosemia
309297	Galactosamine-6-sulfatase deficiency
79238	Galactose epimerase deficiency
79239	Galactose-1-phosphate uridyltransferase deficiency
352	Galactosemia
79239	Galactosemia type 1
79237	Galactosemia type 2
79238	Galactosemia type 3
351	Galactosialidosis
487	Galactosylceramidase deficiency
75496	Galactosyltransferase I deficiency
487	GALC deficiency
79238	GALE deficiency
79238	GALE-D
79237	GALK deficiency
79237	GALK-D
56044	Gallbladder cancer
100086	Gall-bladder endocrine tumor
2065	Galloway syndrome
2065	Galloway-Mowat syndrome
309297	GALNS deficiency
306661	GALNT3-CDG syndrome
79239	GALT deficiency
2325	Gamborg-Nielsen syndrome
3035	Game-Friedman-Paradice syndrome
100026	Gamma heavy-chain disease
2066	Gamma-aminobutyric acid transaminase deficiency
212	Gamma-cystathionase deficiency
33573	Gamma-glutamyl transpeptidase deficiency
33574	Gamma-glutamylcysteine synthetase deficiency
100026	Gamma-HCD
353	Gamma-sarcoglycanopathy
682	Gamstorp disease
682	Gamstorp episodic adynamy
382	GAMT deficiency
251937	Gangliocytoma
251949	Ganglioglioma
251877	Ganglioneuroblastoma
251992	Ganglioneuroma
2067	GAPO syndrome
314022	GAPPS
3469	Garcia-Lurie syndrome
79665	Gardner syndrome
324636	Gardner-Diamond syndrome
2075	Gardner-Silengo-Wachtel syndrome
314022	Gastric adenocarcinoma and proximal polyposis of the stomach
63443	Gastric cancer
141071	Gastric duplication cyst of the tongue
100075	Gastric endocrine tumor
332	Gastric intrinsic factor deficiency
36273	Gastric linitis plastica
913	Gastrinoma
2069	Gastrocutaneous syndrome
2930	Gastrointestinal polyposis - ectodermal changes
2930	Gastrointestinal polyposis - skin pigmentation - alopecia - fingernail changes
44890	Gastrointestinal stromal tumor
2368	Gastroschisis
355	Gaucher disease
2072	Gaucher disease - ophthalmoplegia - cardiovascular calcification
77259	Gaucher disease type 1
77260	Gaucher disease type 2
77261	Gaucher disease type 3
2072	Gaucher disease type 3C
77261	Gaucher disease, subacute neuronopathic type
2072	Gaucher-like disease
56044	GBC
308712	GBE deficiency, adult neuromuscular form
308684	GBE deficiency, childhood combined hepatic and myopathic form
308698	GBE deficiency, childhood neuromuscular form
308670	GBE deficiency, congenital neuromuscular form
308655	GBE deficiency, fatal perinatal neuromuscular form
308638	GBE deficiency, non progressive hepatic form
308621	GBE deficiency, progressive hepatic form
360	GBM
2103	GBS
98916	GBS, acute inflammatory demyelinating polyradiculoneuropathic form
329984	GCC
98962	GCD1
98963	GCD2
25	GCDHD
98962	GCDI
98963	GCDII
528	GCL
228429	GCL4
380	GCPS
79330	GCS1-CDG syndrome
98957	GDCD
53697	GDD
366	GDE deficiency
324636	GDS
36387	GEFS+
26790	Gelatinous ascites
98957	Gelatinous drop-like corneal dystrophy
2623	Geleophysic dwarfism
2623	Geleophysic dysplasia
2073	Gélineau disease
85448	Gelsolin amyloidosis
2074	Gemignani syndrome
251604	Gemistocytic astrocytoma
2084	Gemss syndrome
51608	Generalized arterial calcification of infancy
79402	Generalized atrophic benign epidermolysis bullosa
168632	Generalized basaloid follicular hamartoma syndrome
528	Generalized congenital lipodystrophy
228429	Generalized congenital lipodystrophy type 4
228429	Generalized congenital lipodystrophy with myopathy
263543	Generalized deciduous skin
263548	Generalized deciduous skin type A
263553	Generalized deciduous skin type B
263558	Generalized deciduous skin type C
231568	Generalized dominant dystrophic epidermolysis bullosa
79399	Generalized EBS, non-Dowling-Meara type
79399	Generalized epidermolysis bullosa simplex, non-Dowling-Meara type
79137	Generalized epilepsy - paroxysmal dyskinesia
99649	Generalized epilepsy and praxis-induced seizures
36387	Generalized epilepsy with febrile seizures-plus context
308487	Generalized epimerase deficiency galactosemia
157991	Generalized eruptive histiocytoma
157991	Generalized eruptive histiocytosis
280774	Generalized essential telangiectasia
36236	Generalized exfoliative disease

ORPHA Number	Disease name
308487	Generalized galactose epimerase deficiency
308487	Generalized GALE deficiency
308487	Generalized GALE-D
33355	Generalized hematopoietic hypoplasia
79402	Generalized junctional epidermolysis bullosa, non-Herlitz type
329971	Generalized juvenile polyposis/juvenile polyposis coli
167635	Generalized lichenoid papular eruption
89842	Generalized mitis RDEB
167635	Generalized papular and sclerodermoid lichen myxedematosus
263543	Generalized peeling skin syndrome
263548	Generalized peeling skin syndrome type A
263553	Generalized peeling skin syndrome type B
263558	Generalized peeling skin syndrome type C
171876	Generalized pseudohypoaldosteronism type 1
247353	Generalized pustular psoriasis
3221	Generalized resistance to thyroid hormone
308487	Generalized UDP-galactose-4-epimerase deficiency
308487	Generalized uridine diphosphate galactose-4-epimerase deficiency
183450	Genetic hair anomaly
254704	Genetic hyperferritinemia without iron overload
99845	Genetic recurrent myoglobinuria
226316	Genetic transient congenital hypothyroidism
182734	Genetic urticaria
2075	Genito-palato-cardiac syndrome
85201	Genitopatellar syndrome
2163	Genoa syndrome
85197	Genochondromatosis
93398	Genochondromatosis type 2
329813	Genome-wide paternal uniparental disomy mosaicism
1454	Gentile syndrome
217008	Genuine diffuse phlebectasia
98961	Geographic corneal dystrophy
35686	Geographic helicoid peripapillary choroidopathy
79137	GEPD
99095	Gerbode defect
2808	Gerhardt syndrome
213837	Germ cell cancer of the cervix uteri
213751	Germ cell cancer of the corpus uteri
2077	German syndrome
91352	Germinoma of the central nervous system
2078	Geroderma osteodysplastica
1117	Gershoni-Baruch-Leibo syndrome

ORPHA Number	Disease name
221117	Gerstmann syndrome
356	Gerstmann-Straussler-Scheinker syndrome
99926	Gestational choriocarcinoma
59305	Gestational trophoblastic neoplasm
280774	GET
84090	GFND
314769	GH and PRL co-secreting pituitary adenoma
633	GH receptor deficiency
1802	Ghosal hematodiaphyseal dysplasia
1802	Ghosal syndrome
83450	Ghost teeth
180267	Giant adenofibroma of the breast
643	Giant axonal neuropathy
397	Giant cell arteritis
1190	Giant cell chondrodysplasia
251579	Giant cell glioblastoma
139436	Giant cell histiocytomatosis
626	Giant congenital melanocytic nevus
2494	Giant hypertrophic gastritis
210592	Giant infantile hemangioma
626	Giant pigmented hairy nevus
274	Giant platelet syndrome
99725	Gigantism
1065	Gillespie syndrome
2025	Gingival fibromatosis - facial dysmorphism
3473	Gingival fibromatosis - hepatosplenomegaly - other anomalies
2026	Gingival fibromatosis - hypertrichosis
2027	Gingival fibromatosis - progressive deafness
2709	Gingival hypertrophy - corneal dystrophy
44890	GIST
97286	GIST-paraganglioma dyad
358	Gitelman syndrome
3268	Giuffré-Tsukahara syndrome
849	Glanzmann thrombasthenia
666	Glass bone disease
1535	Glass-Chapman-Hockley syndrome
213833	Glassy cell carcinoma of the cervix uteri
2084	Glaucoma - ectopia - microspherophakia - stiff joints - short stature
2085	Glaucoma - sleep apnea
238763	Glaucoma secondary to spherophakia/ ectopia lentis and megalocornea
354	GLB1 deficiency
360	Glioblastoma
360	Glioblastoma multiforme
269197	Glioependymal/ependymal cyst
251582	Gliomatosis cerebri
251576	Gliosarcoma

ORPHA Number	Disease name
73223	Global developmental delay - osteopenia - ectodermal defect
2791	Globodontia
487	Globoid cell leukodystrophy
171709	Globozoospermia
83454	Glomangiomatosis
2087	Glomerulonephritis - sparse hair - telangiectasis
84090	Glomerulopathy with fibronectin deposits
83454	Glomuvenous malformation
2616	Gloomy syndrome
141163	Glossopalatine ankylosis
221098	Glossopharyngeal neuralgia
221098	Glossovasopharyngeal neuralgia
97280	Glucagonoma
355	Glucocerebrosidase deficiency
786	Glucocorticoid resistance
403	Glucocorticoid sensitive hypertension
403	Glucocorticoid-remediable aldosteronism
79272	Glucosamine N-acetyl-6-sulfatase deficiency
71277	Glucose transporter type 1 deficiency
35710	Glucose-galactose malabsorption
79330	Glucosidase 1 deficiency
79320	Glucosyltransferase 1 deficiency
79325	Glucosyltransferase 2 deficiency
71277	Glut-1 deficiency Syndrome
71277	Glut1-DS
3006	Glutamate decarboxylase deficiency
51208	Glutamate formiminotransferase deficiency
2195	Glutamate-aspartate transport defect
33574	Glutamate-cysteine ligase deficiency
25	Glutaric acidemia type 1
26791	Glutaric acidemia type 2
35706	Glutaric acidemia type 3
25	Glutaric aciduria type 1
26791	Glutaric aciduria type 2
35706	Glutaric aciduria type 3
25	Glutaryl-CoA dehydrogenase deficiency
35706	Glutaryl-CoA oxidase deficiency
25	Glutaryl-coenzyme A dehydrogenase deficiency
32	Glutathione synthetase deficiency
289846	Glutathione synthetase deficiency with 5-oxoprolinuria
289849	Glutathione synthetase deficiency without 5-oxoprolinuria
33573	Glutathionuria
284414	Glycerol kinase deficiency, adult form
284408	Glycerol kinase deficiency, infantile form
284411	Glycerol kinase deficiency, juvenile form

ORPHA Number	Disease name
261476	Glycerol kinase deficiency-contiguous gene syndrome
255182	Glycine cleavage system L protein deficiency
407	Glycine encephalopathy
289891	Glycine N-methyltransferase deficiency
365	Glycogen storage disease due to acid maltase deficiency
308604	Glycogen storage disease due to acid maltase deficiency, adult onset
308552	Glycogen storage disease due to acid maltase deficiency, infantile onset
308573	Glycogen storage disease due to acid maltase deficiency, juvenile onset
57	Glycogen storage disease due to aldolase A deficiency
364	Glycogen storage disease due to G6P deficiency
79258	Glycogen storage disease due to G6P deficiency type a
79259	Glycogen storage disease due to G6P deficiency type b
364	Glycogen storage disease due to glucose-6-phosphatase deficiency
79258	Glycogen storage disease due to glucose-6-phosphatase deficiency type a
79259	Glycogen storage disease due to glucose-6-phosphatase deficiency type b
2088	Glycogen storage disease due to GLUT2 deficiency
367	Glycogen storage disease due to glycogen branching enzyme deficiency
308712	Glycogen storage disease due to glycogen branching enzyme deficiency, adult neuromuscular form
308684	Glycogen storage disease due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form
308698	Glycogen storage disease due to glycogen branching enzyme deficiency, childhood neuromuscular form
308670	Glycogen storage disease due to glycogen branching enzyme deficiency, congenital neuromuscular form
308655	Glycogen storage disease due to glycogen branching enzyme deficiency, fatal perinatal neuromuscular form
308638	Glycogen storage disease due to glycogen branching enzyme deficiency, non progressive hepatic form
308621	Glycogen storage disease due to glycogen branching enzyme deficiency, progressive hepatic form
366	Glycogen storage disease due to glycogen debranching enzyme deficiency
263297	Glycogen storage disease due to glycogenin deficiency
2089	Glycogen storage disease due to hepatic glycogen synthase deficiency

ORPHA Number	Disease name
2364	Glycogen storage disease due to lactate dehydrogenase deficiency
284435	Glycogen storage disease due to lactate dehydrogenase H-subunit deficiency
284426	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency
34587	Glycogen storage disease due to LAMP-2 deficiency
79240	Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency
369	Glycogen storage disease due to liver glycogen phosphorylase deficiency
2089	Glycogen storage disease due to liver glycogen synthase deficiency
264580	Glycogen storage disease due to liver phosphorylase kinase deficiency
137625	Glycogen storage disease due to muscle and heart glycogen synthase deficiency
99849	Glycogen storage disease due to muscle beta-enolase deficiency
368	Glycogen storage disease due to muscle glycogen phosphorylase deficiency
371	Glycogen storage disease due to muscle phosphofructokinase deficiency
715	Glycogen storage disease due to muscle phosphorylase kinase deficiency
370	Glycogen storage disease due to PhK deficiency
711	Glycogen storage disease due to phosphoglucomutase deficiency
713	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency
97234	Glycogen storage disease due to phosphoglycerate mutase deficiency
370	Glycogen storage disease due to phosphorylase kinase deficiency
2089	Glycogen storage disease type 0a
137625	Glycogen storage disease type 0b
364	Glycogen storage disease type 1
79258	Glycogen storage disease type 1a
79259	Glycogen storage disease type 1b
365	Glycogen storage disease type 2
308604	Glycogen storage disease type 2, adult onset
308552	Glycogen storage disease type 2, infantile onset
308573	Glycogen storage disease type 2, juvenile onset
366	Glycogen storage disease type 3
367	Glycogen storage disease type 4
308712	Glycogen storage disease type 4, adult neuromuscular form
308684	Glycogen storage disease type 4, childhood combined hepatic and myopathic form
308698	Glycogen storage disease type 4, childhood neuromuscular form

ORPHA Number	Disease name
308670	Glycogen storage disease type 4, congenital neuromuscular form
308655	Glycogen storage disease type 4, fatal perinatal neuromuscular form
308638	Glycogen storage disease type 4, non progressive hepatic form
308621	Glycogen storage disease type 4, progressive hepatic form
368	Glycogen storage disease type 5
369	Glycogen storage disease type 6B
371	Glycogen storage disease type 7
370	Glycogen storage disease type 9
264580	Glycogen storage disease type 9A
79240	Glycogen storage disease type 9B
264580	Glycogen storage disease type 9C
715	Glycogen storage disease type 9D
715	Glycogen storage disease type 9E
284426	Glycogen storage disease type 11
57	Glycogen storage disease type 12
711	Glycogen storage disease type 14
263297	Glycogen storage disease type 15
370	Glycogen storage disease type IX
264580	Glycogen storage disease type IXa
79240	Glycogen storage disease type IXb
264580	Glycogen storage disease type IXc
715	Glycogen storage disease type IXd
715	Glycogen storage disease type IXe
263297	Glycogen storage disease type XV
365	Glycogenosis due to acid maltase deficiency
308604	Glycogenosis due to acid maltase deficiency, adult onset
308552	Glycogenosis due to acid maltase deficiency, infantile onset
308573	Glycogenosis due to acid maltase deficiency, juvenile onset
57	Glycogenosis due to aldolase A deficiency
79258	Glycogenosis due to glucose-6-phosphatase deficiency type a
79259	Glycogenosis due to glucose-6-phosphatase deficiency type b
79259	Glycogenosis due to glucose-6-phosphatase transport defect
2088	Glycogenosis due to GLUT2 deficiency
367	Glycogenosis due to glycogen branching enzyme deficiency
308712	Glycogenosis due to glycogen branching enzyme deficiency, adult neuromuscular form
308684	Glycogenosis due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form
308698	Glycogenosis due to glycogen branching enzyme deficiency, childhood neuromuscular form

ORPHA Number	Disease name
308670	Glycogenosis due to glycogen branching enzyme deficiency, congenital neuromuscular form
308655	Glycogenosis due to glycogen branching enzyme deficiency, fatal perinatal neuromuscular form
308638	Glycogenosis due to glycogen branching enzyme deficiency, non progressive hepatic form
308621	Glycogenosis due to glycogen branching enzyme deficiency, progressive hepatic form
366	Glycogenosis due to glycogen debranching enzyme deficiency
263297	Glycogenosis due to glycogenin deficiency
2364	Glycogenosis due to lactate dehydrogenase deficiency
284435	Glycogenosis due to lactate dehydrogenase H-subunit deficiency
284426	Glycogenosis due to lactate dehydrogenase M-subunit deficiency
34587	Glycogenosis due to LAMP-2 deficiency
79240	Glycogenosis due to liver and muscle phosphorylase kinase deficiency
369	Glycogenosis due to liver glycogen phosphorylase deficiency
264580	Glycogenosis due to liver phosphorylase kinase deficiency
137625	Glycogenosis due to muscle and heart glycogen synthase deficiency
99849	Glycogenosis due to muscle beta-enolase deficiency
368	Glycogenosis due to muscle glycogen phosphorylase deficiency
371	Glycogenosis due to muscle phosphofructokinase deficiency
715	Glycogenosis due to muscle phosphorylase kinase deficiency
711	Glycogenosis due to phosphoglucomutase deficiency
713	Glycogenosis due to phosphoglycerate kinase 1 deficiency
97234	Glycogenosis due to phosphoglycerate mutase deficiency
370	Glycogenosis due to phosphorylase kinase deficiency
2089	Glycogenosis type 0a
137625	Glycogenosis type 0b
364	Glycogenosis type 1
365	Glycogenosis type 2
308604	Glycogenosis type 2, adult onset
308552	Glycogenosis type 2, infantile onset
308573	Glycogenosis type 2, juvenile onset
366	Glycogenosis type 3
367	Glycogenosis type 4
308712	Glycogenosis type 4, adult neuromuscular form
308684	Glycogenosis type 4, childhood combined hepatic and myopathic form
308698	Glycogenosis type 4, childhood neuromuscular form
308670	Glycogenosis type 4, congenital neuromuscular form
308655	Glycogenosis type 4, fatal perinatal neuromuscular form
308638	Glycogenosis type 4, non progressive hepatic form
308621	Glycogenosis type 4, progressive hepatic form
368	Glycogenosis type 5
369	Glycogenosis type 6B
371	Glycogenosis type 7
370	Glycogenosis type 9
264580	Glycogenosis type 9A
79240	Glycogenosis type 9B
264580	Glycogenosis type 9C
715	Glycogenosis type 9D
715	Glycogenosis type 9E
284426	Glycogenosis type 11
57	Glycogenosis type 12
99849	Glycogenosis type 13
711	Glycogenosis type 14
263297	Glycogenosis type 15
79258	Glycogenosis type Ia
79259	Glycogenosis type Ib
370	Glycogenosis type IX
264580	Glycogenosis type IXa
79240	Glycogenosis type IXb
264580	Glycogenosis type IXc
715	Glycogenosis type IXd
715	Glycogenosis type IXe
263297	Glycogenosis type XV
93598	Glycolic aciduria
354	GM1 gangliosidosis
79255	GM1 gangliosidosis type 1
79256	GM1 gangliosidosis type 2
79257	GM1 gangliosidosis type 3
309152	GM2 gangliosidosis
796	GM2 gangliosidosis 0 variant
309246	GM2-gangliosidosis, AB variant
309192	GM2-gangliosidosis, B variant, adult form
309178	GM2-gangliosidosis, B variant, infantile form
309185	GM2-gangliosidosis, B variant, juvenile form
845	GM2-gangliosidosis, B, B1 variant
309239	GM2-gangliosidosis, B1 variant
171714	GM3 synthase deficiency
626	GMN
2090	GMS syndrome
53697	Gnathodiaphyseal dysplasia
79272	GNS deficiency
329984	Goblet cell adenocarcinoid
329984	Goblet cell carcinoid
329984	Goblet cell carcinoma
329984	Goblet cell tumor
705	Goiter - deafness
373	Golabi-Rosen syndrome
351	Goldberg syndrome
754	Goldberg-Maxwell syndrome
66629	Goldberg-Shprintzen megacolon syndrome
166272	Goldblatt chondrodysplasia
166272	Goldblatt syndrome
3026	Goldblatt-Viljoen syndrome
2261	Goldblatt-Wallis syndrome
374	Goldenhar syndrome
53540	Goldmann-Favre syndrome
3362	Goldstein-Hutt syndrome
3032	Goldston syndrome
1791	Gollop syndrome
1986	Gollop-Wolfgang complex
2092	Goltz syndrome
2092	Goltz-Gorlin syndrome
1770	Gonadal dysgenesis, XY type - associated anomalies
432	Gonadotropic deficiency
759	Gonadotropin-dependant precocious puberty
562	Gonadotropin-independent female-limited sexual precocity
2090	Goniodysgenesis - intellectual deficit - short stature
1482	Gonococcal conjunctivitis
3034	Gonzales-del Angel syndrome
169105	Good syndrome
1321	Goodman camptodactyly
65798	Goodman syndrome
375	Goodpasture syndrome
75389	Goossens-Devriendt syndrome
757	Gordon hyperkalemia-hypertension syndrome
376	Gordon syndrome
73	Gorham disease
73	Gorham syndrome
73	Gorham-Stout disease
377	Gorlin syndrome
2095	Gorlin-Chaudhry-Moss syndrome
66629	GOSHS
2500	Gottron syndrome
900	GPA

ORPHA Number	Disease name
280586	gPAPP deficiency
247353	GPP
721	GPS
403	GRA
2763	Gracile bone dysplasia
53693	GRACILE syndrome
39812	Graft versus host disease
505	Graham Little syndrome
505	Graham Little-Piccardi-Lassueur syndrome
2111	Graham-Boyle-Troxell syndrome
52055	Graham-Cox syndrome
3421	Grand-Kaine-Fulling syndrome
79094	Grange occlusive arterial syndrome
79094	Grange syndrome
2097	Grant syndrome
98962	Granular corneal dystrophy type 1
98963	Granular corneal dystrophy type 2
98962	Granular corneal dystrophy type I
98963	Granular corneal dystrophy type II
98961	Granular corneal dystrophy type III
98963	Granular-lattice corneal dystrophy
86850	Granulocytic sarcoma
900	Granulomatosis with polyangiitis
183	Granulomatous allergic angiitis
64722	Granulomatous mastitis
33111	Granulomatous slack skin
99915	Granulosa cell cancer
99915	Granulosa cell malignant tumor
35858	Gräsbeck-Imerslund disease
69665	Gravidic intrahepatic cholestasis
721	Gray platelet syndrome
293375	Grayson-Wilbrandt corneal dystrophy
276405	Green jaundice
99826	Green monkey disease
1426	Greenberg dysplasia
380	Greig cephalopolysyndactyly syndrome
495	Greither disease
721	Grey platelet syndrome
97261	GRF tumor
97261	GRFoma
139474	Grisart-Destrée syndrome
381	Griscelli disease
79476	Griscelli disease type 1
79477	Griscelli disease type 2
79478	Griscelli disease type 3
381	Griscelli-Pruniéras syndrome
79476	Griscelli-Pruniéras syndrome type 1
79477	Griscelli-Pruniéras syndrome type 2
79478	Griscelli-Pruniéras syndrome type 3
2099	Grix-Blankenship-Peterson syndrome

ORPHA Number	Disease name
3217	Groll-Hirschowitz syndrome
758	Gronblad-Strandberg-Touraine syndrome
1339	Grosse syndrome
314613	Growing teratoma syndrome
2055	Growth deficiency - brachydactyly - dysmorphism
2067	Growth delay - alopecia - pseudoanodontia - optic atrophy
53693	Growth delay - aminoaciduria - cholestasis - iron overload - lactic acidosis - early death
73272	Growth delay - deafness- intellectual deficit
3035	Growth delay - hydrocephaly - lung hypoplasia
79113	Growth delay - intellectual deficit - mandibulofacial dysostosis - microcephaly - cleft palate
73272	Growth delay due to insulin-like growth factor I deficiency
73273	Growth delay due to insulin-like growth factor I resistance
314769	Growth hormone and prolactin co-secreting pituitary adenoma
633	Growth hormone receptor deficiency
97261	Growth hormone releasing factor tumor
53693	Growth restriction - aminoaciduria - cholestasis - iron overload - lactic acidosis - early death
2101	Grubben-de Cock-Borghgraef syndrome
365	GSD due to acid maltase deficiency
308604	GSD due to acid maltase deficiency, adult onset
308552	GSD due to acid maltase deficiency, infantile onset
308573	GSD due to acid maltase deficiency, juvenile onset
57	GSD due to aldolase A deficiency
364	GSD due to G6P deficiency
79258	GSD due to G6P deficiency type a
79259	GSD due to G6P deficiency type b
79259	GSD due to G6PT deficiency
2088	GSD due to GLUT2 deficiency
367	GSD due to glycogen branching enzyme deficiency
308712	GSD due to glycogen branching enzyme deficiency, adult neuromuscular form
308684	GSD due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form
308698	GSD due to glycogen branching enzyme deficiency, childhood neuromuscular form
308670	GSD due to glycogen branching enzyme deficiency, congenital neuromuscular form
308655	GSD due to glycogen branching enzyme deficiency, fatal perinatal neuromuscular form

ORPHA Number	Disease name
308638	GSD due to glycogen branching enzyme deficiency, non progressive hepatic form
308621	GSD due to glycogen branching enzyme deficiency, progressive hepatic form
366	GSD due to glycogen debranching enzyme deficiency
263297	GSD due to glycogenin deficiency
2089	GSD due to hepatic glycogen synthase deficiency
2364	GSD due to lactate dehydrogenase deficiency
284435	GSD due to lactate dehydrogenase H-subunit deficiency
284426	GSD due to lactate dehydrogenase M-subunit deficiency
34587	GSD due to LAMP-2 deficiency
79240	GSD due to liver and muscle phosphorylase kinase deficiency
369	GSD due to liver glycogen phosphorylase deficiency
264580	GSD due to liver phosphorylase kinase deficiency
137625	GSD due to muscle and heart glycogen synthase deficiency
99849	GSD due to muscle beta-enolase deficiency
368	GSD due to muscle glycogen phosphorylase deficiency
371	GSD due to muscle phosphofructokinase deficiency
715	GSD due to muscle phosphorylase kinase deficiency
711	GSD due to phosphoglucomutase deficiency
713	GSD due to phosphoglycerate kinase 1 deficiency
97234	GSD due to phosphoglycerate mutase deficiency
370	GSD due to phosphorylase kinase deficiency
2089	GSD type 0a
137625	GSD type 0b
364	GSD type 1
79259	GSD type 1 non a
79258	GSD type 1a
79259	GSD type 1b
365	GSD type 2
308604	GSD type 2, adulte onset
308552	GSD type 2, infantile onset
308573	GSD type 2, juvenile onset
366	GSD type 3
367	GSD type 4
308712	GSD type 4, adult neuromuscular form
308684	GSD type 4, childhood combined hepatic and myopathic form
308698	GSD type 4, childhood neuromuscular form

ORPHA Number	Disease name
308670	GSD type 4, congenital neuromuscular form
308655	GSD type 4, fatal perinatal neuromuscular form
308638	GSD type 4, non progressive hepatic form
308621	GSD type 4, progressive hepatic form
368	GSD type 5
369	GSD type 6B
371	GSD type 7
370	GSD type 9
264580	GSD type 9A
79240	GSD type 9B
264580	GSD type 9C
715	GSD type 9D
715	GSD type 9E
284426	GSD type 11
57	GSD type 12
711	GSD type 14
263297	GSD type 15
370	GSD type IX
264580	GSD type IXa
79240	GSD type IXb
264580	GSD type IXc
715	GSD type IXd
715	GSD type IXe
263297	GSD type XV
79258	GSDIa
79259	GSDIb
366	GSDIII
308712	GSDIV, adult neuromuscular form
308684	GSDIV, childhood combined hepatic and myopathic form
308698	GSDIV, childhood neuromuscular form
308670	GSDIV, congenital neuromuscular form
308655	GSDIV, fatal perinatal neuromuscular form
308638	GSDIV, non progressive hepatic form
308621	GSDIV, progressive hepatic form
99849	GSDXIII
711	GSDXIV
59305	GTN
2102	GTP cyclohydrolase I deficiency
2102	GTPCH deficiency
90020	Guam disease
319234	Guanarito hemorrhagic fever
382	Guanidinoacetate methyltransferase deficiency
2785	Guibaud-Vainsel syndrome
2103	Guillain-Barré syndrome
98916	Guillain-Barré syndrome, acute inflammatory demyelinating polyradiculoneuropathic form
2103	Guillain-Barré-Strohl syndrome

ORPHA Number	Disease name
231	Guinea worm disease
1661	Guizar-Vasquez-Luengas syndrome
2104	GuizarVazquez-Sanchez-Manzano syndrome
1562	Gunal-Seber-Basaran syndrome
79277	Günther disease
1858	Gurrieri-Sammito-Bellussi syndrome
324561	Guttate hypopigmentation and punctate palmoplantar keratoderma
2957	Guttmacher syndrome
39812	GVH
293375	GWCD
370	Gycogenosis due to PhK deficiency
99914	Gynandroblastoma
414	Gyrate atrophy of choroid and retina
168569	H syndrome
139441	H-ABC
2396	Haberland syndrome
99803	Haddad syndrome
217026	Hadziselimovic syndrome
91378	HAE
100051	HAE 2
100054	HAE 3
100050	HAE-I
100051	HAE-II
100054	HAE-III
2026	HAFF
79263	Hagberg-Santavuori disease
2841	Hailey-Hailey disease
2342	Haim-Munk syndrome
1408	Hair defect - photosensitivity - intellectual deficit
69084	Hair-nail ectodermal dysplasia
58017	Hairy cell leukemia
300878	Hairy cell leukemia variant
2220	Hairy elbows
3387	Hairy throat syndrome
955	Hajdu-Cheney syndrome
2521	Halal syndrome
1809	Halal-Setton-Wang syndrome
185	Halasz syndrome
2985	Hal-Berg-Rudolph syndrome
2108	Hallermann-Streiff syndrome
2109	Hallermann-Streiff-François syndrome, severe form
2109	Hallermann-Streiff-like syndrome
2109	Hallerman-Streiff-François syndrome, severe form
2109	Hallerman-Streiff-like syndrome
157850	Hallervorden-Spatz syndrome
138	Hall-Hittner syndrome
2107	Hall-Riggs syndrome

ORPHA Number	Disease name
2110	Hallux varus - preaxial polysyndactyly
3453	HAM syndrome
289326	HAM/TSP
314555	Hamamy syndrome
2926	Hamanishi-Ueba-Tsuji syndrome
1217	Hamano-Tsukamoto syndrome
2869	Hamartomatous intestinal polyposis
93946	Hamel cerebro-palato-cardiac syndrome
79126	Hamman-Rich syndrome
73229	HANAC syndrome
1927	Hand and foot deformity - flat facies
2438	Hand-foot-genital syndrome
2438	Hand-foot-uterus syndrome
99873	Hand-Schüller-Christian disease
989	Hanhart syndrome
186	Hanot syndrome
340	Hantavirosis
340	Hantavirus fever
319247	Hantavirus pulmonary syndrome
3294	Hapnes-Boman-Skeie syndrome
1490	Harboyan syndrome
899	HARD syndrome
85182	Hardcastle syndrome
1415	Hardikar syndrome
1177	Harding ataxia
2812	Hard-skin syndrome, Parana type
457	Harlequin ichthyosis
199282	Harlequin syndrome
2115	Harrod syndrome
2116	Hartnup disorder
2116	Hartnup syndrome
2117	Hartsfield-Bixler-Demyer syndrome
84085	HAS
83601	Hashimoto encephalitis
99872	Hashimoto-Pritzker syndrome
2994	Haspeslagh-Fryns-Muelenaere syndrome
3325	HAT
2118	Hawkinsinuria
1071	Hay-Wells syndrome
163596	Hb Bart's hydrops fetalis
231242	HbC - beta-thalassemia
231249	HbE - beta-thalassemia
93616	HbH disease
352657	HBID
330032	HbLepore - beta-thalassemia
251359	HbS - beta-thalassemia
251365	HbSC disease
251370	HbSD disease
251375	HbSE disease
86864	HCD

ORPHA Number	Disease name
93556	HCDD
85458	HCHWA
324723	HCHWA, Arctic type
100006	HCHWA, Dutch type
324718	HCHWA, Flemish type
100008	HCHWA, Icelandic type
324708	HCHWA, Iowa type
324713	HCHWA, Italian type
324703	HCHWA, Piedmont type
100006	HCHWA-D
58017	HCL
300878	HCL-v
163690	HCS
306741	HD-HA syndrome
157941	HDL1
98934	HDL2
157946	HDL3
98759	HDL4
313808	HDLS
2237	HDR syndrome
67037	Head and neck squamous cell carcinoma
254898	Hearing loss - encephaloneuropathy - obesity - valvulopathy
3225	Hearing loss - familial salivary gland insensitivity to aldosterone
1355	Heart defect - round face - congenital developmental delay
1338	Heart defect - tongue hamartoma - polysyndactyly
1354	Heart defects - limb shortening
875	Heart tumor of the child
392	Heart-hand syndrome type 1
1350	Heart-hand syndrome type 2
1342	Heart-hand syndrome type 3
168796	Heart-hand syndrome, Slovenian type
1342	Heart-hand syndrome, Spanish type
1342	Heart-limb syndrome type 3
93556	Heavy chain deposition disease
86864	Heavy chain disease
2119	HEC syndrome
3377	Hecht syndrome
3377	Hecht-Beals syndrome
2492	Hecht-Scott syndrome
2120	Heckenlively syndrome
238468	HED
98813	HED-ID
2787	Heide syndrome
3220	Heimler syndrome
99932	Heiner syndrome
178330	Heinz body anemia
86813	Helicoid peripapillary chorioretinal degeneration
168782	Heller syndrome
252054	Hemangioblastoma
2330	Hemangioma-thrombocytopenia syndrome
90053	Hematopoietic stem cell transplantation
2128	Hemi 3 syndrome
86908	Hemiconvulsion-hemiplegia-epilepsy syndrome
2128	Hemicorporal hypertrophy
306741	Hemidystonia-hemiatrophy syndrome
1241	Hemifacial hyperplasia - strabismus
141145	Hemifacial hypertrophy
141136	Hemifacial microsomia
2549	Hemifacial microsomia - radial defects
141148	Hemifacial myohyperplasia
276280	Hemihyperplasia-multiple lipomatosis syndrome
2128	Hemihypertrophy
2129	Hemihypertrophy - intestinal web - corneal opacity
99802	Hemimegalencephaly
306669	Hemiparkinsonism-hemiatrophy syndrome
139491	Hemochromatosis due to defect in ferroportin
79230	Hemochromatosis type 2
225123	Hemochromatosis type 3
139491	Hemochromatosis type 4
163596	Hemoglobin Bart's hydrops fetalis
231242	Hemoglobin C - beta-thalassemia
2132	Hemoglobin C disease
90039	Hemoglobin D disease
231249	Hemoglobin E - beta-thalassemia
2133	Hemoglobin E disease
93616	Hemoglobin H disease
330032	Hemoglobin Lepore - beta-thalassemia
330041	Hemoglobin M disease
280615	Hemoglobinopathy Toms River
86817	Hemolytic anemia due to adenylate kinase deficiency
714	Hemolytic anemia due to diphosphoglycerate mutase deficiency
99138	Hemolytic anemia due to erythrocyte adenosine deaminase overproduction
712	Hemolytic anemia due to glucophosphate isomerase deficiency
90030	Hemolytic anemia due to glutathione reductase deficiency
248305	Hemolytic anemia due to glyceraldehyde-3-phosphate dehydrogenase deficiency
35120	Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency
766	Hemolytic anemia due to red cell pyruvate kinase deficiency
275944	Hemolytic disease of the newborn with Kell allo-immunization
90038	Hemolytic-uremic syndrome with diarrhea
2134	Hemolytic-uremic syndrome without diarrhea
93581	Hemolytic-uremic syndrome without diarrhea with anti-factor H antibodies
93578	Hemolytic-uremic syndrome without diarrhea with B factor anomaly
93575	Hemolytic-uremic syndrome without diarrhea with C3 anomaly
357008	Hemolytic-uremic syndrome without diarrhea with DGKE deficiency
93579	Hemolytic-uremic syndrome without diarrhea with H factor anomaly
93580	Hemolytic-uremic syndrome without diarrhea with I factor anomaly
93576	Hemolytic-uremic syndrome without diarrhea with MCP/CD46 anomaly
217023	Hemolytic-uremic syndrome without diarrhea with thrombomodulin anomaly
448	Hemophilia
98878	Hemophilia A
98879	Hemophilia B
329	Hemophilia C
178396	Hemorrhagic disease due to alpha-1-antitrypsin Pittsburgh mutation
340	Hemorrhagic fever - renal syndrome
274	Hemorrhagiparous thrombocytic dystrophy
324632	Hendra virus infection
2136	Hennekam syndrome
2135	Hennekam-Beemer syndrome
761	Henoch-Schönlein purpura
95159	HEP
79269	Heparan sulfamidase deficiency
79271	Heparan-alpha-glucosaminide N-acetyltransferase deficiency
3325	Heparin-associated thrombocytopenia
3325	Heparin-induced thrombocytopenia
3325	Heparin-induced thrombocytopenia type 2
102069	Hepatic amyloidosis with intrahepatic cholestasis
156	Hepatic carnitine palmitoyl transferase 1 deficiency
156	Hepatic carnitine palmitoyl transferase I deficiency
386	Hepatic cystic hamartoma
100085	Hepatic endocrine tumor
2031	Hepatic fibrosis - renal cysts - intellectual deficit
369	Hepatic glycogen phosphorylase deficiency
369	Hepatic phosphorylase deficiency
890	Hepatic veno-occlusive disease
79124	Hepatic veno-occlusive disease - immunodeficiency
90073	Hepatitis B re-infection following liver transplantation

ORPHA Number	Disease name
449	Hepatoblastoma
54272	Hepatocellular adenoma
88673	Hepatocellular carcinoma
33402	Hepatocellular carcinoma, childhood-onset
137681	Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1
137681	Hepatoencephalopathy due to COXPD1
95159	Hepatoerythropoietic porphyria
905	Hepatolenticular degeneration
64743	Hepatoportal sclerosis
364	Hepatorenal glycogenosis
882	Hepatorenal tyrosinemia
86882	Hepatosplenic T-cell lymphoma
306539	Hereditary acrokeratotic poikiloderma of Kindler-Weary
2907	Hereditary acrokeratotic poikiloderma, Weary type
85450	Hereditary amyloid nephropathy
93560	Hereditary amyloid nephropathy due to apolipoprotein AI variant
238269	Hereditary amyloid nephropathy due to Apolipoprotein AII variant
93562	Hereditary amyloid nephropathy due to fibrinogen A alpha-chain variant
93561	Hereditary amyloid nephropathy due to lysozyme variant
85448	Hereditary amyloidosis, Finnish type
228277	Hereditary anetoderma
91378	Hereditary angioedema
100050	Hereditary angioedema type 1
100051	Hereditary angioedema type 2
100054	Hereditary angioedema type 3
91378	Hereditary angioneurotic edema
100050	Hereditary angioneurotic edema type 1
100051	Hereditary angioneurotic edema type 2
100054	Hereditary angioneurotic edema type 3
73229	Hereditary angiopathy-nephropathy-aneurysms-muscle cramps syndrome
3115	Hereditary areflexic dystasia, Roussy-Lévy type
289601	Hereditary arterial and articular multiple calcification syndrome
1429	Hereditary benign chorea
352657	Hereditary benign corneal intraepithelial dyskeratosis
352657	Hereditary benign intraepithelial dyskeratosis
91378	Hereditary bradykinine-induced angioedema
145	Hereditary breast and ovarian cancer syndrome
227535	Hereditary breast cancer
227535	Hereditary breast carcinoma
36382	Hereditary CAD
30925	Hereditary CDI
30925	Hereditary central diabetes insipidus
221061	Hereditary cerebral cavernoma
221061	Hereditary cerebral cavernous malformation
85458	Hereditary cerebral hemorrhage with amyloidosis
324723	Hereditary cerebral hemorrhage with amyloidosis, Arctic type
100006	Hereditary cerebral hemorrhage with amyloidosis, Dutch type
324718	Hereditary cerebral hemorrhage with amyloidosis, Flemish type
100008	Hereditary cerebral hemorrhage with amyloidosis, Icelandic type
324708	Hereditary cerebral hemorrhage with amyloidosis, Iowa type
324713	Hereditary cerebral hemorrhage with amyloidosis, Italian type
324703	Hereditary cerebral hemorrhage with amyloidosis, Piedmont type
48818	Hereditary ceruloplasmin deficiency
36382	Hereditary cervical artery dissections
53372	Hereditary chin myoclonus
53372	Hereditary chin-trembling
676	Hereditary chronic pancreatitis
238578	Hereditary clubfoot due to 17q23.1-q23.2 microduplication
293144	Hereditary clubfoot due to 5q31 microdeletion
293150	Hereditary clubfoot due to PITX1 point mutation
98434	Hereditary combined deficiency of factors II, VII, IX and X
98434	Hereditary combined deficiency of vitamin K-dependent clotting factors
238722	Hereditary congenital controlateral synkinesia
238722	Hereditary congenital mirror movements
972	Hereditary continuous muscle fiber activity
79273	Hereditary coproporphyria
60015	Hereditary cranium bifidum
168577	Hereditary cryohydrocytosis type 2
168577	Hereditary cryohydrocytosis with reduced stomatin
98967	Hereditary crystalline stromal dystrophy of Schnyder
100008	Hereditary cystatin C amyloid angiopathy
313808	Hereditary diffuse leukoencephalopathy with spheroids
288	Hereditary elliptocytosis
63261	Hereditary endotheliopathy - retinopathy - nephropathy - stroke
98873	Hereditary erythroblastic multinuclearity with a positive acidified-serum test (hempas)
85195	Hereditary expansile polyostotic osteolytic dysplasia
157846	Hereditary ferritinopathy
90045	Hereditary folate malabsorption
469	Hereditary fructose intolerance
469	Hereditary fructose-1-phosphate aldolase deficiency
469	Hereditary fructosemia
53372	Hereditary geniospasm
2024	Hereditary gingival fibromatosis
2024	Hereditary gingival hyperplasia
359	Hereditary glaucoma
774	Hereditary hemorrhagic telangiectasia
2604	Hereditary hollow visceral myopathy
199285	Hereditary hypercarotenemia and vitamin A deficiency
238475	Hereditary hypercholanemia
3197	Hereditary hyperekplexia
3197	Hereditary hyperexplexia
163	Hereditary hyperferritinemia with congenital cataracts
163	Hereditary hyperferritinemia-cataract syndrome
2801	Hereditary hyperphosphatasia
157215	Hereditary hypophosphatemic rickets with hypercalciuria
55654	Hereditary hypotrichosis simplex
90368	Hereditary hypotrichosis simplex of the scalp
217407	Hereditary hypotrichosis with recurrent skin vesicles
79091	Hereditary inclusion body myopathy - joint contractures - ophthalmoplegia
602	Hereditary inclusion body myopathy type 2
79091	Hereditary inclusion body myopathy type 3
324381	Hereditary inclusion body myopathy type 4
178464	Hereditary inclusion body myopathy with early respiratory failure
300373	Hereditary infantile gigantism
332	Hereditary juvenile meganoblastic anemia due to intrinsic factor deficiency
2334	Hereditary keratitis
493	Hereditary keratoacanthoma
523	Hereditary leiomyomatosis
523	Hereditary leiomyomatosis with renal carcinoma
523	Hereditary leiomyomatosis with renal cell cancer
79452	Hereditary lymphedema type I
90186	Hereditary lymphedema type II

ORPHA Number	Disease name
228277	Hereditary macular atrophy
621	Hereditary methemoglobinemia
330041	Hereditary methemoglobinemia due to hemoglobin mutation
157794	Hereditary mixed polyposis syndrome
65753	Hereditary motor and sensory neuropathy type 1
64748	Hereditary motor and sensory neuropathy type 3
773	Hereditary motor and sensory neuropathy type 4
64751	Hereditary motor and sensory neuropathy type 5
90120	Hereditary motor and sensory neuropathy type 6
90103	Hereditary motor and sensory neuropathy with deafness, intellectual deficit and absent sensory large myelinated fibers
99950	Hereditary motor and sensory neuropathy, Lom type
90117	Hereditary motor and sensory neuropathy, Okinawa type
90117	Hereditary motor and sensory neuropathy, proximal type
99953	Hereditary motor and sensory neuropathy, Russe Type
1839	Hereditary mucoepithelial dysplasia
171723	Hereditary mucosal leukokeratosis
136	Hereditary multi-infarct dementia
523	Hereditary multiple cutaneous leiomyomas
83454	Hereditary multiple glomangiomas
2590	Hereditary myoclonus - progressive distal muscular atrophy
43115	Hereditary myopathy with lactic acidosis due to ISCU deficiency
1062	Hereditary neurocutaneous angioma
30925	Hereditary neurogenic diabetes insipidus
640	Hereditary neuropathy with liability to pressure palsies
279943	Hereditary neutrophilia
91378	Hereditary non histamine-induced angioedema
144	Hereditary nonpolyposis colon cancer
144	Hereditary nonpolyposis colorectal cancer
168583	Hereditary North American Indian childhood cirrhosis
56	Hereditary ochronosis
30	Hereditary orotic aciduria
79141	Hereditary painful callosities
86923	Hereditary palmoplantar hyperkeratosis, Gamborg-Nielsen type
86923	Hereditary palmoplantar keratoderma, Gamborg-Nielsen type
99878	Hereditary parathyroids hyperplasia
2828	Hereditary Parkinson disease
168615	Hereditary persistence of alpha-fetoprotein
46532	Hereditary persistence of fetal hemoglobin - beta-thalassemia
251380	Hereditary persistence of fetal hemoglobin - sickle cell disease
29072	Hereditary pheochromocytoma-paraganglioma
300373	Hereditary pituitary hyperplasia
330061	Hereditary polymorphous light eruption of American Indians
178345	Hereditary prepubertal gynecomastia
828	Hereditary progressive arthroophthalmopathy
98808	Hereditary progressive dystonia with marked diurnal fluctuation
158025	Hereditary progressive mucinous histiocytosis
178464	Hereditary proximal myopathy with early respiratory failure
275777	Hereditary pulmonary arterial hypertension
98867	Hereditary pyropoikilocytosis
85450	Hereditary renal amyloidosis
93560	Hereditary renal amyloidosis due to apolipoprotein AI variant
238269	Hereditary renal amyloidosis due to apolipoprotein AII variant
93562	Hereditary renal amyloidosis due to fibrinogen A alpha-chain variant
93561	Hereditary renal amyloidosis due to lysozyme variant
94088	Hereditary renal hypouricemia
788	Hereditary resistance to anti-vitamin K
357027	Hereditary retinoblastoma
221043	Hereditary sclerosing poikiloderma with tendon and pulmonary involvement
221039	Hereditary sclerosing poikiloderma, Weary type
280598	Hereditary sensorimotor neuropathy with hyperelastic skin
36386	Hereditary sensory and autonomic neuropathy type 1
139564	Hereditary sensory and autonomic neuropathy type 1 with cough and gastroesophageal reflux
139564	Hereditary sensory and autonomic neuropathy type 1B
970	Hereditary sensory and autonomic neuropathy type 2
1764	Hereditary sensory and autonomic neuropathy type 3
642	Hereditary sensory and autonomic neuropathy type 4
64752	Hereditary sensory and autonomic neuropathy type 5
314381	Hereditary sensory and autonomic neuropathy type 6
139573	Hereditary sensory and autonomic neuropathy with deafness and global delay
139578	Hereditary sensory and autonomic neuropathy with spastic paraplegia
213524	Hereditary site-specific ovarian cancer syndrome
685	Hereditary spastic paraparesis
685	Hereditary spastic paraplegia
822	Hereditary spherocytosis
79133	Hereditary symmetrical aplastic nevi of temples
84093	Hereditary thermosensitive neuropathy
71493	Hereditary thrombocythemia
268322	Hereditary thrombocytopenia with normal platelets
329319	Hereditary thrombocytosis with transverse limb defect
82	Hereditary thrombophilia due to congenital antithrombin 3 deficiency
82	Hereditary thrombophilia due to congenital antithrombin deficiency
217467	Hereditary thrombophilia due to congenital histidine-rich (poly-L) glycoprotein deficiency
217467	Hereditary thrombophilia due to congenital HRG deficiency
745	Hereditary thrombophilia due to congenital protein C deficiency
743	Hereditary thrombophilia due to congenital protein S deficiency
205	Hereditary unconjugated hyperbilirubinemia
79234	Hereditary unconjugated hyperbilirubinemia type 1
79235	Hereditary unconjugated hyperbilirubinemia type 2
71291	Hereditary vascular retinopathy
71291	Hereditary vascular retinopathy - Raynaud phenomenon - migraine
93160	Hereditary vitamin D-resistant rickets
98805	Hereditary whispering dysphonia
903	Hereditary Willebrand disease
170	Hereditary woolly hair syndrome
170	Hereditary wooly hair syndrome
3467	Hereditary xanthinuria
3202	Hereditary xerocytosis
64746	Hereditarymotor and sensoryneuropathy type 2
773	Heredopathia atactica polyneuritiformis
275777	Heritable pulmonary arterial hypertension
3411	Herlyn-Werner syndrome
79430	Hermansky-Pudlak syndrome
183678	Hermansky-Pudlak syndrome type 2
231531	Hermansky-Pudlak syndrome type 7
231537	Hermansky-Pudlak syndrome type 8
280663	Hermansky-Pudlak syndrome type 9
183678	Hermansky-Pudlak syndrome with neutropenia

ORPHA Number	Disease name
231500	Hermansky-Pudlak syndrome with pulmonary fibrosis
231512	Hermansky-Pudlak syndrome without pulmonary fibrosis
2139	Hernandez-Aguirre Negrete syndrome
2786	Hernandez-Fragoso syndrome
63261	HERNS syndrome
1930	Herpes simplex encephalitis
1930	Herpes simplex neuroinvasion
137586	Herpes simplex virus keratitis
293	Herpes virus antenatal infection
1930	Herpetic encephalitis
137586	Herpetic keratitis
208524	Herpetiform pemphigus
369	Hers disease
1486	Herva disease
314970	HES-L
314950	HES-M
314950	HES-N
314962	HES-R
450	Heterotaxia
450	Heterotaxy syndrome
640	Heterozygous microdeletion 17p11.2p12
3450	Heterozygous OSMED
3450	Heterozygous otospondylomegaepiphyseal dysplasia
845	Hexosaminidase A deficiency
309192	Hexosaminidase A deficiency, adult form
309239	Hexosaminidase A deficiency, B1 variant
309178	Hexosaminidase A deficiency, infantile form
309185	Hexosaminidase A deficiency, juvenile form
309246	Hexosaminidase activator deficiency
796	Hexosaminidases A and B deficiency
309169	Hexosaminidases A and B deficiency, adult form
309155	Hexosaminidases A and B deficiency, infantile form
309162	Hexosaminidases A and B deficiency, juvenile form
2438	HFGS
2744	HGPPS
79271	HGSNAT deficiency
163	HHCS
86908	HHE syndrome
415	HHH syndrome
276280	HHML
157215	HHRH
774	HHT
457	HI
435	HI syndrome
35878	HI/HA syndrome
602	HIBM2
79091	HIBM3
324381	HIBM4
178464	HIBM-ERF
189	Hidrotic ectodermal dysplasia
1808	Hidrotic ectodermal dysplasia, Christianson-Fourie type
1809	Hidrotic ectodermal dysplasia, Halal type
343	HIDS
137577	HIE
330012	High altitude pulmonary edema
314029	High bone mass OI
314029	High bone mass osteogenesis imperfecta
3181	High scapula
231080	High-grade dysplasia in patients with Barrett esophagus
251646	High-grade ependymoma
101088	HIGM1
101089	HIGM2
101090	HIGM3
101091	HIGM4
101092	HIGM5
99978	Hilar CCA
99978	Hilar cholangiocarcinoma
84085	Hinman syndrome
84085	Hinman-Allen syndrome
84085	Hinman's syndrome
1164	Hinson-Pepys disease
3408	Hip dysplasia - enchondromata - ecchondroma
2114	Hip dysplasia, Beukes type
2129	HIPO syndrome
892	Hippel-Lindau disease
99151	Hippocampal tauopathy in cerebral aging
65684	Hirayama disease
388	Hirschsprung disease
2155	Hirschsprung disease - deafness - polydactyly
2151	Hirschsprung disease - ganglioneuroblastoma
2152	Hirschsprung disease - intellectual deficit
2153	Hirschsprung disease - nail hypoplasia - dysmorphism
2150	Hirschsprung disease - type D brachydactyly
261537	Hirschsprung disease and intellectual deficit due to 2q22 microdeletion
261552	Hirschsprung disease and intellectual deficit due to a point mutation
261537	Hirschsprung disease and intellectual deficit due to del(2)(q22)
261537	Hirschsprung disease and intellectual deficit due to monosomy 2q22
2026	Hirsutism - congenital gingival hyperplasia
2156	Hirsutism - skeletal dysplasia - intellectual deficit
3283	His bundle tachycardia
2157	Histidase deficiency
2157	Histidinemia
2157	Histidinuria
2158	Histidinuria - renal tubular defect
50918	Histiocytic necrotizing lymphadenitis
86896	Histiocytic sarcoma
137675	Histiocytoid cardiomyopathy
389	Histiocytosis X
264955	Histiocytosis X in childhood and adulthood
264750	Histiocytosis X specific to adulthood
264724	Histiocytosis X specific to childhood
390	Histoplasmosis
3325	HIT
1573	HJMD
572	HLA class 2-negative severe combined immunodeficiency
523	HLRCC
2213	HMC syndrome
35701	HMG-CoA synthase deficiency
64748	HMSN 3
773	HMSN 4
64751	HMSN 5
90117	HMSNP
99953	HMSNR
69084	HNED
144	HNPCC
640	HNPP
67037	HNSCC
391	Hodgkin lymphoma, classical
1979	Hoepffner-Dreyer-Reimers syndrome
2349	Hoffman syndrome
414	HOGA
3328	Holmes-Collins syndrome
93970	Holmes-Gang syndrome
2143	Holmes-Schepens syndrome
2161	Holoacardius amorphus
79242	Holocarboxylase synthetase deficiency
2162	Holoprosencephaly
2165	Holoprosencephaly - caudal dysgenesis
2163	Holoprosencephaly - craniosynostosis
2117	Holoprosencephaly - ectrodactyly - cleft lip palate
2166	Holoprosencephaly - postaxial polydactyly
3186	Holoprosencephaly - radial heart renal anomalies
392	Holt-Oram syndrome

ORPHA Number	Disease name
2167	Holzgreve-Wagner-Rehder syndrome
30924	HOMG1
34528	HOMG2
31043	HOMG3
2168	Homocarnosinase deficiency
2168	Homocarnosinosis
394	Homocystinuria due to cystathionine beta-synthase deficiency
395	Homocystinuria due to methylene tetrahydrofolate reductase deficiency
622	Homocystinuria without methylmalonic aciduria
56	Homogentisic acid oxidase deficiency
163596	Homozygous alpha0-thalassemia
14	Homozygous familial hypobetalipoproteinemia
98865	Homozygous hereditary elliptocytosis
98960	Honeycomb corneal dystrophy
98958	Honey-droplet corneal dystrophy
78	Hookworms infection
307936	HOPP syndrome
2744	Horizontal gaze palsy with progressive scoliosis
397	Horton disease
392	HOS
166412	Hot water reflex epilepsy
1352	Houlston-Ironton-Temple syndrome
99907	House allergic alveolitis
2198	Howell-Evans syndrome
3322	Hoyeraal-Hreidarsson syndrome
31740	HP
98808	HPD with marked diurnal fluctuation
2162	HPE
46532	HPFH - beta-thalassemia
251380	HPFH - sickle cell disease
306669	HP-HA syndrome
247262	HPMR
436	HPP
293958	HPPD
510	HPRT complete deficiency
510	HPRT deficiency grade IV
79233	HPRT deficiency, grade I
79233	HPRT partial deficiency
79233	HPRT1 partial deficiency
79233	HPRT-related gout
79233	HPRT-related hyperuricemia
79430	HPS
231500	HPS with pulmonary fibrosis
231512	HPS without pulmonary fibrosis
183678	HPS2
231531	HPS7
231537	HPS8
280663	HPS9
99880	HPT-JT
2323	HRD syndrome
84085	HS
139564	HSAN with cough and gastroesophageal reflux
139573	HSAN with deafness and global delay
139578	HSAN with spastic paraplegia
139564	HSAN1B
970	HSAN2
1764	HSAN3
642	HSAN4
64752	HSAN5
314381	HSAN6
2182	HSAS
388	HSCR
35123	HSD deficiency
685	HSP
1930	HSV encephalitis
137586	HSV keratitis
285	HT-EDS
289326	HTLV-1-associated myelopathy/tropical spastic paraparesis
228116	Hughes-Stovin syndrome
289326	Human T-lymphotropic virus type I-associated myelopathy/tropical spastic paraparesis
289326	Human T-lymphotropic virus type-1-associated myelopathy/tropical spastic paraparesis
294973	Humeral agenesis/hypoplasia
295063	Humeral agenesis/hypoplasia, bilateral
295061	Humeral agenesis/hypoplasia, unilateral
294973	Humeral intercalary meromelia
295063	Humeral intercalary meromelia, bilateral
295061	Humeral intercalary meromelia, unilateral
295211	Humero-radial fusion, bilateral
295209	Humero-radial fusion, unilateral
3265	Humero-radial synostosis
295211	Humero-radial synostosis, bilateral
295209	Humero-radial synostosis, unilateral
295207	Humero-radio-ulnar fusion, bilateral
295205	Humero-radio-ulnar fusion, unilateral
294975	Humero-radio-ulnar intercalary transverse meromelia
295087	Humero-radio-ulnar intercalary transverse meromelia, bilateral
295085	Humero-radio-ulnar intercalary transverse meromelia, unilateral
3266	Humero-radio-ulnar synostosis
295207	Humero-radio-ulnar synostosis, bilateral
295205	Humero-radio-ulnar synostosis, unilateral
295215	Humero-ulnar fusion, bilateral
295213	Humero-ulnar fusion, unilateral
94056	Humero-ulnar synostosis
295215	Humero-ulnar synostosis, bilateral
295213	Humero-ulnar synostosis, unilateral
3383	Humerus trochlea aplasia
580	Hunter syndrome
217085	Hunter syndrome type A
217093	Hunter syndrome type B
2715	Hunter-Jurenka-Thompson syndrome
97340	Hunter-McAlpine craniosynostosis
3365	Hunter-Rudd-Hoffmann syndrome
1390	Hunter-Thompson-Reed syndrome
399	Huntington chorea
399	Huntington disease
157941	Huntington disease-like 1
98934	Huntington disease-like 2
157946	Huntington disease-like 3
98759	Huntington disease-like 4
384	Huriez syndrome
93473	Hurler syndrome
93476	Hurler-Scheie syndrome
330061	Hutchinson summer prurigo
740	Hutchinson-Gilford progeria syndrome
93160	HVDRR
71291	HVR
53698	Hyaline body myopathy
70587	Hyaline membrane disease
530	Hyalinosis cutis et mucosae
67041	Hyaluronidase deficiency
86851	Hybrid acute leukemia
99927	Hydatidiform mole
400	Hydatidosis
2898	Hyde Forster-McCarthy-Berry syndrome
2177	Hydranencephaly
330021	Hydrargyria
330061	Hydroa aestivale
330058	Hydroa vacciniforme
899	Hydrocephalus - agyria - retinal dysplasia
2186	Hydrocephalus - blue sclerae - nephropathy
916	Hydrocephalus - cleft palate - joint contractures
2180	Hydrocephalus - costovertebral dysplasia - Sprengel anomaly
2119	Hydrocephalus - endocardial fibroelastosis - cataract
2183	Hydrocephalus - obesity - hypogonadism
2182	Hydrocephalus with stenosis of aqueduct of Sylvius
899	Hydrocephalus-agyria-retinal dysplasia syndrome
2184	Hydrocephaly - low insertion umbilicus

ORPHA Number	Disease name
2181	Hydrocephaly - tall stature - joint laxity
221126	Hydrocephaly/hydranencephaly due to cerebral vasculopathy
2189	Hydrolethalus
3280	Hydromelia
2473	Hydrometrocolpos - postaxial polydactyly
2704	Hydronephrosis - inverted smile
1426	Hydrops - ectopic calcification - motheaten
1041	Hydrops fetalis
20	Hydroxymethylglutaric aciduria
401	Hymenolepiasis
553	Hyperadrenocorticism
309147	Hyperalaninemia
927	Hyperammonemia due to N-acetylglutamate synthetase deficiency
168588	Hyperandrogenism due to cortisone reductase deficiency
90	Hyperargininemia
309147	Hyper-beta-alaninemia
234	Hyperbilirubinemia type 2
3111	Hyperbilirubinemia, Rotor type
276405	Hyperbiliverdinemia
306661	Hypercalcemic tumoral calcinosis
2196	Hypercalciuria - bilateral macular coloboma
209902	Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency
83639	Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency
553	Hypercortisolism
1032	Hyperdibasic aminoaciduria type 1
470	Hyperdibasic aminoaciduria type 2
3197	Hyperekplexia
163985	Hyperekplexia - epilepsy
168956	Hypereosinophilic syndrome
408	Hyperglycerolemia
2410	Hypergonadotropic hypogonadism - cataract syndrome
243	Hypergonadotropic ovarian dysgenesis
343	Hyper-IgD syndrome
101090	Hyper-IgM syndrome due to CD40 deficiency
101088	Hyper-IgM syndrome due to CD40 ligand deficiency
101088	Hyper-IgM syndrome due to CD40L deficiency
101092	Hyper-IgM syndrome due to UNG deficiency
101092	Hyper-IgM syndrome due to uracil N glycosylase
101088	Hyper-IgM syndrome type 1
101089	Hyper-IgM syndrome type 2
101090	Hyper-IgM syndrome type 3
101091	Hyper-IgM syndrome type 4
101092	Hyper-IgM syndrome type 5
742	Hyperimidodipeptiduria
343	Hyperimmunoglobinemia D with recurrent fever
2314	Hyperimmunoglobulin E syndrome type 1
169446	Hyperimmunoglobulin E syndrome type 2
2314	Hyperimmunoglobulin E-recurrent infection syndrome
343	Hyperimmunoglobulinemia D syndrome
343	Hyperimmunoglobulinemia D with periodic fever
71212	Hyperinsulinemic hypoglycemia due to 3-hydroxylacyl-CoA dehydrogenase deficiency
79299	Hyperinsulinemic hypoglycemia due to glucokinase deficiency
324575	Hyperinsulinemic hypoglycemia due to HNF1A deficiency
263455	Hyperinsulinemic hypoglycemia due to HNF4A deficiency
263458	Hyperinsulinemic hypoglycemia due to INSR deficiency
263458	Hyperinsulinemic hypoglycemia due to insulin receptor deficiency
276603	Hyperinsulinemic hypoglycemia due to Kir6.2 deficiency, diazoxide-resistant focal form
276598	Hyperinsulinemic hypoglycemia due to SUR1 deficiency, diazoxide-resistant focal form
276556	Hyperinsulinemic hypoglycemia due to UCP2 deficiency
71212	Hyperinsulinism due to 3-hydroxylacyl-CoA dehydrogenase deficiency
79299	Hyperinsulinism due to glucokinase deficiency
71212	Hyperinsulinism due to glutamodehydrogenase deficiency
71212	Hyperinsulinism due to HADH deficiency
324575	Hyperinsulinism due to HNF1A deficiency
263455	Hyperinsulinism due to HNF4A deficiency
263458	Hyperinsulinism due to INSR deficiency
165991	Hyperinsulinism due to monocarboxylate transporter 1 deficiency
71212	Hyperinsulinism due to SCHAD deficiency
71212	Hyperinsulinism due to short-chain 3-hydroxylacyl-CoA dehydrogenase deficiency
165991	Hyperinsulinism due to SLC16A1 deficiency
276556	Hyperinsulinism due to UCP2 deficiency
35878	Hyperinsulinism-hyperammonemia syndrome
682	Hyperkalemic periodic paralysis
682	Hyperkalemic PP
89939	Hyperkalemic renal tubular acidosis
757	Hyperkaliemia - hypertension, Gordon type
409	Hyperkeratosis lenticularis perstans
1336	Hyperkeratosis-hyperpigmentation syndrome
682	HyperKPP
140905	Hyperlipidemia due to hepatic triglyceride lipase deficiency
412	Hyperlipidemia type 3
411	Hyperlipoproteinemia type 1
412	Hyperlipoproteinemia type 3
413	Hyperlipoproteinemia type 4
70470	Hyperlipoproteinemia type 5
2203	Hyperlysinemia
2203	Hyperlysinemia type I
3124	Hyperlysinemia type II
289891	Hypermethioninemia due to glycine N-methyltransferase deficiency
289891	Hypermethioninemia due to GNMT deficiency
88618	Hypermethioninemia due to S-adenosylhomocysteine hydrolase deficiency
289290	Hypermethioninemia encephalopathy due to adenosine kinase deficiency
289290	Hypermethioninemia encephalopathy due to ADK deficiency
73267	Hypernychthemeral syndrome
414	Hyperornithinemia
414	Hyperornithinemia - gyrate atrophy of choroid and retina
415	Hyperornithinemia-hyperammonemia-homocitrullinuria
2801	Hyperostosid corticalis deformans juvenilis
3416	Hyperostosis corticalis generalisata
77296	Hyperostosis frontalis interna
2780	Hyperostosis generalisata with striations
99880	Hyperparathyroidism - jaw tumor syndrome
295002	Hyperphalangy
295140	Hyperphalangy in digits 2-5
295142	Hyperphalangy, bilateral
295140	Hyperphalangy, unilateral
1388	Hyperphalangy-clinodactyly of index finger with Pierre Robin syndrome
238583	Hyperphenylalaninemia
13	Hyperphenylalaninemia due to 6-pyruvoyltetrahydropterin synthase deficiency
238583	Hyperphenylalaninemia due to BH4 deficiency
1578	Hyperphenylalaninemia due to dehydratase deficiency
226	Hyperphenylalaninemia due to dihydropteridine reductase deficiency

ORPHA Number	Disease name
2102	Hyperphenylalaninemia due to GTP cyclohydrolase deficiency
1578	Hyperphenylalaninemia due to pterin-4-alpha-carbinolamine dehydratase deficiency
238583	Hyperphenylalaninemia due to tetrahydrobiopterin deficiency
2209	Hyperphenylalaninemic embryopathy
247262	Hyperphosphatasia-intellectual deficiency syndrome
34	Hyperpipecolatemia
157798	Hyperplastic polyposis syndrome
682	HyperPP
419	Hyperprolinemia type 1
79101	Hyperprolinemia type 2
93604	Hyperprostaglandin E syndrome
727	Hypersensitivity angiitis
31740	Hypersensitivity pneumonitis
2211	Hypertelorism - hypospadias - polysyndactyly syndrome
1519	Hypertelorism, Teebi type
2213	Hypertelorism-microtia-facial clefting syndrome
2745	Hypertelorism-oesophageal abnormality-hypospadias syndrome
293958	Hypertelorism-preauricular sinus-punctual pits-deafness syndrome
293958	Hypertelorism-preauricular sinus-punctual pits-hearing loss syndrome
88660	Hypertension due to gain-of-function mutations in the mineralocorticoid receptor
757	Hypertensive hyperkalemia
423	Hyperthermia of anesthesia
1231	Hypertrichosis - atrophic skin - ectropion - macrostomia
2220	Hypertrichosis cubiti - short stature
2222	Hypertrichosis lanuginosa congenita
2222	Hypertrichosis universalis
2026	Hypertrichosis with acromegaloid facial appearence
319182	Hypertrichosis-short stature-facial dysmorphism-developmental delay syndrome
2765	Hypertrichotic osteochondrodysplasia
324525	Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation
324525	Hypertrophic cardiomyopathy and renal tubular disease due to mtDNA mutation
217601	Hypertrophic cardiomyopathy due to intensive athletic training
329883	Hypertrophic gastropathy without hypoproteinemia
64748	Hypertrophic neuropathy of infancy
90282	Hypertrophic or verrucous lupus erythematosus
2224	Hypertryptophanemia
217330	Hyperuricemia - anemia - renal failure
251523	Hyperzincemia and hypercalprotectinemia
276429	Hypnic headache
289157	Hypocalcemic vitamin D-dependent rickets
93160	Hypocalcemic vitamin D-resistant rickets
100032	Hypocalcified amelogenesis imperfecta
93297	Hypochondrogenesis
429	Hypochondroplasia
36412	Hypocomplementemic urticarial vasculitis
430	Hypodermyiasis
2228	Hypodontia - dysplasia of nails
2228	Hypodontia - nail dysgenesis
185	Hypogenetic lung syndrome
989	Hypoglossia - hypodactyly
3423	Hypogonadism - gynecomastia - X-linked intellectual deficit
2233	Hypogonadism - mitral valve prolapse - intellectual deficit
141333	Hypogonadism-short stature-coloboma-preaxial polydactyly syndrome
2230	Hypogonadotropic hypogonadism - frontoparietal alopecia
2235	Hypogonadotropic hypogonadism - retinitis pigmentosa
293967	Hypogonadotropic hypogonadism-severe microcephaly-sensorineural deafness-dysmorphism syndrome
293967	Hypogonadotropic hypogonadism-severe microcephaly-sensorineural hearing loss-dysmorphism syndrome
238468	Hypohidrotic ectodermal dysplasia
1882	Hypohidrotic ectodermal dysplasia - hypothyroidism - ciliary dyskinesia
98813	Hypohidrotic ectodermal dysplasia with immunodeficiency
293964	Hypoinsulinemic hypoglycemia and body hemihypertrophy
112	Hypokalemic alkalosis - hypercalciuria
681	Hypokalemic periodic paralysis
30924	Hypomagnesemia caused by selective magnesium malabsorption
30924	Hypomagnesemia intestinal type 1
31043	Hypomagnesemia renal type 3
1790	Hypomandibular faciocranial dysostosis
100033	Hypomaturation amelogenesis imperfecta
100034	Hypomaturation-hypoplastic amelogenesis imperfecta with taurodontism
435	Hypomelanosis of Ito
2435	Hypomelanotic and hypermelanotic cutaneous macules - retarded growth - intellectual deficiency
85163	Hypomyelination - congenital cataract
88637	Hypomyelination - hypogonadotropic hypogonadism - hypodontia
2680	Hypomyelination neuropathy - arthrogryposis
139441	Hypomyelination with atrophy of basal ganglia and cerebellum
3453	Hypoparathyroidism - Addison's disease - mucocutaneous candidiasis
3453	Hypoparathyroidism - Addison's disease - mucocutaneous candidosis
2237	Hypoparathyroidism - deafness - renal disease
2323	Hypoparathyroidism - intellectual deficit - dysmorphism
2323	Hypoparathyroidism - short stature - intellectual deficit - seizures
436	Hypophosphatasia
314621	Hypophyseal duplication
79477	Hypopigmentation - immunodeficiency with or without neurologic impairment
79476	Hypopigmentation - neurologic impairment
324561	Hypopigmentation and punctate keratosis of the palms and soles
42665	Hypopigmentation-deafness syndrome
324561	Hypopigmentation-punctate palmoplantar keratoderma syndrome
2626	Hypopituitarism - short stature - skeletal anomalies
91354	Hypopituitarism due to empty sella turcica syndrome
1863	Hypoplasia of the femoral trochlea
99058	Hypoplasia of the mitral valve annulus
722	Hypoplasminogenemia
100031	Hypoplastic amelogenesis imperfecta
2248	Hypoplastic left heart syndrome
293864	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbalder syndrome
3332	Hypoplastic tibiae - postaxial polydactyly
327	Hypoproconvertinemia
2494	Hypoproteinemic hypertrophic gastropathy
325	Hypoprothrombinemia
2250	Hyposmia - nasal and ocular hypoplasia - hypogonadotropic hypogonadism
157788	Hypospadias - hypertelorism - coloboma and deafness
2261	Hypospadias - intellectual deficit, Goldblatt type
2745	Hypospadias-dysphagia syndrome
2745	Hypospadias-hypertelorism syndrome
2353	Hypotelorism - cleft palate - hypospadias
672	Hypothalamic hamartoblastoma syndrome
86906	Hypothalamic hamartomas with gelastic seizures
1226	Hypothyroidism - cleft palate
3047	Hypothyroidism - dysmorphism - postaxial polydactyly - intellectual deficit

ORPHA Number	Disease name
226307	Hypothyroidism due to deficient transcription factors involved in pituitary development or function
90673	Hypothyroidism due to TSH receptor mutations
163690	Hypotonia - cystinuria syndrome
79507	Hypotonia - failure to thrive - microcephaly
91131	Hypotonia and ichthyosis due to dolichol phosphate deficiency
137908	Hypotonia with lactic acidemia and hyperammonemia
91132	Hypotrichosis - congenital ichthyosis
69735	Hypotrichosis - lymphedema - telangiectasia
55654	Hypotrichosis simplex
90368	Hypotrichosis simplex of the scalp
1573	Hypotrichosis with juvenile macular degeneration
1573	Hypotrichosis with juvenile macular dystrophy
444	Hypotrichosis, Marie Unna type
330029	Hypotrichosis-deafness syndrome
2266	Hypotrichosis-intellectual deficit, Lopes type
307936	Hypotrichosis-osteolysis-periodontitis-palmoplantar hyperkeratosis syndrome
307936	Hypotrichosis-osteolysis-periodontitis-palmoplantar keratoderma syndrome
307936	Hypotrichosis-striate palmoplantar hyperkeratosis-acroosteolysis-periodontitis syndrome
307936	Hypotrichosis-striate palmoplantar keratoderma-acroosteolysis-periodontitis syndrome
79233	Hypoxanthine guanine phosphoribosyltransferase 1 partial deficiency
510	Hypoxanthine guanine phosphoribosyltransferase complete deficiency
79233	Hypoxanthine guanine phosphoribosyltransferase deficiency, grade I
510	Hypoxanthine guanine phosphoribosyltransferase deficiency, grade IV
79233	Hypoxanthine guanine phosphoribosyltransferase partial deficiency
137577	Hypoxic and ischemic brain injury in the newborn
137577	Hypoxic-ischemic encephalopathy
682	HYPP
293168	IAHSP
254509	Iatrogenic botulism
453	IBIDS syndrome
611	IBM
602	IBM2

ORPHA Number	Disease name
79091	IBM3
52430	IBMPFD
1576	IBSN
31709	ICCA syndrome
64734	ICE syndrome
576	I-cell disease
2268	ICF syndrome
2269	Ichthyosis - alopecia - eclabion - ectropion - intellectual deficit
2274	Ichthyosis - hepatosplenomegaly - cerebellar degeneration
59303	Ichthyosis - hypotrichosis - sclerosing cholangitis
2278	Ichthyosis - intellectual deficit - dwarfism - renal impairment
2272	Ichthyosis - oral and digital anomalies
455	Ichthyosis bullosa of Siemens
457	Ichthyosis congenita, harlequin type
289586	Ichthyosis exfoliativa
457	Ichthyosis fetalis, Harlequin type
2273	Ichthyosis follicularis - alopecia - photophobia
2273	Ichthyosis follicularis - atrichia - photophobia
79504	Ichthyosis hystrix gravior
79503	Ichthyosis hystrix of Curth-Macklin
79503	Ichthyosis hystrix, Curth-Macklin type
88621	Ichthyosis prematurity syndrome
281190	Ichthyosis variegata
281190	Ichthyosis with confetti
79504	Ichthyosis, Lambert type
2267	Ichthyosis-cheek-eyebrow syndrome
91132	Ichthyosis-hypotrichosis syndrome
276	IDCS T-B+ par déficit en chaîne gamma
289347	IDH
73014	IDI
3306	idic(15)
930	Idiopathic achalasia
724	Idiopathic acute eosinophilic pneumonia
139423	Idiopathic acute transverse myelitis
422	Idiopathic and/or familial pulmonary arterial hypertension
280914	Idiopathic anterior uveitis
88	Idiopathic aplastic anemia
1980	Idiopathic basal ganglia calcification
171684	Idiopathic bilateral vestibulopathy
84065	Idiopathic bile acid malabsorption
60033	Idiopathic bronchiectasis
188	Idiopathic capillary leak syndrome
163703	Idiopathic catastrophic epileptic encephalopathy
228000	Idiopathic CD4 lymphocytopenia
169615	Idiopathic central precocious puberty

ORPHA Number	Disease name
2902	Idiopathic chronic eosinophilic pneumonia
95717	Idiopathic congenital hypothyroidism
209919	Idiopathic copper-associated cirrhosis
35066	Idiopathic cutaneous and mucosal candidiasis
35066	Idiopathic cutaneous and mucosal candidosis
256	Idiopathic dystonia
247724	Idiopathic eosinophilic myositis
2810	Idiopathic facial palsy
329874	Idiopathic giant cell myocarditis
64722	Idiopathic granulomatous mastitis
89845	Idiopathic hydrops fetalis
2197	Idiopathic hypercalciuria
3260	Idiopathic hypereosinophilic syndrome
33208	Idiopathic hypersomnia
228315	Idiopathic hypersomnia with long sleep time
228318	Idiopathic hypersomnia without long sleep time
1572	Idiopathic immunoglobulin deficiency
51608	Idiopathic infantile arterial calcification
35062	Idiopathic infection disseminated by cytomegalovirus
238624	Idiopathic intracranial hypertension
85193	Idiopathic juvenile osteoporosis
314017	Idiopathic linear interstitial keratitis
33577	Idiopathic lobular panniculitis
90158	Idiopathic localized lipodystrophy
353344	Idiopathic macular telangiectasia type 1
353351	Idiopathic macular telangiectasia type 3
84065	Idiopathic malabsorption due to bile acid synthesis defects
73	Idiopathic massive osteolysis
97560	Idiopathic membranous glomerulonephritis
2774	Idiopathic multicentric osteolysis with or without nephropathy
824	Idiopathic myelofibrosis
45452	Idiopathic neonatal atrial flutter
33577	Idiopathic nodular panniculitis
51608	Idiopathic obliterative arteriopathy
441	Idiopathic orthostatic hypotension
280921	Idiopathic panuveitis
747	Idiopathic PAP
280917	Idiopathic posterior uveitis
747	Idiopathic pulmonary alveolar proteinosis
275766	Idiopathic pulmonary arterial hypertension
1676	Idiopathic pulmonary artery dilatation
2032	Idiopathic pulmonary fibrosis
99931	Idiopathic pulmonary hemosiderosis

ORPHA Number	Disease name
35061	Idiopathic recurrent and disabling cutaneous herpes
251307	Idiopathic recurrent pericarditis
276174	Idiopathic recurrent stupor
251307	Idiopathic relapsing pericarditis
209943	Idiopathic retinal-aneurysms-neuroretinitis syndrome
35065	Idiopathic severe pneumococcemia
69061	Idiopathic steroid-sensitive nephrotic syndrome
93209	Idiopathic steroid-sensitive nephrotic syndrome with diffuse mesangial proliferation
93206	Idiopathic steroid-sensitive nephrotic syndrome with focal segmental glomerulosclerosis
93206	Idiopathic steroid-sensitive nephrotic syndrome with focal segmental hyalinosis
93207	Idiopathic steroid-sensitive nephrotic syndrome with minimal change
99858	Idiopathic syringomyelia
256	Idiopathic torsion dystonia
98806	Idiopathic torsion dystonia of mixed type
209956	Idiopathic uveal effusion syndrome
130	Idiopathic ventricular fibrillation, Brugada type
228140	Idiopathic ventricular fibrillation, not Brugada type
280384	IDMDC
580	Iduronate 2-sulfatase deficiency
217085	Iduronate 2-sulfatase deficiency type A
217093	Iduronate 2-sulfatase deficiency type B
92050	IED
2273	IFAP syndrome
332	IFD
99972	IgA1 deficiency
99973	IgA2 deficiency
329874	IGCM
79099	IGDA
73272	IGF-1 deficiency
183675	IgG subclass deficiency with IgA subclass deficiency
79078	IgG4-related dacryoadenitis and sialoadenitis
284264	IgG4-related sclerosing disease
284264	IgG4-related systemic disease
329903	Ig-mediated membranoproliferative glomerulonephritis
329903	Ig-mediated MPGN
59303	IHSC
238624	IIH
85193	IJO
100078	Ileal endocrine tumor
1150	Illum syndrome
85173	IMAGe syndrome
1534	Imaizumi-Kuroki syndrome
247718	IMAM
42062	Iminoglycinuria
284362	Immature interstitial mesenchymal tumor
289465	Immigration delay disease
244	Immotile cilia syndrome
2901	Immune brachial plexus neuropathy
169090	Immune dysfunction due to T-cell inactivation due to calcium entry defect
3002	Immune thrombocytopenia
3002	Immune thrombocytopenic purpura
206569	Immune-mediated necrotizing myopathy
94075	Immune-mediated protracted diarrhea of infancy
206575	Immune-mediated rippling muscle disease
86886	Immunoblastic lymphadenopathy
2268	Immunodeficiency - centromeric instability - facial anomalies
647	Immunodeficiency - microcephaly - chromosomal instability
935	Immunodeficiency - short limb dwarfism
34592	Immunodeficiency by defective expression of HLA class 1
572	Immunodeficiency by defective expression of HLA class 2
169147	Immunodeficiency due to a C1, C4, or C2 component complement deficiency
169150	Immunodeficiency due to a C5 to C9 component complement deficiency
169150	Immunodeficiency due to a late component of complements deficiency
169147	Immunodeficiency due to an early component of complement deficiency
169100	Immunodeficiency due to CD25 deficiency
331190	Immunodeficiency due to ficolin3 deficiency
70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency
331187	Immunodeficiency due to MASP-2 deficiency
70593	Immunodeficiency due to selective anti-polysaccharide antibody deficiency
200421	Immunodeficiency with factor H anomaly
200418	Immunodeficiency with factor I anomaly
75391	Immunodeficiency with natural-killer cell deficiency
99972	Immunoglobulin A1 deficiency
99973	Immunoglobulin A2 deficiency
284264	Immunoglobulin G4-related sclerosing disease
169110	Immunoglobulin heavy chain deficiency
85443	Immunoglobulinic amyloidosis
329903	Immunoglobulin-mediated membranoproliferative glomerulonephritis
329903	Immunoglobulin-mediated MPGN
103915	Immunoproliferative small intestinal disease
97567	Immunotactoid glomerulopathy
91137	Immunotactoid or fibrillary glomerulonephritis
91137	Immunotactoid or fibrillary glomerulopathy
2759	Imperforate oropharynx - costo vetebral anomalies
71276	Imploding antrum syndrome
35069	INAD
35069	INAD1
254509	Inadvertent botulism
45453	Incessant infant ventricular tachycardia
79263	INCL
231226	Inclusion body beta-thalassemia
199267	Inclusion body fibromatosis
602	Inclusion body myopathy type 2
79091	Inclusion body myopathy type 3
52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia
611	Inclusion body myositis
254693	Incomplete hydatidiform mole
254693	Incomplete molar pregnancy
157769	Incomplete situs inversus
180079	Incomplete unilateral aplasia of the Müllerian duct
464	Incontinentia pigmenti
435	Incontinentia pigmenti type 1
464	Incontinentia pigmenti type 2
158019	Indeterminate cell histiocytosis
1388	Index finger anomaly - Pierre Robin syndrome
101335	Indian tick typhus
98848	Indolent systemic mastocytosis
1909	Indomethacin embryofetopathy
70587	Infant acute respiratory distress syndrome
70587	Infant ARDS
178478	Infant botulism
1943	Infant epilepsy with migrant focal crisis
178478	Infant intestinal botulism
178478	Infant intestinal toxemia botulism
178478	Infant intestinal toxin-mediated botulism
70587	Infant respiratory distress syndrome
247165	Infantile acrodynia
99725	Infantile and juvenile forms of acromegaly
70590	Infantile apnea
51608	Infantile arteriosclerosis
93591	Infantile autosomal recessive medullary cystic kidney disease

ORPHA Number	Disease name
2679	Infantile axonal neuropathy
89938	Infantile Bartter syndrome with deafness
1576	Infantile bilateral striatal necrosis
178478	Infantile botulism
314911	Infantile Canavan disease
137675	Infantile cardiomyopathy with histiocytoid change
217557	Infantile cellular interstitial pneumonitis
313850	Infantile cerebellar-retinal degeneration
77260	Infantile cerebral Gaucher disease
1313	Infantile choroido cerebral calcification syndrome
31709	Infantile convulsions and choreoathetosis
1310	Infantile cortical hyperostosis
199267	Infantile digital fibromatosis
87876	Infantile dysmorphic sialidosis
238455	Infantile dystonia-parkinsonism
300373	Infantile gigantism due to pituitary hyperplasia
289860	Infantile glycine encephalopathy
79255	Infantile GM1 gangliosidosis
309155	Infantile GM2 gangliosidosis 0 variant
293603	Infantile hereditary endothelial dystrophy
352563	Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency
247651	Infantile hypophosphatasia
79076	Infantile juvenile polyposis syndrome
206436	Infantile Krabbe disease
667	Infantile malignant osteopetrosis
247165	Infantile mercury intoxication
247165	Infantile mercury poisoning
2591	Infantile myofibromatosis
79263	Infantile NCL
35069	Infantile neuroaxonal dystrophy
79263	Infantile neuronal ceroid lipofuscinosis
251304	Infantile onset panniculitis with uveitis and systemic granulomatosis
1186	Infantile onset spinocerebellar ataxia
67047	Infantile optic atrophy with chorea and spastic paraplegia
85179	Infantile osteopetrosis with neuroaxonal dysplasia
247651	Infantile phosphoethanolaminuria
247651	Infantile Rathburn disease
772	Infantile Refsum disease
300293	Infantile regressive hypertriglyceridemia and fatty liver
300293	Infantile regressive hypertriglyceridemia and hepatosteatosis
254864	Infantile reversible cytochrome c oxidase deficiency myopathy
263410	Infantile spams - psychomotor retardation - progressive brain atrophy - basal ganglia disease
3451	Infantile spasms
3173	Infantile spasms - broad thumbs
83330	Infantile spinal muscular atrophy
1575	Infantile striato thalamic degeneration
1576	Infantile striatonigral degeneration
1576	Infantile striatonigral necrosis
506	Infantile subacute necrotizing encephalopathy
255241	Infantile subacute necrotizing encephalopathy with leukodystrophy
255249	Infantile subacute necrotizing encephalopathy with nephrotic syndrome
3311	Infantile symmetrical thalamic degeneration
2176	Infantile systemic hyalinosis
1577	Infantile thalamic degeneration
2768	Infantile tibia vara
137675	Infantile xanthomatous cardiomyopathy
293168	Infantile-onset ascending hereditary spastic paralysis
284332	Infantile-onset autosomal recessive nonprogressive cerebellar ataxia
1451	Infantile-onset multisystem inflammatory disease
171714	infantile-onset symptomatic epilepsy syndrome - developmental stagnation - blindness
178487	Infant-like botulism
137593	Infectious epithelial keratitis
289347	Infective dermatitis associated with HTLV-1
289347	Infective dermatitis associated with human T-lymphotropic virus type 1
289347	Infective dermatitis associated with human T-lymphotropic virus type I
99123	Inferior caval vein interruption
99123	Inferior vena cava interruption
280794	Infiltrative small vesicular DCM
280794	Infiltrative small vesicular diffuse cutaneous mastocytosis
85445	Inflammatory amyloidosis
178342	Inflammatory myofibroblastic tumor
160148	Inflammatory myoglandular polyps
247718	Inflammatory myopathy with abundant macrophages
48918	Inflammatory pseudotumor of skeletal muscle
90003	Inflammatory pseudotumor of the liver
238305	Infundibulo-neurohypophysitis
95513	Infundibulo-panhypophysitis
1849	Infundibulopelvic stenosis - multicystic kidney
254504	Inhalation botulism
247257	Inhalational anthrax
254504	Inhalational botulism
319465	Inherited acute myeloid leukemia
319465	Inherited AML
319462	Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations
282166	Inherited CJD
210141	Inherited congenital spastic quadriplegia
210141	Inherited congenital spastic tetraplegia
282166	Inherited Creutzfeldt-Jakob disease
100054	Inherited estrogen-associated angioedema
100054	Inherited estrogen-associated angioneurotic edema
100054	Inherited estrogen-dependent angioedema
100054	Inherited estrogen-dependent angioneurotic edema
71278	Inherited glutamine synthetase deficiency
71278	Inherited GS deficiency
289548	Inherited isolated adrenal insufficiency due to CYP11A1 deficiency
225968	Inherited predisposition to essential thrombocythemia
63259	Iniencephaly
178475	Inoculation botulism
642	Insensitivity to pain - anhidrosis
97279	Insulinoma
2297	Insulin-resistance syndrome type A
2298	Insulin-resistance syndrome type B
97279	Insuloma
127	Intelectual deficiency - epilepsy - endocrine disorders
2466	Intellectual defici-aphasia-shuffling gait-adducted thumbs syndrome
356996	Intellectual deficiency - hypotonia - spasticity - sleep disorder
289483	Intellectual deficit - alacrima - achalasia
1236	Intellectual deficit - athetosis - microphthalmia
3041	Intellectual deficit - balding - patella luxation - acromicria
3042	Intellectual deficit - cataracts - calcified pinnae - myopathy
171860	Intellectual deficit - cataracts - kyphosis
2474	Intellectual deficit - coloboma - slimness
329224	Intellectual deficit - craniofacial dysmorphism - cryptorchidism
3044	Intellectual deficit - dysmorphism - hypogonadism - diabetes mellitus
171851	Intellectual deficit - enteropathy - deafness - peripheral neuropathy - ichthyosis - keratodermia
2139	Intellectual deficit - epilepsy - bulbous nose
1495	Intellectual deficit - hypoplastic corpus callosum - preauricular tag
166108	Intellectual deficit - hypotonia - facial dysmorphism
3050	Intellectual deficit - hypotonia - skin hyperpigmentation

ORPHA Number	Disease name
3451	Intellectual deficit - hypsarrhythmia
3067	Intellectual deficit - microcephaly - phalangeal - facial abnormalities
3313	Intellectual deficit - microcephaly - unusual facies
3068	Intellectual deficit - myopathy - short stature - endocrine defect
3071	Intellectual deficit - nasal papillomata
352530	Intellectual deficit - obesity - brain malformations - facial dysmorphism
3082	Intellectual deficit - polydactyly - uncombable hair
3409	Intellectual deficit - short stature - hand contractures - genital anomalies
3074	Intellectual deficit - short stature - hypertelorism
1240	Intellectual deficit - short stature - wedge shaped epiphyses of knees
3051	Intellectual deficit - sparse hair - brachydactyly
1891	Intellectual deficit - spasticity - ectrodactyly
75858	Intellectual deficit - truncal obesity - retinal dystrophy - micropenis
3043	Intellectual deficit - unusual facies
100973	Intellectual deficit associated with fragile site FRAXE
166108	Intellectual deficit, Birk-Barel type
3079	Intellectual deficit, Buenos-Aires type
2557	Intellectual deficit, Mietens-Weber type
3080	Intellectual deficit, Wolff type
85327	Intellectual deficit, X-linked - acromegaly - hyperactivity
85295	Intellectual deficit, X-linked - choreoathetosis - abnormal behavior
163979	Intellectual deficit, X-linked - craniofacioskeletal syndrome
85280	Intellectual deficit, X-linked - cubitus valgus - dysmorphism
1568	Intellectual deficit, X-linked - Dandy-Walker malformation - basal ganglia disease - Seizures
2958	Intellectual deficit, X-linked - dysmorphism - cerebral atrophy
85319	Intellectual deficit, X-linked - epilepsy - progressive joint contractures - dysmorphism
85317	Intellectual deficit, X-linked - hypogammaglobulinemia - progressive neurological deterioration
85331	Intellectual deficit, X-linked - hypogonadism - ichthyosis - obesity - short stature
85329	Intellectual deficit, X-linked - hypotonia - facial dysmorphism - aggressive behavior
85320	Intellectual deficit, X-linked - macrocephaly - macro-orchidism
2898	Intellectual deficit, X-linked - plagiocephaly
85318	Intellectual deficit, X-linked - precocious puberty - obesity
3077	Intellectual deficit, X-linked - psychosis - macroorchidism
85332	Intellectual deficit, X-linked - retinitis pigmentosa
3052	Intellectual deficit, X-linked - seizures - psoriasis
3055	Intellectual deficit, X-linked - short stature - obesity
85273	Intellectual deficit, X-linked, Abidi type
85276	Intellectual deficit, X-linked, Armfield type
3056	Intellectual deficit, X-linked, Brooks type
85277	Intellectual deficit, X-linked, Cantagrel type
163971	Intellectual deficit, X-linked, Cilliers type
3059	Intellectual deficit, X-linked, Gu type
163961	Intellectual deficit, X-linked, Kroes type
775	Intellectual deficit, X-linked, Martinez type
85283	Intellectual deficit, X-linked, Miles-Carpenter type
163956	Intellectual deficit, X-linked, Nascimento type
85322	Intellectual deficit, X-linked, Pai type
163953	Intellectual deficit, X-linked, Raymond type
3061	Intellectual deficit, X-linked, Raynaud type
85285	Intellectual deficit, X-linked, Schimke type
3062	Intellectual deficit, X-linked, Schutz type
85323	Intellectual deficit, X-linked, Seemanova type
85286	Intellectual deficit, X-linked, Shashi type
85324	Intellectual deficit, X-linked, Shrimpton type
85287	Intellectual deficit, X-linked, Siderius type
3063	Intellectual deficit, X-linked, Snyder type
85325	Intellectual deficit, X-linked, Stevenson type
85288	Intellectual deficit, X-linked, Stocco Dos Santos type
85328	Intellectual deficit, X-linked, Turner type
163976	Intellectual deficit, X-linked, Van Esch type
85289	Intellectual deficit, X-linked, Vitale type
85290	Intellectual deficit, X-linked, Wilson type
3064	Intellectual deficit, X-linked, Wittner type
85291	Intellectual deficit, X-linked, Wittwer type
85337	Intellectual deficit, X-linked, Zorick type
314575	Intellectual deficit-hypotonia-brachycephaly-pyloric stenosis-cryptorchidism syndrome
3046	Intellectual deficit-unusual facies, Davis-Lafer type
1478	Interauricular communication
51890	Intercostal nerve syndrome
86900	Interdigitating cell sarcoma
86900	Interdigitating dendritic cell sarcoma
210115	Interleukin-1 receptor antagonist deficiency
169100	Interleukin-2 receptor alpha chain deficiency
268162	Intermediate BCKD deficiency
268162	Intermediate branched-chain ketoacid dehydrogenase deficiency
268162	Intermediate branched-chain ketoaciduria
99989	Intermediate DEND syndrome
268162	Intermediate leucinosis
268162	Intermediate maple syrup urine disease
268162	Intermediate MSUD
171433	Intermediate nemaline myopathy
210110	Intermediate osteopetrosis
309331	Intermediate severe Salla disease
83418	Intermediate spinal muscular atrophy
99134	Intermediate stomatocytosis syndrome
268173	Intermittent BCKD deficiency
268173	Intermittent branched-chain ketoacid dehydrogenase deficiency
268173	Intermittent branched-chain ketoaciduria
329967	Intermittent hydrarthrosis
268173	Intermittent leucinosis
268173	Intermittent maple syrup urine disease
268173	Intermittent MSUD
981	Internal carotid agenesis
37202	Interstitial cystitis
79099	Interstitial granulomatous dermatitis with arthritis
1480	Interventricular communication
99092	Interventricular septum aneurysm
178481	Intestinal botulism
178481	Intestinal colonization botulism
92050	Intestinal epithelial dysplasia
30924	Intestinal hypomagnesemia with secondary hypocalcemia
3452	Intestinal lipodystrophy
3452	Intestinal lipophagic granulomatosis
36204	Intestinal lymphangiectasia
314376	Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency
2869	Intestinal polyposis - cutaneous pigmentation syndrome
86880	Intestinal T-cell lymphoma
178481	Intestinal toxemia botulism
178481	Intestinal toxin-mediated botulism
228371	Intoxication botulism
1057	Intracranial aneurysms - multiple congenital anomalies
46724	Intracranial arteriovenous malformation

ORPHA Number	Disease name
48736	Intracranial embryonal carcinoma
252006	Intracranial endodermal sinus tumor
91352	Intracranial germinoma
252006	Intracranial yolk sac tumor
137622	Intractable diarrhea - choanal atresia - eye anomalies
73014	Intractable diarrhea of infancy
69665	Intrahepatic cholestasis of pregnancy
280802	Intralobar congenital bronchopulmonary sequestration
280802	Intralobar congenital pulmonary sequestration
99088	Intramural coronary arterial course
100003	Intraneural perineurioma
268139	Intraocular medulloepithelioma
140436	Intraosseous hemangioma
137686	Intrauterine adhesions
85173	Intrauterine growth retardation - metaphyseal dysplasia - adrenal hypoplasia congenita - genital anomalies
137686	Intrauterine synechiae
98839	Intravascular large B-cell lymphoma
98839	Intravascular lymphomatosis
332	Intrinsic factor deficiency
3306	Inv dup(15)
90078	Invasive infections due to vancomycin-resistant enterococci
90078	Invasive infections due to VRE
99925	Invasive mole
324648	Invasive non-typhoidal salmonellosis
96092	Invdupdel(8p)
79405	Inverse JEB
329324	Inverse Klippel-Trénaunay syndrome
98951	Inverse Marcus-Gunn phenomenon
79409	Inverse RDEB
79409	Inverse recessive dystrophic epidermolysis bullosa
96092	Inverted 8p duplication/deletion syndrome
2704	Inverted smile - neurogenic bladder
1451	IOMID syndrome
1186	IOSCA
275766	IPAH
37042	IPEX
88621	IPS
103915	IPSID
70592	IRAK4 deficiency
772	IRD
209981	IRIDA syndrome
64734	Iridocorneal endothelial syndrome
2995	Iris coloboma-ptosis-intellectual deficit syndrome
39044	Iris melanoma
209981	Iron-refractory iron deficiency anemia
86915	Irons-Bianchi syndrome
43115	Iron-sulphur cluster deficiency myopathy
209943	IRVAN syndrome
84142	Isaac-Mertens syndrome
84142	Isaac's syndrome
972	Isaacs-Mertens syndrome
1509	Ischiopatellar dysplasia
85200	Ischio-spinal dysostosis
85200	Ischio-vertebral dysplasia
85200	Ischio-vertebral syndrome
43115	ISCU myopathy
79159	Isobutyric aciduria
79159	Isobutyryl-CoA dehydrogenase deficiency
3309	Isochromosome 5p
3310	Isochromosome 9p
884	Isochromosome 12p mosaicism
884	Isochromosome 12p syndrome
3307	Isochromosome 18p
96055	Isochromosome 21
96325	Isochromosome Y
98797	Isochromosomy Yp
98798	Isochromosomy Yq
99731	ISOD
3306	Isodicentric 15 chromosome
79144	Iso-Kikuchi syndrome
6	Isolated 3-methylcrotonyl-CoA carboxylase deficiency
263524	Isolated acute necrotizing encephalopathy
289465	Isolated adermatoglyphia
229717	Isolated agammaglobulinemia
180188	Isolated amastia
268868	Isolated amyelia
263524	Isolated ANE
1048	Isolated anencephaly/exencephaly
140989	Isolated angiitis of the central nervous system
250923	Isolated aniridia
91397	Isolated ankyloblepharon filiforme adnatum
2542	Isolated anophthalmia - microphthalmia
557	Isolated anorectal malformation
3387	Isolated anterior cervical hypertrichosis
162516	Isolated apertura pyriformis stenosis
268936	Isolated arhinencephaly
1166	Isolated asymmetric crying facies
206599	Isolated asymptomatic elevation of creatine phosphokinase
254913	Isolated ATP synthase deficiency
199326	Isolated autosomal dominant hypomagnesemia, Glaudemans type
269221	Isolated bilateral hemispheric cerebellar hypoplasia
158778	Isolated bone marrow mastocytosis
35099	Isolated brachycephaly
180188	Isolated breast aplasia
1398	Isolated cerebellar hypoplasia/agenesis
269203	Isolated cerebellar vermis agenesis
199630	Isolated cerebellar vermis hypoplasia
2343	Isolated cloverleaf skull syndrome
1460	Isolated coenzyme Q-cytochrome C reductase deficiency
217059	Isolated congenital acropachy
91416	Isolated congenital alacrima
88620	Isolated congenital anosmia
162526	Isolated congenital auditory ossicle malformation
238722	Isolated congenital controlateral synkinesia
217059	Isolated congenital digital clubbing
99171	Isolated congenital ectropion
432	Isolated congenital gonadotropin deficiency
141152	Isolated congenital hypoglossia/aglossia
91489	Isolated congenital megalocornea
238722	Isolated congenital mirror movements
217059	Isolated congenital nail clubbing
162516	Isolated congenital nasal pyriform aperture stenosis
91490	Isolated congenital sclerocornea
216718	Isolated congenitally uncorrected transposition of the great arteries
216718	Isolated congenitally uncorrected transposition of the great vessels
1460	Isolated CoQ-cytochrome C reductase deficiency
254905	Isolated COX deficiency
91396	Isolated cryptophthalmia
254905	Isolated cytochrome C oxidase deficiency
217	Isolated Dandy-Walker malformation
269212	Isolated Dandy-Walker malformation with hydrocephalus
269215	Isolated Dandy-Walker malformation without hydrocephalus
248340	Isolated delta-SPD
248340	Isolated delta-storage pool disease
248340	Isolated dense-SPD
248340	Isolated dense-storage pool disease
99177	Isolated distichiasis
35093	Isolated dolichocephaly
1885	Isolated ectopia lentis
221106	Isolated facial myokymia
65683	Isolated focal cortical dysplasia
268961	Isolated focal cortical dysplasia type I
268973	Isolated focal cortical dysplasia type Ia
268980	Isolated focal cortical dysplasia type Ib
268987	Isolated focal cortical dysplasia type Ic

ORPHA Number	Disease name
268994	Isolated focal cortical dysplasia type II
269001	Isolated focal cortical dysplasia type IIa
269008	Isolated focal cortical dysplasia type IIb
52901	Isolated follicle stimulating hormone deficiency
52901	Isolated FSH deficiency
408	Isolated glycerol kinase deficiency
231662	Isolated growth hormone deficiency type IA
231671	Isolated growth hormone deficiency type IB
231679	Isolated growth hormone deficiency type II
231692	Isolated growth hormone deficiency type III
2128	Isolated hemihyperplasia
306527	Isolated hereditary congenital facial paralysis
183675	Isolated IgG subclass deficiency
2345	Isolated Klippel-Feil syndrome
1084	Isolated lissencephaly type 1 without known genetic defects
268920	Isolated macrencephaly
268920	Isolated megalencephaly
238593	Isolated mesenteric lipodystrophy
95707	Isolated micropenis
90641	Isolated mitochondrial neurosensory deafness
2609	Isolated mitochondrial respiratory chain complex I deficiency
3208	Isolated mitochondrial respiratory chain complex II deficiency
1460	Isolated mitochondrial respiratory chain complex III deficiency
254905	Isolated mitochondrial respiratory chain complex IV deficiency
254913	Isolated mitochondrial respiratory chain complex V deficiency
90641	Isolated mitochondrial sensorineural deafness
2609	Isolated NADH-coenzyme Q reductase deficiency
2609	Isolated NADH-CoQ reductase deficiency
2609	Isolated NADH-ubiquinone reductase deficiency
162516	Isolated nasal pyriform aperture hypoplasia
137902	Isolated optic nerve hypoplasia
166119	Isolated osteopoikilosis
63440	Isolated oxycephaly
269209	Isolated partial cerebellar vermis agenesis
96269	Isolated partial vaginal agenesis
718	Isolated Pierre Robin sequence
718	Isolated Pierre Robin syndrome
35098	Isolated plagiocephaly
2924	Isolated polycystic liver disease
2456	Isolated polythelia
216452	Isolated postlingual genetic deafness
216445	Isolated prelingual genetic deafness
238670	Isolated prothyroliberin deficiency
238670	Isolated protirelin deficiency
264691	Isolated pulmonary capillaritis
2542	Isolated pure microphthalmia
35093	Isolated scaphocephaly
823	Isolated spina bifida
178311	Isolated sternocostoclavicular hyperostosis
3208	Isolated succinate-coenzyme Q reductase deficiency
3208	Isolated succinate-CoQ reductase deficiency
3208	Isolated succinate-ubiquinone reductase deficiency
99731	Isolated sulfite oxidase deficiency
90674	Isolated thyroid-stimulating hormone deficiency
238670	Isolated thyroliberin deficiency
238670	Isolated thyrotropin-releasing factor deficiency
238670	Isolated thyrotropin-releasing hormone deficiency
269206	Isolated total cerebellar vermis agenesis
103909	Isolated trehalose intolerance
238670	Isolated TRF deficiency
238670	Isolated TRH deficiency
3366	Isolated trigonocephaly
90674	Isolated TSH deficiency
238670	Isolated TSH-releasing factor deficiency
1460	Isolated ubiquinone-cytochrome C reductase deficiency
269218	Isolated unilateral hemispheric cerebellar hypoplasia
96	Isolated vitamin E deficiency
472	Isosporiasis
2305	Isotretinoin embryopathy
2305	Isotretinoin syndrome
2306	Isotretinoin-like syndrome
33	Isovaleric acid CoA dehydrogenase deficiency
33	Isovaleric acidemia
309324	ISSD
2739	Itin syndrome
435	Ito hypomelanosis
3002	ITP
99123	IVC interruption
294415	Ivemark II syndrome
97548	Ivemark syndrome
2307	IVIC syndrome
281190	IWC
3236	Jackson-Barr syndrome
1540	Jackson-Weiss syndrome
2848	Jacobs syndrome
2308	Jacobsen syndrome
1941	JAE
2029	Jaffe-Campanacci syndrome
93276	Jaffe-Lichtenstein disease
2269	Jagell-Holmgren-Hofer syndrome
1873	Jalili syndrome
300605	JALS
73423	Jamaican vomiting sickness
73423	Jamaican vomiting syndrome
1891	Jancar syndrome
2590	Jankovic-Rivera syndrome
168491	Jansky-Bielschowsky disease
79139	Japanese encephalitis
2311	Jarcho-Levin syndrome
474	JATD
313795	Jawad syndrome
91412	Jaw-winking syndrome
139431	Jeavons syndrome
305	JEB
79404	JEB-H
79405	JEB-I
79406	JEB-lo
79402	JEB-nH gen
251393	JEB-nH loc
79403	JEB-PA
1201	Jejunal atresia
100077	Jejunal endocrine tumor
89840	JEN-nH
90647	Jervell and Lange-Nielsen syndrome
33314	Jessner-Kanof lymphocytic infiltration of the skin
33314	Jessner's benign lymphocytic infiltration of the skin
33314	Jessner's lymphocytic infiltration of the skin
3283	JET
474	Jeune asphyxiating thoracic dystrophy
474	Jeune syndrome
248111	JHD
2929	JIP
65684	JMADUE
324999	JMP syndrome
289596	JNA
79264	JNCL
2314	Job syndrome
2315	Johanson-Blizzard syndrome
85320	Johnson syndrome
2316	Johnson-McMillin syndrome

ORPHA Number	Disease name
1112	Johnson-Munson syndrome
1485	Johnston-Aarons-Schelley syndrome
324999	Joint contractures-muscular atrophy-microcytic anemia-panniculitis-associated lipodystrophy syndrome
2295	Joint instability syndrome
2027	Jones syndrome
1256	Jorgenson-Lenz syndrome
475	Joubert syndrome
475	Joubert syndrome type A
1454	Joubert syndrome with congenital hepatic fibrosis
1454	Joubert syndrome with hepatic defect
220493	Joubert syndrome with ocular defect
2318	Joubert syndrome with oculorenal defect
2754	Joubert syndrome with orofaciodigital defect
220497	Joubert syndrome with renal defect
220493	Joubert syndrome with retinopathy
2318	Joubert syndrome with Senior-Loken syndrome
475	Joubert-Boltshauser syndrome
2801	JPG
2929	JPS
2318	JS type B
1454	JS-H
220493	JS-O
2318	JS-OR
220497	JS-R
2319	Juberg-Hayward syndrome
93972	Juberg-Marsidi syndrome
3283	Junctional ectopic tachycardia
305	Junctional epidermolysis bullosa
79403	Junctional epidermolysis bullosa - pyloric atresia
79404	Junctional epidermolysis bullosa generalisata gravis
79402	Junctional epidermolysis bullosa generalisata mitis
79405	Junctional epidermolysis bullosa inversa
79402	Junctional epidermolysis bullosa, Disentis type
79404	Junctional epidermolysis bullosa, Herlitz type
79404	Junctional epidermolysis bullosa, Herlitz-Pearson type
89840	Junctional epidermolysis bullosa, non-Herlitz type
2321	Jung-Wolff-Back-Stahl syndrome
319223	Junin hemorrhagic fever
989	Jussieu syndrome
1941	Juvenile absence epilepsy
300605	Juvenile amyotrophic lateral sclerosis
199260	Juvenile aponeurotic fibromatosis
93592	Juvenile autosomal recessive medullary cystic kidney disease
314918	Juvenile Canavan disease
247794	Juvenile cataract - microcornea - renal glucosuria
300605	Juvenile Charcot disease
86834	Juvenile chronic myelomonocytic leukemia
2778	Juvenile chronic recurrent multifocal osteomyelitis
2778	Juvenile CRMO
93672	Juvenile dermatomyositis
93672	Juvenile DM
228254	Juvenile elastoma without osteopoikilosis
2929	Juvenile gastrointestinal polyposis
98977	Juvenile glaucoma
79256	Juvenile GM1 gangliosidosis
309162	Juvenile GM2 gangliosidosis 0 variant
79230	Juvenile hemochromatosis
98954	Juvenile hereditary epithelial dystrophy of Meesmann
248111	Juvenile Huntington chorea
248111	Juvenile Huntington disease
2028	Juvenile hyaline fibromatosis
2929	Juvenile intestinal polyposis
300605	Juvenile Lou-Gehrig disease
65684	Juvenile muscular atrophy of distal upper extremity
65684	Juvenile muscular atrophy of the distal upper limb
86834	Juvenile myelomonocytic leukemia
307	Juvenile myoclonic epilepsy
289596	Juvenile nasopharyngeal angiofibroma
79264	Juvenile NCL
79264	Juvenile neuronal ceroid lipofuscinosis
85193	Juvenile osteoporosis
329894	Juvenile overlap myositis
2801	Juvenile Paget disease
2801	Juvenile Paget's disease
352497	Juvenile parkinsonism with intellectual deficit due to DNAJC6 deficiency
247604	Juvenile PLS
93568	Juvenile PM
93568	Juvenile polymyositis
79076	Juvenile polyposis of infancy
2929	Juvenile polyposis syndrome
247604	Juvenile primary lateral sclerosis
85436	Juvenile psoriatic arthritis
85408	Juvenile rheumatoid factor-negative polyarthritis
247854	Juvenile rheumatoid factor-negative polyarthritis with anti-nuclear antibodies
247861	Juvenile rheumatoid factor-negative polyarthritis without anti-nuclear antibodies
85435	Juvenile rheumatoid factor-positive polyarthritis
93399	Juvenile sialidosis type 2
83419	Juvenile spinal muscular atrophy
209341	Juvenile spinal muscular atrophy, autosomal dominant
85438	Juvenile spondylarthropathy
26137	Juvenile temporal arteritis
158000	Juvenile xanthogranuloma
79241	Juvenile-onset multiple carboxylase deficiency
324989	Juvenile-onset SAPHO syndrome
324989	Juvenile-onset synovitis-acne-pustulosis-hyperostosis-osteitis syndrome
99100	Juxtaposition of the atrial appendages
99100	Juxtaposition of the atrial auricles
1540	JWS
612	K+-aggravated myotonia
2322	Kabuki make-up syndrome
2322	Kabuki syndrome
85146	Kaeser syndrome
612	K-aggravated myotonia
29073	Kahler's disease
2324	Kaler-Garrity-Stern syndrome
2325	Kallin syndrome
478	Kallmann syndrome
2326	Kallmann syndrome - heart disease
99179	Kandori fleck retina
1836	Kantaputra mesomelic dysplasia
79280	Kanzaki disease
949	Kaplan-Plauchu-Fitch syndrome
2122	Kaposiform hemangioendothelioma
33276	Kaposi's sarcoma
91136	Kappa light chain-associated Fanconi syndrome
183675	Kappa-chain deficiency
2328	Kapur-Toriello syndrome
1381	Karandikar-Maria-Kamble syndrome
2329	Karsch-Neugebauer syndrome
2330	Kasabach-Merritt syndrome
1137	Kashani-Strom-Utley syndrome
1894	Kasznica-Carlson-Coppedge syndrome
3360	Katsantoni-Papadakou Lagoyanni syndrome
2473	Kaufman-Mckusick syndrome
2331	Kawasaki disease
2306	Kawashima syndrome
2533	Kawashima-Tsuji syndrome
2332	KBG syndrome
96169	KdVS

ORPHA Number	Disease name
480	Kearns-Sayre syndrome
199260	Keasby tumor
2662	Keipert syndrome
79233	Kelley-Seegmiller syndrome
137653	Kelly-Kirson-Wyatt syndrome
54028	Kelly-Paterson syndrome
481	Kennedy disease
64542	Kennedy-Teebi syndrome
991	Kennerknecht syndrome
2333	Kenny syndrome
2333	Kenny-Caffey syndrome
101336	Kenya tick typhus
101336	Kenya tick-bite fever
477	Keratitis - ichthyosis - deafness/Hystrix-like ichthyosis - deafness
79395	Keratoderma - ichthyosiform dermatosis - elevated beta-glucuronidase
494	Keratoderma hereditarium mutilans
79395	Keratoderma hereditarium mutilans with ichthyosis
79501	Keratodermia palmoplantaris papulosa, Buschke-Fischer-Brauer type
50943	Keratolytic winter erythema
495	Keratosis extremitatum hereditaria progrediens
218	Keratosis follicularis
2339	Keratosis follicularis - dwarfism - cerebral atrophy
2340	Keratosis follicularis spinulosa decalvans
281201	Keratosis linearis-ichthyosis congenita-sclerosing keratoderma syndrome
86919	Keratosis palmaris et plantaris - clinodactyly
678	Keratosis palmoplantar - periodontopathy
28378	Keratosis palmoplantaris - corneal dystrophy
50944	Keratosis palmoplantaris - cystic eyelids - hypodontia - hypotrichosis
2342	Keratosis palmoplantaris - periodontopathia - onychogryposis
79141	Keratosis palmoplantaris nummularis
50942	Keratosis palmoplantaris striata
50942	Keratosis palmoplantaris striata et areata
495	Keratosis palmoplantaris transgrediens et progrediens
87503	Keratosis palmoplantaris transgrediens of Siemens
50942	Keratosis palmoplantaris varians of Wachters
34217	Keratosis palmoplantaris with arrythmogenic cardiomyopathy
2198	Keratosis palmoplantaris-esophageal carcinoma syndrome
498	Keratosis pilaris atrophicans
499	Kerion celsi
3351	Kersey syndrome
293807	Ketamine-induced biliary dilatation
134	Ketoacidosis due to beta-ketothiolase deficiency
1399	Ketoaciduria - intellectual deficit - ataxia - deafness
2056	Ketohexokinase deficiency
35	Ketotic glycinemia
85202	Keutel syndrome
2988	Khalifa-Graham syndrome
477	KID syndrome
477	KID/HID syndrome
97332	Kienbock disease
50918	Kikuchi disease
50918	Kikuchi-Fujimoto disease
482	Kimura disease
2908	Kindler syndrome
99741	King-Denborough syndrome
565	Kinky hair disease
565	Kinky hair syndrome
1183	Kinsbourne syndrome
100996	Kjellin syndrome
98673	Kjer disease
99978	Klatskin tumor
2343	Kleeblattschaedel syndrome
261494	Kleefstra syndrome
96147	Kleefstra syndrome due to 9q subtelomeric deletion
96147	Kleefstra syndrome due to 9q34 microdeletion
261652	Kleefstra syndrome due to a point mutation
96147	Kleefstra syndrome due to del(9)(q34)
96147	Kleefstra syndrome due to monosomy 9q34
33543	Kleine-Levin syndrome
2110	Kleiner-Holmes syndrome
896	Klein-Waardenburg syndrome
281201	KLICK syndrome
2578	Klippel-Feil deformity - conductive deafness - absent vagina
2345	Klippel-Feil malformation
2345	Klippel-Feil sequence
90308	Klippel-Trénaunay syndrome
2346	Klippel-Trénaunay-Weber syndrome
157823	Klüver-Bucy syndrome
485	Kniest dysplasia
1571	Knobloch syndrome
1571	Knobloch-Layer syndrome
2698	Knuckle pads-leuconychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome
2698	Knuckle pads-leuconychia-sensorineural deafness-palmoplantar keratoderma syndrome
2349	Kocher-Debré-Semelaigne syndrome
679	Köhlmeier-Degos disease
679	Köhlmeier-Degos-Delort-Tricort syndrome
1946	Kohlschutter-Tonz syndrome
3197	Kok disease
51890	Komar syndrome
99077	Kommerell diverticulum
3212	Konigsmark-Knox-Hussels syndrome
96169	Koolen-De Vries syndrome
2892	Kopysc-Barczyk-Krol syndrome
2839	Kosenow syndrome
99749	Kostmann syndrome
1129	Kosztolanyi syndrome
2351	Kousseff syndrome
99741	Koussef-Nichols syndrome
629	Kowarski syndrome
2352	Kozlowski-Brown-Hardwick syndrome
3082	Kozlowski-Krajewska syndrome
2204	Kozlowski-Tsuruta syndrome
487	Krabbe disease
206436	Krabbe disease, classic form
206436	Krabbe disease, early-onset
206443	Krabbe disease, late-onset
1345	Krasnow-Qazi syndrome
284149	Kreiborg-Pakistani syndrome
2908	KS
293936	KTCNCT
306674	Kufor-Rakeb syndrome
79262	Kufs disease
83419	Kugelberg-Welander disease
209341	Kugelberg-Welander disease, autosomal dominant
2505	Kunze-Riehm syndrome
1149	Kuskokwim disease
767	Küssmaul-Maier disease
2798	Kuzniecky syndrome
319254	Kyasanur forest disease
319254	Kyasanur hemorrhagic fever
79155	Kynureninase deficiency
1801	Kyphomelic dysplasia
275543	L1 syndrome
275543	L1CAM syndrome
79314	L-2-HGA
79314	L-2-hydroxyglutaric acidemia
79314	L-2-hydroxyglutaric aciduria
53696	LAAHD
3473	Laband syndrome
2363	Lacrimo-auriculo-dento-digital syndrome

ORPHA Number	Disease name
2363	Lacrimo-auriculo-radio-dental syndrome
284426	Lactate dehydrogenase A deficiency
284435	Lactate dehydrogenase B deficiency
2965	Lactotroph adenoma
2968	LAD
99844	LAD-1 variant
2363	LADD syndrome
1484	Ladda-Zonana-Ramer syndrome
99842	LAD-I
99843	LAD-II
99844	LAD-III
158687	LAEB
501	Lafora disease
1997	Lagophthalmia - cleft lip and palate
59135	Laing distal myopathy
59135	Laing early-onset distal myopathy
275761	LAL deficiency
538	LAM
306507	LAMB-2-related infantile-onset nephrotic syndrome
1296	Lambert syndrome
43393	Lambert-Eaton myasthenic syndrome
98995	Lamellar cataract
313	Lamellar ichthyosis
137871	Laminopathy type Decaudain-Vigouroux
137871	Laminopathy with severe metabolic syndrome and myopathy
90024	LAMM syndrome
98818	Landau-Kleffner syndrome
354	Landing disease
269	Landouzy-Dejerine myopathy
231031	Lane disease
2632	Langer mesomelic dysplasia
502	Langer-Giedion syndrome
389	Langerhans cell granulomatosis
264955	Langerhans cell granulomatosis in childhood and adulthood
264750	Langerhans cell granulomatosis specific to adulthood
264724	Langerhans cell granulomatosis specific to childhood
389	Langerhans cell histiocytosis
264955	Langerhans cell histiocytosis in childhood and adulthood
264750	Langerhans cell histiocytosis specific to adulthood
264724	Langerhans cell histiocytosis specific to childhood
86897	Langerhans cell sarcoma
2368	Laparoschisis
2363	LARD syndrome
98838	Large cell lymphoma of the mediastinum
626	Large congenital melanocytic nevus
633	Laron syndrome
220465	Laron syndrome with immunodeficiency
220465	Laron-like syndrome
633	Laron-type dwarfism
2370	Larsen-like osseous dysplasia - short stature
284139	Larsen-like syndrome, B3GAT3 type
2808	Laryngeal abductor paralysis
2375	Laryngeal abductor paralysis - intellectual deficit
2407	Laryngeal and ocular granulation tissue in children from the Indian subcontinent syndrome
93961	Laryngeal dyskinesia
93961	Laryngeal dystonia
100083	Laryngeal endocrine tumor
2372	Laryngocele
2407	Laryngo-onycho-cutaneous syndrome
137935	Laryngotracheal angioma
2004	Laryngo-tracheo-esophageal cleft
2005	Laryngo-tracheo-esophageal cleft - pulmonary hypoplasia
280205	Laryngo-tracheo-esophageal cleft type 0
93938	Laryngo-tracheo-esophageal cleft type 1
93939	Laryngo-tracheo-esophageal cleft type 2
93940	Laryngo-tracheo-esophageal cleft type 3
93941	Laryngo-tracheo-esophageal cleft type 4
2004	Laryngo-tracheo-esophageal diastema
1202	Larynx atresia
99824	Lassa fever
99824	Lassa hemorrhagic fever
98974	Late hereditary endothelial dystrophy
168491	Late infantile NCL
168491	Late infantile neuronal ceroid lipofuscinosis
98816	Late onset benign childhood occipital epilepsy
79256	Late-infantile GM1 gangliosidosis
206443	Late-infantile/juvenile Krabbe disease
93589	Late-onset autosomal recessive medullary cystic kidney disease
247573	Late-onset citrullinemia type 1
247573	Late-onset citrullinemia type I
228227	Late-onset focal dermal elastosis
163708	Late-onset infantile spasms
199299	Late-onset isolated ACTH deficiency
79406	Late-onset junctional epidermolysis bullosa
231556	Late-onset localized junctional epidermolysis bullosa - intellectual deficit
79241	Late-onset multiple carboxylase deficiency
67042	Late-onset retinal degeneration
2789	Lateral meningocele syndrome
450	Lateralization defect
141136	Laterofacial microsomia
46059	Lathosterolosis
98964	Lattice corneal dystrophy type 1
98964	Lattice corneal dystrophy type I
99094	Laubry-Pezzi syndrome
2398	Launois-Bensaude adenolipomatosis
2377	Laurence-Moon syndrome
2378	Laurin-Sandrow syndrome
79086	Lawrence syndrome
79086	Lawrence-Seip syndrome
2379	Laxova-Opitz syndrome
137898	LBSL
2369	LBWC syndrome
2004	LC
99900	LCAD
650	LCAT deficiency
1486	LCCS1
137776	LCCS2
137783	LCCS3
98964	LCD1
93558	LCDD
98964	LCDI
5	LCHAD deficiency
52416	LCM
157973	L-CMD
626	LCMN
156	L-CPT1 deficiency
156	L-CPTI deficiency
65285	LDD
2364	LDH deficiency
284435	LDH-H subunit deficiency
284426	LDH-M subunit deficiency
352504	L-DOPA-unresponsive juvenile parkinsonism
2616	Le Merrer syndrome
330015	Lead intoxication
330015	Lead poisoning
3246	Learman syndrome
65	Leber congenital amaurosis
104	Leber hereditary optic neuropathy
190	Leber miliary aneurysm
104	Leber optic atrophy
99718	Leber 'plus' disease
98955	LECD
650	Lecithin-cholesterol acyltransferase deficiency
199251	Ledderhose disease
71273	Left renal vein entrapment syndrome
99111	Left superior caval vein persisting to left-sided atrium
99111	Left superior vena cava persisting to left-sided atrium

ORPHA Number	Disease name
99111	Left SVC persisting to left-sided atrium
54260	Left ventricular hypertrabeculation
54260	Left ventricular noncompaction
99095	Left ventricular-to-right atrial communication
1757	Leg duplication - mirror foot
2380	Legg-Calvé-Perthes disease
549	Legionellosis
549	Legionnaires' disease
137605	Legius syndrome
2789	Lehman syndrome
1647	Leichtman-Wood-Rohn syndrome
506	Leigh disease
255241	Leigh disease with leukodystrophy
70474	Leigh disease with myopathy
255249	Leigh disease with nephrotic syndrome
506	Leigh syndrome
70474	Leigh syndrome with cardiomyopathy
255241	Leigh syndrome with leukodystrophy
255249	Leigh syndrome with nephrotic syndrome
314	Leiner disease
71274	Leiomyomatosis peritonealis disseminate
64720	Leiomyosarcoma
213807	Leiomyosarcoma of the cervix uteri
213625	Leiomyosarcoma of the corpus uteri
507	Leishmaniasis
2044	Leisti-Hollister-Rimoin syndrome
140936	Lelis syndrome
137839	Lemierre postanginal sepsis
137839	Lemierre syndrome
2382	Lennox-Gastaut syndrome
209959	Lens-induced endophthalmitis
209959	Lens-induced iridocyclitis
209959	Lens-induced uveitis
568	Lenz microphthalmia
2658	Lenz-Majewski hyperostotic dwarfism
500	LEOPARD syndrome
330032	Lepore - beta-thalassemia
508	Leprechaunism
548	Leprosy
252031	Leptomeningeal melanomatosis
268838	Leptomyelolipoma
509	Leptospirosis
2900	Leri pleonosteosis
240	Léri-Weill dyschondrosteosis
240	Léri-Weill syndrome
510	Lesch-Nyhan syndrome
158687	Lethal acantholytic epidermolysis bullosa
314718	Lethal arteriopathy syndrome due to FBLN4 deficiency
53696	Lethal arthrogryposis - anterior horn cell disease
1187	Lethal ataxia with deafness and optic atrophy
1842	Lethal bone dysplasia, Holmgren type
1420	Lethal chondrodysplasia, Moerman type
1421	Lethal chondrodysplasia, Seller type
1486	Lethal congenital contracture syndrome type 1
137776	Lethal congenital contracture syndrome type 2
137783	Lethal congenital contracture syndrome type 3
330050	Lethal encephalopathy due to mitochondrial and peroxisomal fission defect
1972	Lethal faciocardiomelic dysplasia
1046	Lethal hemolytic anemia - genital anomalies
35064	Lethal idiopathic viral infection
254857	Lethal infantile mitochondrial disease
254857	Lethal infantile mitochondrial myopathy
2347	Lethal Kniest-like dysplasia
2371	Lethal Larsen-like syndrome
86879	Lethal midline granuloma
33108	Lethal multiple pterygium syndrome
300313	Lethal neurodegenerative disorder due to copper transport defect
293925	Lethal occipital encephalocele-skeletal dysplasia syndrome
2736	Lethal omphalocele-cleft palate syndrome
216804	Lethal osteogenesis imperfecta
1832	Lethal osteosclerotic bone dysplasia
210144	Lethal polymalformative syndrome, Boissel type
1234	Lethal popliteal pterygium syndrome
1423	Lethal recessive chondrodysplasia
1662	Lethal restrictive dermopathy
79022	Lethal variant of Simpson-Golabi-Behmel syndrome
99870	Letterer-Siwe disease
511	Leucinosis
58017	Leukemic reticuloendotheliosis
300878	Leukemic reticuloendotheliosis variant
2968	Leukocyte adhesion deficiency
99842	Leukocyte adhesion deficiency type I
99843	Leukocyte adhesion deficiency type II
99844	Leukocyte adhesion deficiency type III
99844	Leukocyte adhesion deficiency-1 variant
77295	Leukodystrophy with oligodontia
137639	Leukoencephalopathy - ataxia - hypodontia - hypomyelination
163684	Leukoencephalopathy - dystonia - motor neuropathy
83629	Leukoencephalopathy - metaphyseal chondrodysplasia
314051	Leukoencephalopathy - thalamus and brainstem anomalies - high lactate
139444	Leukoencephalopathy with bilateral anterior temporal lobe cysts
137898	Leukoencephalopathy with brain stem and spinal cord involvement - lactate elevation
135	Leukoencephalopathy with vanishing white matter
2386	Leukoencephalopathy-palmoplantar keratoderma syndrome
1816	Leukomelanoderma - intellectual deficit - hypotrichosis
2387	Leukonychia totalis
210133	Leukonychia totalis - acanthosis-nigricans-like lesions - abnormal hair
2045	Leukonychia totalis - trichilemmal cysts - ciliary dystrophy
79507	Leukotriene C4 synthase deficiency
2743	Levic-Stefanovic-Nikolic syndrome
2388	Levine-Critchley syndrome
871	Lev-Lenegre disease
95854	Levocardia
95854	Levocardia-situs inversus
352504	Levodopa-unresponsive juvenile parkinsonism
216694	Levo-transposition of the great arteries
2363	Levy-Hollister syndrome
302	Lewandowsky-Lutz syndrome
48162	Lewis-Sumner syndrome
96265	Leydig cel hypoplasia due to complete LH receptor inactivation
96265	Leydig cel hypoplasia due to complete luteinizing hormone receptor inactivation
96265	Leydig cel hypoplasia due to complete luteinizing hormone resistance
755	Leydig cell hypoplasia
96265	Leydig cell hypoplasia due to complete LH resistance
325448	Leydig cell hypoplasia due to LHB deficiency
325448	Leydig cell hypoplasia due to luteinizing hormone subunit beta deficiency
96266	Leydig cell hypoplasia due to partial LH receptor inactivation
96266	Leydig cell hypoplasia due to partial LH resistance
96266	Leydig cell hypoplasia due to partial luteinizing hormone receptor inactivation
96266	Leydig cell hypoplasia due to partial luteinizing hormone resistance
99824	LF
93599	L-glyceric aciduria
266	LGMD1A
264	LGMD1B

ORPHA Number	Disease name
265	LGMD1C
34516	LGMD1D
34517	LGMD1E
55595	LGMD1F
55596	LGMD1G
238755	LGMD1H
267	LGMD2A
268	LGMD2B
353	LGMD2C
62	LGMD2D
119	LGMD2E
219	LGMD2F
34514	LGMD2G
1878	LGMD2H
34515	LGMD2I
140922	LGMD2J
86812	LGMD2K
206549	LGMD2L
206554	LGMD2M
206559	LGMD2N
206564	LGMD2O
254361	LGMD2Q
93557	LHCDD
65285	Lhermitte-Duclos disease
104	LHON
313	LI
49804	Lichen amyloidosis
49804	Lichen amyloidosus
525	Lichen follicularis
525	Lichen planopilaris
254395	Lichen planus actinus
525	Lichen planus follicularis
254478	Lichen planus pemphigoides
254463	Lichen planus pigmentosa
254463	Lichen planus pigmentosus
254463	Lichen planus pigmentosus inversus
254395	Lichen planus subtropicus
254395	Lichen planus tropicus
254395	Lichenoid melanodermatitis
2390	Lichstenstein syndrome
526	Liddle syndrome
1275	Liebenberg syndrome
524	Li-Fraumeni syndrome
99812	LIG4 syndrome
99812	Ligase 4 syndrome
93557	Light and heavy chain deposition disease
93558	Light chain deposition disease
85443	Light-chain amyloidosis
97231	Ligneous conjunctivitis
2369	Limb body wall complex
93957	Limb dystonia
257	Limb girdle dystrophy with epidermolysis bullosa simplex
267	Limb girdle muscular dystrophy due to calpain deficiency
34515	Limb girdle muscular dystrophy due to FKRP deficiency
34514	Limb girdle muscular dystrophy due to telethonin deficiency
2492	Limb transversal defect - cardiac anomaly
974	Limb, scalp and skull defects
86812	Limb-girdle muscular dystrophy - intellectual deficit
62	Limb-girdle muscular dystrophy due to alpha-sarcoglycan deficiency
119	Limb-girdle muscular dystrophy due to beta-sarcoglycan deficiency
265	Limb-girdle muscular dystrophy due to caveolin-3 deficiency
219	Limb-girdle muscular dystrophy due to delta-sarcoglycan deficiency
268	Limb-girdle muscular dystrophy due to dysferlin deficiency
353	Limb-girdle muscular dystrophy due to gamma-sarcoglycan deficiency
264	Limb-girdle muscular dystrophy due to lamin A/C deficiency
266	Limb-girdle muscular dystrophy due to myotilin deficiency
1878	Limb-girdle muscular dystrophy due to TRIM32 deficiency
52430	Limb-girdle muscular dystrophy with Paget disease of bone
163892	Limbic encephalitis
83467	Limbic encephalitis - neuromyotonia - hyperhidrosis - polyneuropathy
276402	Limbic encephalitis with caspr2 antibodies
329341	Limbic encephalitis with dipeptidyl-peptidase 6 antibodies
329341	Limbic encephalitis with DPP6 antibodies
163908	Limbic encephalitis with leucine-rich glioma-inactivated 1 antibodies
163908	Limbic encephalitis with LGI1 antibodies
163914	Limbic encephalitis with nCMAgs antibodies
217253	Limbic encephalitis with NMDA receptor antibodies
217253	Limbic encephalitis with N-methyl-D-aspartate receptor antibodies
163914	Limbic encephalitis with novel Cell Membrane Antigens antibodies
69085	Limb-mammary syndrome
254857	LIMD
366	Limit dextrinosis
220402	Limited cutaneous systemic scleroderma
220402	Limited cutaneous systemic sclerosis
220407	Limited systemic sclerosis
168491	LINCL
892	Lindau disease
3077	Lindsay-Burn syndrome
79150	Linear and whorled nevoid hypermelanosis
140933	Linear atrophoderma of Moulin
228236	Linear focal dermal elastosis
2611	Linear hamartoma syndrome
46488	Linear IgA dermatosis
79466	Linear inflammatory verrucous epidermal nevus
254379	Linear lichen planus
254379	Linear LP
2612	Linear nevus sebaceus syndrome
2611	Linear verrucous nevus syndrome
36273	Linitis plastica of the stomach
77243	Lipedema
165	Lipidosis with triglyceride storage disease
255182	Lipoamide dehydrogenase deficiency
528	Lipoatrophic diabetes
156156	Lipoatrophy with diabetes, leukomelanodermic papules, liver steatosis, and hypertrophic cardiomyopathy
247762	Lipoblastoma
50811	Lipodystrophy - intellectual deficit - deafness
3163	Lipodystrophy - Rieger anomaly - diabetes
1979	Lipodystrophy due to peptidic growth factors deficiency
293838	Lipoic acid synthase deficiency
139436	Lipoid dermatoarthritis
530	Lipoid proteinosis
36397	Lipomatosis dolorosa
238593	Lipomatous mesenteritis
812	Lipomucopolysaccharidosis
268835	Lipomyelomeningocele
329481	Lipoprotein glomerulopathy
69078	Liposarcoma
238593	Liposclerotic mesenteritis
888	Lip-pit syndrome
98955	Lisch epithelial corneal dystrophy
2400	Lisker-Garcia-Ramos syndrome
101003	Lison syndrome
101356	Lissencephaly - demyelinating axonal neuropathy
531	Lissencephaly due to 17p13.3 deletion
95232	Lissencephaly due to LIS1 mutation
171680	Lissencephaly due to TUBA1A mutation
89844	Lissencephaly syndrome, Norman-Roberts type
2148	Lissencephaly type 1 due to doublecortin gene mutation
352682	Lissencephaly type 2 without muscular or eye involvement

ORPHA Number	Disease name
352682	Lissencephaly type 2 without muscular or ocular involvement
352704	Lissencephaly type 2B
86821	Lissencephaly type 3 - familial fetal akinesia sequence
86822	Lissencephaly type 3 - metacarpal bone dysplasia
100011	Lissencephaly with cerebellar hypoplasia type A
100012	Lissencephaly with cerebellar hypoplasia type B
100013	Lissencephaly with cerebellar hypoplasia type C
100014	Lissencephaly with cerebellar hypoplasia type D
100015	Lissencephaly with cerebellar hypoplasia type E
100016	Lissencephaly with cerebellar hypoplasia type F
533	Listeriosis
1680	Little syndrome
820	Livedo racemosa and cerebrovascular accidents
820	Livedo reticularis and cerebrovascular accidents
79095	Liver disease - retinitis pigmentosa - polyneuropathy - epilepsy
369	Liver glycogen phosphorylase deficiency
2415	LM
157973	LMNA-related congenital muscular dystrophy
33108	LMPS
69085	LMS
93924	Lobar holoprosencephaly
666	Lobstein disease
2440	Lobster-claw deformity
2407	LOC syndrome
314709	Localized AL amyloidosis
93685	Localized Castleman disease
263534	Localized deciduous skin
79400	Localized epidermolysis bullosa simplex
1823	Localized epiphyseal dysplasia
90289	Localized fibrosing scleroderma
314709	Localized immunoglobulinic amyloidosis
251393	Localized junctional epidermolysis bullosa, non-Herlitz type
86795	Localized lichen myxedematosus
90398	Localized lichen myxedematosus with mixed features of different subtypes
90399	Localized lichen myxedematosus with monoclonal gammopathy or systemic symptoms
178517	Localized pagetoid reticulosis
263534	Localized peeling skin syndrome
163927	Localized pustular psoriasis
90289	Localized scleroderma
2406	Locked-in syndrome
75566	Loeffler endocarditis
60030	Loeys-Dietz syndrome type 1
284973	Loeys-Dietz syndrome type 2
284984	Loeys-Dietz syndrome with osteoarthritis
2407	LOGIC syndrome
250831	Logopenic primary progressive aphasia
250831	Logopenic progressive aphasia
250831	Logopenic variant PPA
2404	Loiasis
5	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
99900	Long chain acyl-CoA dehydrogenase deficiency
3363	Long eyelashes - intellectual deficit
90647	Long QT interval - deafness
65283	Long QT syndrome - syndactyly
65283	Long QT syndrome type 8
180157	Longitudinal vaginal septum
52054	Longman-Tolmie syndrome
168	Loose anagen syndrome
2832	Lopes-Gorlin syndrome
2266	Lopes-Marques de Faria syndrome
1532	Lopez-Hernandez syndrome
67042	LORD
79395	Loricrin keratoderma
803	Lou-Gehrig disease
100	Louis-Bar syndrome
2621	Low birth weight - dwarfism - dysgammaglobulinemia
251633	Low grade ependymoma
69663	Low phospholipid associated cholelithiasis
534	Lowe disease
534	Lowe oculo-cerebro-renal syndrome
534	Lowe syndrome
2408	Lowe-Kohn-Cohen syndrome
2487	Lower limb deficiency - hypospadias
295051	Lower limb hypertrophy
141064	Lower lip fistula
276435	Lower motor neuron syndrome with late-adult onset
140949	Low-flow priapism
1652	Low-molecular-weight proteinuria with hypercalciuria and nephrocalcinosis
844	Lown-Ganong-Levine syndrome
1533	Lowry syndrome
2409	Lowry-MacLean syndrome
1824	Lowry-Wood syndrome
2003	Lowry-Yong syndrome
254478	LP pemphigoides
254463	LP pigmentosa
254463	LP pigmentosus
250831	LPA
71274	LPD
329481	LPG
470	LPI
309015	LPL deficiency
163927	LPP*
525	LPP*
37553	LQT7
65283	LQT8
314051	LTBL
79507	LTC4 synthase deficiency
2004	LTEC
93938	LTEC I
93939	LTEC II
93940	LTEC III
93941	LTEC IV
280205	LTEC0
93938	LTEC1
93939	LTEC2
93940	LTEC3
93941	LTEC4
216694	L-transposition of the great arteries
53351	Lubag
2575	Lubani-Al Saleh-Teebi syndrome
2410	Lubinsky syndrome
2312	Lucey-Driscoll syndrome
776	Lujan syndrome
776	Lujan-Fryns syndrome
319213	Lujo hemorrhagic fever
268388	Lumbosacral spina bifida aperta
268758	Lumbosacral spina bifida cystica
97332	Lunatomalacia
2928	Lundberg syndrome
1120	Lung agenesis - heart defect - thumb anomalies
137631	Lung fibrosis - immunodeficiency - 46,XX gonadal dysgenesis
90285	Lupus erythematosus panniculitis
90285	Lupus erythematosus profundus
90283	Lupus erythematosus tumidus
1173	Luteinizing hormone-releasing hormone deficiency with ataxia
302	Lutz-Lewandowsky epidermodysplasia verruciformis
54260	LVNC
537	Lyell syndrome
86869	LYG
91546	Lyme borreliosis
91546	Lyme disease

*Caution: one same acronym may correspond to different diseases in medical terms. Please refer to the full name of the disease to get the correct Orpha code.

ORPHA Number	Disease name
538	Lymphangioleiomyomatosis
2415	Lymphangioma
2035	Lymphatic filariasis
2415	Lymphatic malformation
86915	Lymphedema - atrial septal defects - facial changes
86914	Lymphedema - cerebral arteriovenous anomaly
86917	Lymphedema - cleft palate
33001	Lymphedema - distichiasis
1563	Lymphedema - hypoparathyroidism
2136	Lymphedema - lymphangiectasia - intellectual deficit
662	Lymphedema with yellow nails
158793	Lymphoadenopathic mastocytosis with eosinophilia
86870	Lymphoblastoid variant of NK-cell lymphoma
98846	Lymphocyte-depleted classical Hodgkin lymphoma
98845	Lymphocyte-rich classical Hodgkin lymphoma
65279	Lymphocytic colitis
79128	Lymphocytic interstitial pneumonia
289682	Lymphoepithelial-like carcinoma
86886	Lymphogranulomatosis X
314970	Lymphoid HES
314970	Lymphoid hypereosinophilic syndrome
79128	Lymphoid interstitial pneumonia
86869	Lymphomatoid granulomatosis
98842	Lymphomatoid papulosis
329998	Lymphomatous meningitis
33226	Lymphoplasmacytic immunocytoma
67038	Lymphoplasmacytic leukemia
33226	Lymphoplasmacytic lymphoma
280302	Lymphoplasmacytic sclerosing pancreatitis
67038	Lymphoplasmacytoid immunocytoma
144	Lynch syndrome
1123	Lynch-Lee-Murday syndrome
3196	Lyngstadaas syndrome
2203	Lysine alpha-ketoglutarate reductase deficiency
470	Lysinuric protein intolerance
275761	Lysosomal acid lipase deficiency
61	Lysosomal alpha-D-mannosidase deficiency
309288	Lysosomal alpha-D-mannosidase deficiency, adult form
309282	Lysosomal alpha-D-mannosidase deficiency, infantile form
34587	Lysosomal glycogen storage disease with normal acid maltase activity
79284	Lysosomal membrane cobalamin transporter deficiency
93561	Lysozyme amyloidosis
90020	Lytico-Bodig disease
330041	M hemoglobinopathy
247262	Mabry syndrome
98938	MAC
2083	Mac Dermot-Winter syndrome
36412	Mac Duffie hypocomplementemic urticarial vasculitis
36412	Mac Duffie syndrome
2675	Maccario-Mena syndrome
2220	MacDermot-Patton-Williams syndrome
98757	Machado disease
98757	Machado-Joseph disease
276238	Machado-Joseph disease type 1
276241	Machado-Joseph disease type 2
276244	Machado-Joseph disease type 3
319229	Machupo hemorrhagic fever
79495	Macias Flores-Garcia Cruz-Rivera syndrome
1574	Mackay-Shek-Carr syndrome
2477	Macrencephaly
357158	Macroblepharon - ectropion - hypertelorism - macrostomia syndrome
217335	Macrocephaly - alopecia - cutis laxa - scoliosis
60040	Macrocephaly - cutis marmorata telangiectatica congenita
94061	Macrocephaly - immune deficiency - anemia
2427	Macrocephaly - short stature - paraplegia
2429	Macrocephaly - spastic paraplegia - dysmorphism
210548	Macrocephaly-autism syndrome
60040	Macrocephaly-capillary malformation syndrome
2563	Macrocephaly-obesity-mental disability-ocular abnormalities syndrome
79489	Macrocystic lymphangioma
79489	Macrocystic lymphatic malformation
295044	Macrodactyly of fingers
295241	Macrodactyly of fingers, bilateral
295239	Macrodactyly of fingers, unilateral
295047	Macrodactyly of foot
295044	Macrodactyly of hand
295047	Macrodactyly of toes
295245	Macrodactyly of toes, bilateral
295243	Macrodactyly of toes, unilateral
592	Macrophagic myofasciitis
2432	Macrosomia - microphthalmia - cleft palate
2563	Macrosomia-obesity-macrocephaly-ocular abnormalities syndrome
141276	Macrostomia
83619	Macrostomia - preauricular tags - external ophthalmoplegia
220448	Macrothrombocytopenia with mitral valve insufficiency
217335	MACS syndrome
137814	Macular amyloidosis
91494	Macular coloboma - cleft palate - hallux valgus
98969	Macular corneal dystrophy
827	Macular dystrophy with flecks
79457	Maculopapular cutaneous mastocytosis
90287	Maculopapular lupus rash
2457	MAD
26791	MAD deficiency
26791	MADD
35688	Madelung deformity
295223	Madelung deformity, bilateral
295221	Madelung deformity, unilateral
2398	Madelung disease
137867	Madras motor neuron disease
48162	MADSAM
2583	Madura foot
1942	MAE
199354	Maeda syndrome
163634	Maffucci syndrome
324972	MAGIC syndrome
77297	Majeed syndrome
70470	Major hyperlipidemia
210272	Mal de débarquement
87503	Mal de Meleda
556	Malakoplakia
673	Malaria
75376	Malattia leventinese
2234	Male hypergonadotropic hypogonadism - intellectual deficit - skeletal anomalies
137893	Male infertility associated with large-headed multiflagellar polyploid spermatozoa
352613	Male infertility due to NANOS1 mutation
217034	Male infertility with normal virilization due to maturation arrest
217034	Male infertility with normal virilization due to meiosis defect
3000	Male limited precocious puberty
755	Male pseudohermaphrodism due to LH resistance or LHB deficiency
755	Male pseudohermaphrodism due to luteinizing hormone resistance or luteinizing hormone beta subunit deficiency
752	Male pseudohermaphroditism due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
753	Male pseudohermaphroditism due to 5-alpha-reductase 2 deficiency
1646	Male sterility due to chromosome Y deletion

ORPHA Number	Disease name
289385	Malignancy diagnosed during pregnancy
98839	Malignant angioendotheliomatosis
679	Malignant atrophic papulosis
99912	Malignant dysgerminomatous germ cell tumor
2023	Malignant fibrous histiocytoma
213837	Malignant germ cell tumor of the cervix uteri
213751	Malignant germ cell tumor of the corpus uteri
423	Malignant hyperpyrexia
423	Malignant hyperthermia
2215	Malignant hyperthermia - arthrogryposis - torticollis
168999	Malignant melanoma of the mucosa
293181	Malignant migrating partial epilepsy of infancy
293181	Malignant migrating partial seizures of infancy
213512	Malignant mixed epithelial mesenchymal tumor of the ovary
213610	Malignant mixed müllerian tumor of the corpus uteri
213787	Malignant müllerian mixed tumor of the cervix uteri
3148	Malignant neurilemmoma
3148	Malignant neurofibroma
3286	Malignant paroxysmal ventricular tachycardia
252128	Malignant perineurioma
3148	Malignant peripheral nerve sheath tumor
252212	Malignant peripheral nerve sheath tumor with rhabdomyosarcomatous differenciation
213812	Malignant peripheral neuroectodermal tumor of the cervix uteri
213630	Malignant peripheral neuroectodermal tumor of the corpus uteri
168811	Malignant peritoneal mesothelioma
3148	Malignant schwannoma
99868	Malignant thymoma
252212	Malignant triton tumor
180242	Malignant tumor of fallopian tube
943	Malonic aciduria
943	Malonyl-CoA decarboxylase deficiency
2229	Malouf syndrome
99090	Malposition of the coronary ostium
52417	MALT lymphoma
103907	Maltase-glucoamylase deficiency
52417	MALToma
50920	Mammary polyadenomatosis
238744	Mammary-digital-nail syndrome
337	Man of stone
244310	Man5GlcNAc2-PP-Dol flippase deficiency
141174	Mandibular arteriovenous malformation
246	Mandibulofacial dysostosis with postaxial limb anomalies
2457	Mandibuloacral dysplasia
90153	Mandibuloacral dysplasia with type A lipodystrophy
90154	Mandibuloacral dysplasia with type B lipodystrophy
2458	Mandibulofacial dysostosis - deafness - postaxial polydactyly
99143	Mandibulofacial dysostosis - lymphedema syndrome
357158	Mandibulofacial dysostosis - macroblepharon - macrostomia
245	Mandibulofacial dysostosis with preaxial limb anomalies
861	Mandibulofacial dysostosis without limb anomalies
79113	Mandibulofacial dysostosis, Guion-Almeida type
1131	Mandibulofacial dysostosis, Toriello type
79113	Mandibulofacial dysostosis-microcephaly syndrome
91412	Mandibulo-palpebral synkinesis - ptosis
306682	Manganese intoxication
306682	Manganese poisoning
2717	Manitoba oculotrichoanal syndrome
79327	Mannosyltransferase 1 deficiency
79326	Mannosyltransferase 2 deficiency
79321	Mannosyltransferase 6 deficiency
79328	Mannosyltransferase 7-9 deficiency
79324	Mannosyltransferase 8 deficiency
2459	Mansonellosis
52416	Mantle cell lymphoma
52416	Mantle zone lymphoma
511	Maple syrup urine disease
2785	Marble brain disease
228157	Marburg acute multiple sclerosis
99826	Marburg hemorrhagic fever
99826	Marburg virus disease
221074	Marchiafava-Bignami disease
447	Marchiafava-Micheli disease
91412	Marcus-Gunn phenomenon
91412	Marcus-Gunn syndrome
2461	Marden-Walker syndrome
2460	Marden-Walker-like syndrome
1120	Mardini--Nyhan syndrome
558	Marfan syndrome
284963	Marfan syndrome type 1
284973	Marfan syndrome type 2
2462	Marfanoid craniosynostosis syndrome
314041	Marfanoid habitus - inguinal hernia - advanced bone age
2463	Marfanoid habitus - intellectual deficit, autosomal recessive
2464	Marfanoid syndrome, De Silva type
444	Marie Unna congenital hypotrichosis
444	Marie Unna hereditary hypotrichosis
101104	Marin-Amat syndrome
559	Marinesco-Sjögren syndrome
2717	Marles syndrome
2717	Marles-Greenberg-Persaud syndrome
583	Maroteaux-Lamy disease
2767	Maroteaux-Le Merrer-Bensahel syndrome
950	Maroteaux-Malamut syndrome
1423	Maroteaux-Stanescu-Cousin syndrome
1040	Maroteaux-Verloes-Stanescu syndrome
101337	Marseilles fever
560	Marshall syndrome
42642	Marshall syndrome with periodic fever
561	Marshall-Smith syndrome
908	Martin-Bell syndrome
85321	Martin-Probst syndrome
1387	Martsolf syndrome
2466	MASA syndrome
73	Massive osteolysis
2467	Mast cell disease
98851	Mast cell leukemia
66661	Mast cell sarcoma
101001	Mast syndrome
2135	Mastocytosis - short stature - hearing loss
168598	MAT deficiency
168598	MAT I/III deficiency
254534	Maternal 14q32.2 hypermethylation syndrome
254528	Maternal 14q32.2 microdeletion syndrome
275944	Maternal anti-Kell allo-immunization
254528	Maternal del(14)(q32.2)
2209	Maternal hyperphenylalaninemia
2216	Maternal hyperthermia induced birth defects
254528	Maternal monosomy 14q32.2
2209	Maternal phenylketonuria
2209	Maternal PKU
329942	Maternal riboflavin deficiency
251009	Maternal uniparental disomy of chromosome 1
96179	Maternal uniparental disomy of chromosome 2
96180	Maternal uniparental disomy of chromosome 4
96181	Maternal uniparental disomy of chromosome 6
96183	Maternal uniparental disomy of chromosome 9
97678	Maternal uniparental disomy of chromosome 13
96184	Maternal uniparental disomy of chromosome 14

ORPHA Number	Disease name
96185	Maternal uniparental disomy of chromosome 16
96186	Maternal uniparental disomy of chromosome 20
96187	Maternal uniparental disomy of chromosome 21
96188	Maternal uniparental disomy of chromosome 22
261519	Maternal uniparental disomy of chromosome X
1349	Maternally-inherited cardiomyopathy and deafness
1349	Maternally-inherited cardiomyopathy and hearing loss
663	Maternally-inherited chronic progressive external ophthalmoplegia
663	Maternally-inherited CPEO
225	Maternally-inherited diabetes and deafness
255210	Maternally-inherited infantile subacute necrotizing encephalopathy
255210	Maternally-inherited Leigh disease
255210	Maternally-inherited Leigh syndrome
254851	Maternally-inherited mitochondrial dystonia
255225	Maternally-inherited mitochondrial hypertrophic cardiomyopathy
663	Maternally-inherited progressive external ophthalmoplegia
320360	Maternally-inherited spastic paraplegia
320360	Maternally-inherited SPG
2015	Mathieu-De Broca-Bony syndrome
2470	Matthew-Wood syndrome
552	Maturity-onset diabetes of the young
293603	Maumenee corneal dystrophy
141171	Maxillary arteriovenous malformation
1248	Maxillo-nasal dysostosis
1248	Maxillo-nasal dysplasia
3109	Mayer-Rokitansky-Küster-Hauser syndrome
247775	Mayer-Rokitansky-Küster-Hauser syndrome type 1
2578	Mayer-Rokitansky-Küster-Hauser syndrome type 2
57782	Mazabraud syndrome
91138	MC
93554	MC type II
93555	MC type III
71529	MC4R deficiency
254519	MCA due to 14q32.2 maternally expressed gene defect
42	MCAD deficiency
42	MCADD
300496	MCAHS type 2
60040	MCAP
368	McArdle disease
79140	MCC
6	MCC deficiency
85195	McCabe's disease
6	MCCD
562	McCune-Albright syndrome
148	MCD*
93686	MCD*
98969	MCD*
2471	McDonough syndrome
75327	MCDR1
319640	MCDR2
36412	McDuffie hypocomplementemic urticarial vasculitis
36412	McDuffie syndrome
308425	MCEE deficiency
158668	McGrath syndrome
2473	McKusick-Kaufman syndrome
52416	MCL
2474	McLain-Dekaban syndrome
59306	McLeod neuroacanthocytosis syndrome
60040	MCM
60040	MCMTC
77298	MCOPS3
85275	MCOPS4
178364	MCOPS5
139471	MCOPS6
2556	MCOPS7
3434	MCOPS8
2470	MCOPS9
77299	MCOPS10
2512	MCPH
2001	McPherson-Clemens syndrome
2999	McPherson-Hall syndrome
228418	MCSZ
59	MCT8 deficiency
809	MCTD
565	MD
273	MD1
258	MDC1A
210272	MdD
280333	MDDGC7
210272	MdDS
1836	MDK
238744	MDN syndrome
3097	Meacham syndrome
3097	Meacham-Winn-Culler syndrome
588	MEB syndrome
98954	MECD
564	Meckel syndrome
3032	Meckel syndrome type 7
564	Meckel-Gruber syndrome
3032	Meckel-like syndrome type 1
70588	Meconium aspiration syndrome
314376	Meconium ileus due to guanylate cyclase 2C deficiency
93308	MED1
93307	MED4
93311	MED5
3453	MEDAC syndrome
98838	Med-DLBCL
2476	Medeira-Dennis-Donnai syndrome
57196	Medial condensing osteitis of the clavicle
2006	Median cleft lip/mandibule
2006	Median cleft lower facial stage
1993	Median cleft of the upper lip - corpus callosum lipoma - cutaneous polyps
141239	Median cleft of the upper lip and maxilla
250	Median cleft syndrome
2699	Median nodule of the upper lip
98838	Mediastinal diffuse large-cell lymphoma with sclerosis
63999	Mediastinal fibrosis
231	Medina worm disease
231	Medinensis
231214	Mediterranean anemia
83313	Mediterranean boutonneuse fever
103915	Mediterranean lymphoma
101022	Mediterranean macrothrombocytopenia
101338	Mediterranean spotted fever
42	Medium chain acyl-CoA dehydrogenase deficiency
171851	MEDNIK syndrome
3050	Medrano-Roldan syndrome
29073	Medullary plasmacytoma
1309	Medullary sponge kidney
1332	Medullary thyroid carcinoma
616	Medulloblastoma
251858	Medulloblastoma with extensive nodularity
251883	Medulloepithelioma
98954	Meesmann corneal dystrophy
97252	Mega-cisterna magna
66629	Megacolon - microcephaly
280671	Megaconial congénital muscular dystrophy
238637	Megacystis-megaureter syndrome
2241	Megacystis-microcolon-intestinal hypoperistalsis syndrome
2241	Megacystis-microcolon-intestinal hypoperistalsis-hydronephrosis syndrome
2604	Megaduodenum and/or megacystis
2478	Megalencephalic leukodystrophy
2478	Megalencephalic leukoencephalopathy with subcortical cysts
2477	Megalencephaly
60040	Megalencephaly - cutis marmorata telangiectatica congenita

*Caution: one same acronym may correspond to different diseases in medical terms. Please refer to the full name of the disease to get the correct Orpha code.

ORPHA Number	Disease name
2478	Megalencephaly - cystic leukodystrophy
83473	Megalencephaly - polymicrogyria - post-axial polydactyly - hydrocephalus
60040	Megalencephaly-capillary malformation syndrome
60040	Megalencephaly-capillary malformation-polymicrogyria syndrome
238763	Megalocornea - spherophakia - secondary glaucoma
2479	Megalocornea-intellectual deficit syndrome
50815	Mégarbané-Loiselet syndrome
238637	Megaureter-megacystis syndrome
352328	MEGDEL syndrome
3038	Mehes syndrome
85282	MEHMO syndrome
2196	Meier-Blumberg-Imahorn syndrome
2554	Meier-Gorlin syndrome
90186	Meige disease
93964	Meige dystonia
90186	Meige lymphedema
93964	Meige syndrome
90185	Meige-like disease
314451	Meigs syndrome
252206	Melanoma and neural system tumor syndrome
97338	Melanoma of soft part
252206	Melanoma-astrocytoma syndrome
51013	Melanoma-pancreatic cancer syndrome
79146	Melanosis diffusa congenita
79146	Melanosis universalis hereditaria
550	MELAS syndrome
87503	Meleda disease
2482	Melhem-Fahl syndrome
31202	Melioidosis
2483	Melkersson-Rosenthal syndrome
2484	Melnick-Needles syndrome
2485	Melorheostosis
1879	Melorheostosis with osteopoikilosis
93571	Membranoproliferative glomerulonephritis type 2
69063	Membranous congenital glomerulonephritis due to anti-maternal neutral endopeptidase alloimmunisation
652	MEN 1
653	MEN2
247698	MEN2A
247709	MEN2B
276152	MEN4
319552	Mendelian susceptibility to interleukin 12 receptor beta 1 deficiency
99898	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR1 deficiency
319547	Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR2 deficiency
319558	Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency
319552	Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency
99898	Mendelian susceptibility to mycobacterial diseases due to complete interferon gamma receptor 1 deficiency
319547	Mendelian susceptibility to mycobacterial diseases due to complete interferon gamma receptor 2 deficiency
319558	Mendelian susceptibility to mycobacterial diseases due to complete interleukin 12B deficiency
319563	Mendelian susceptibility to mycobacterial diseases due to complete ISG15 deficiency
319600	Mendelian susceptibility to mycobacterial diseases due to partial interferon regulatory factor 8 deficiency
319600	Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency
319595	Mendelian susceptibility to mycobacterial diseases due to partial signal transducer and activator of transcription 1 deficiency
319595	Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency
2494	Menetrier disease
3216	Mengel-Konigsmark syndrome
45360	Meniere disease
252046	Meningeal melanocytoma
2495	Meningioma
33475	Meningococcal meningitis
565	Menkes disease
565	Menkes syndrome
75858	Mental retardation - truncal obesity - retinal dystrophy - micropenis
330021	Mercurialism
330021	Mercury intoxication
330021	Mercury poisoning
79140	Merkel cell carcinoma
258	Merosin-negative congenital muscular dystrophy
551	MERRF syndrome
54370	Mesangiocapillary glomerulonephritis
386	Mesenchymal hamartoma of liver
238593	Mesenteric lipogranuloma
238593	Mesenteric panniculitis
99701	Mesial temporal lobe epilepsy with hippocampal sclerosis
295004	Mesoaxial polydactyly of fingers
295173	Mesoaxial polydactyly of fingers, bilateral
295171	Mesoaxial polydactyly of fingers, unilateral
295010	Mesoaxial polydactyly of toes
295185	Mesoaxial polydactyly of toes, bilateral
295183	Mesoaxial polydactyly of toes, unilateral
157801	Mesoaxial synostotic syndactyly with phalangeal reduction
95443	Mesocardia
289	Mesodermic dysplasia
2496	Mesomelia-synostoses syndrome
2496	Mesomelia-synostoses syndrome, Verloes-David-Pfeiffer type
2631	Mesomelic dwarfism - cleft palate - camptodactyly
2632	Mesomelic dwarfism, Langer type
2633	Mesomelic dwarfism, Nievergelt type
2634	Mesomelic dwarfism, Reinhardt-Pfeiffer type
97360	Mesomelic dwarfism-small genitalia syndrome
85170	Mesomelic dysplasia with absent fibulas and triangular tibias
2496	Mesomelic dysplasia with acral synostoses, Verloes-David-Pfeiffer type
1836	Mesomelic dysplasia, Kantaputra type
85170	Mesomelic dysplasia, Savarirayan type
1836	Mesomelic dysplasia, Thai type
50251	Mesothelioma
95432	Mesulam syndrome
171690	Metabolic myopathy due to lactate transporter defect
2499	Metachondromatosis
512	Metachromatic leukodystrophy
309271	Metachromatic leukodystrophy, adult form
309263	Metachromatic leukodystrophy, juvenile form
309256	Metachromatic leukodystrophy, late infantile form
1240	Metaphyseal acroscyphodysplasia
1040	Metaphyseal anadysplasia
166035	Metaphyseal chondrodysplasia - retinitis pigmentosa
33067	Metaphyseal chondrodysplasia, Jansen type
166038	Metaphyseal chondrodysplasia, Kaitila type
175	Metaphyseal chondrodysplasia, McKusick type
174	Metaphyseal chondrodysplasia, Schmid type
2501	Metaphyseal chondrodysplasia, Spahr type
99646	Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria
2502	Metaphyseal dysostosis - intellectual deficit - conductive deafness
2504	Metaphyseal dysplasia - maxillary hypoplasia - brachydacty
85188	Metaphyseal dysplasia, Braun-Tinschert type
3005	Metaphyseal dysplasia, Pyle type

ORPHA Number	Disease name
213531	Metaplastic carcinoma of the breast
95504	Metastatic pituitary hormone deficiency
99866	Metastatic spermatocytic seminoma
2635	Metatropic dwarfism
2635	Metatropic dysplasia type 1
88639	Methacrylic aciduria
31825	Methanol poisoning
1923	Methimazole embryofetopathy
168598	Methionine adenosyltransferase deficiency
90070	Methotrexate intoxication
90070	Methotrexate poisoning
86904	Methotrexate-associated lymphoproliferative disorders
1917	Methyl mercury antenatal infection
622	Methylcobalamin deficiency
308380	Methylcobalamin deficiency type cblDv1
2169	Methylcobalamin deficiency type cblE
2170	Methylcobalamin deficiency type cblG
395	Methylene tetrahydrofolate reductase deficiency
308425	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency
308425	Methylmalonic acidemia due to methylmalonyl-CoA racemase deficiency
26	Methylmalonic acidemia with homocystinuria
79282	Methylmalonic acidemia with homocystinuria, type cblC
79283	Methylmalonic acidemia with homocystinuria, type cblD
79284	Methylmalonic acidemia with homocystinuria, type cblF
293355	Methylmalonic acidemia without homocystinuria
280183	Methylmalonic acidemia, TCb1R type
280183	Methylmalonic acidemia, TCbIR type
308425	Methylmalonic aciduria due to methylmalonyl-CoA epimerase deficiency
308425	Methylmalonic aciduria due to methylmalonyl-CoA racemase deficiency
280183	Methylmalonic aciduria due to transcobalamin receptor defect
26	Methylmalonic aciduria with homocystinuria
79282	Methylmalonic aciduria with homocystinuria, type cblC
79283	Methylmalonic aciduria with homocystinuria, type cblD
79284	Methylmalonic aciduria with homocystinuria, type cblF
293355	Methylmalonic aciduria without homocystinuria
29	Mevalonic aciduria
2710	Meyer-Schwickerath syndrome
79113	MFDM syndrome
293190	MFH
558	MFS
284963	MFS1
284973	MFS2
67046	MGA type 1
111	MGA2
67047	MGA3
67048	MGA4
66634	MGA5
79329	MGAT2-CDG syndrome
35123	MHBD deficiency
99826	MHF
386	MHL
79651	mHPA
294016	MIC-CAP syndrome
294016	MIC-CM syndrome
2505	Michelin tire baby syndrome
2507	Mickleson syndrome
163937	MICPCH
2508	Micrencephaly - corpus callosum agenesis - abnormal genitalia
2510	Micro syndrome
2511	Microbrachycephaly - ptosis - cleft lip
2512	Microcephalia vera
85172	Microcephalic osteodysplastic dysplasia, Saul-Wilson type
2637	Microcephalic osteodysplastic primordial dwarfism type 2
2636	Microcephalic osteodysplastic primordial dwarfism types 1 and 3
2636	Microcephalic osteodysplastic primordial dwarfism, Taybi-Linder type
329228	Microcephalic primordial dwarfism due to ZNF335 deficiency
319671	Microcephalic primordial dwarfism, Alazami type
319675	Microcephalic primordial dwarfism, Dauber type
2643	Microcephalic primordial dwarfism, Toriello type
329228	Microcephalic primordial dwarfism, Walsh type
2513	Microcephaly - albinism - digital anomalies
3433	Microcephaly - brachydactyly - kyphoscoliosis
2523	Microcephaly - brain defect - spasticity - hypernatremia
2516	Microcephaly - cardiac defect - lung malsegmentation
2515	Microcephaly - cardiomyopathy
2522	Microcephaly - cervical spine fusion anomalies
2521	Microcephaly - cleft palate
99142	Microcephaly - cutis verticis gyrata - lymphedema
2533	Microcephaly - deafness - intellectual deficit
137653	Microcephaly - digital anomalies - intellectual deficit
217026	Microcephaly - facio-cardio-skeletal syndrome, Hadziselimovic type
2172	Microcephaly - glomerulonephritis - marfanoid habitus
2065	Microcephaly - hiatus hernia - nephrotic syndrome
2558	Microcephaly - hypergonadotropic hypogonadism - short stature
3132	Microcephaly - hypogammaglobulinemia - abnormal immunity
647	Microcephaly - immunodeficiency - lymphoreticuloma
137658	Microcephaly - intellectual deficit - phalangeal and neurological anomalies
1305	Microcephaly - intellectual deficit - tracheoesophageal fistula
1229	Microcephaly - intracranial calcification - intellectual deficit
2526	Microcephaly - lymphedema - chorioretinopathy
3434	Microcephaly - microphthalmia - ectrodactyly of lower limbs - prognathism
1305	Microcephaly - oculo-digito-esophageal-duodenal syndrome
171703	Microcephaly - polymicrogyria - corpus callosum agenesis
228418	Microcephaly - seizures - developmental delay
2519	Microcephaly - seizures - intellectual deficit - heart disease
240760	Microcephaly and chromosomal instability without immunodeficiency
2512	Microcephaly vera
294016	Microcephaly-capillary malformation syndrome
329332	Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome
329332	Microcephaly-cerebellar hypoplasia-congenital heart conduction defect syndrome
294016	Microcephaly-cutaneous capillary malformation syndrome
2528	Microcephaly-microcornea syndrome, Seemanova type
2670	Microcoria - congenital nephrosis
2535	Microcornea - corectopia - macular hypoplasia
2536	Microcornea - glaucoma - absent frontal sinuses
231736	Microcornea - posterior megalolenticonus - persistent fetal vasculature - coloboma
263347	Microcornea - rod-cone dystrophy - cataract - posterior staphyloma
98956	Microcystic corneal dystrophy
79490	Microcystic infiltrating lymphatic malformation

ORPHA Number	Disease name
79490	Microcystic lymphangioma
79490	Microcystic lymphatic malformation
83642	Microcytic anemia with liver iron overload
567	Microdeletion 22q11.2
77301	Microdeletion 9q22.3
90024	Microdontia - type I microtia - deafness
101081	Microduplication 17p12
217377	Microduplication Xp11.22-p11.23 syndrome
280200	Microform holoprosencephaly
280200	Microform HPE
2538	Microgastria - limb reduction defect
1388	Micrognathia digital syndrome
1083	Microlissencephaly
50810	Microlissencephaly - micromelia
89844	Microlissencephaly type A
101052	Microlissencephaly type B
2641	Micromelic dwarfism, Fryns type
93329	Micromelic dysplasia - dislocation of radius
85275	Microphthalmia - ankyloblepharon - intellectual deficit
98938	Microphthalmia - anophthalmia - coloboma
77299	Microphthalmia - brain atrophy
2543	Microphthalmia - cataract
2556	Microphthalmia - dermal aplasia - sclerocornea
2547	Microphthalmia - microtia - fetal akinesia
2705	Microphthalmia - optic nerve aplasia
251279	Microphthalmia - retinitis pigmentosa - foveoschisis - optic disc drusen
139471	Microphthalmia with brain and digit anomalies
98938	Microphthalmia with colobomatous cyst
1106	Microphthalmia with limb anomalies
2556	Microphthalmia with linear skin defects syndrome
568	Microphthalmia, Lenz type
727	Micropolyangiitis
58220	Microscopic colitis
727	Microscopic polyangiitis
727	Microscopic polyarteritis
2551	Microspherophakia - metaphyseal dysplasia
2552	Microsporidiosis
83463	Microtia
139450	Microtia - eye coloboma - imperforation of the nasolacrimal duct
2306	Microtia-aortic arch syndrome
289522	Microtriplication 11q24.1
2290	Microvillous inclusion disease
166430	Micturation-induced seizures

ORPHA Number	Disease name
1456	Mid-aortic dysplastic syndrome
1456	Midaortic syndrome
1456	Mid-aortic syndrome
2556	MIDAS syndrome
225	MIDD
228299	Mid-dermal elastolysis
1456	Middle aortic syndrome
100084	Middle ear endocrine tumor
93926	Middle interhemispheric fusion variant
93926	Middle interhemispheric variant of holoprosencephaly
2323	Middle-East syndrome
141288	Midline cervical cleft
95443	Midline heart
93926	Midline interhemispheric variant of holoprosencephaly
2557	Mietens syndrome
2867	Mievis-Verellen Dumoulin syndrome
293181	Migrating partial epilepsy of infancy
293181	Migrating partial seizures of infancy
504	Migratory myiasis
93926	MIH
93926	MIH type HPE
93926	MIHF
93926	MIHV
2558	Mikati-Najjar-Sahli syndrome
79078	Mikulicz disease
314918	Mild Canavan disease
169799	Mild factor IX deficiency
169808	Mild factor VIII deficiency
169808	Mild hemophilia A
169799	Mild hemophilia B
79651	Mild HPA
79651	Mild hyperphenylalaninemia
171439	Mild nemaline myopathy
216796	Mild osteogenesis imperfecta
247815	Mild peroxismal disorder due to PEX10 deficiency
79253	Mild phenylketonuria
79253	Mild PKU
93279	Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis
246	Miller syndrome
531	Miller-Dieker syndrome
98919	Miller-Fisher syndrome
94091	Mills syndrome
79452	Milroy disease
79450	Milroy-like disease
255210	MILS
1917	Minamata disease
757	Mineralocorticoid resistant hyperkalemia

ORPHA Number	Disease name
352734	Minimal pigment oculocutaneous albinism type 1
98832	Minimally differentiated acute myeloblastic leukemia
822	Minkowski-Chauffard disease
1918	Minoxidil antenatal infection
566	Miosis, congenital
94125	MIRAS
295010	Mirror foot
295185	Mirror foot, bilateral
295183	Mirror foot, unilateral
295004	Mirror hand
295173	Mirror hand, bilateral
295171	Mirror hand, unilateral
2378	Mirror hands and feets - nasal defects
3004	Mirror polydactyly - vertebral segmentation - limbs defects
293822	MITF-related melanoma and renal cell carcinoma predisposition syndrome
313850	Mitochondrial aconitase deficiency
353217	Mitochondrial aspartate-glutamate carrier 1 deficiency
225	Mitochondrial diabetes
352470	Mitochondrial DNA deletion syndrome with limb-girdle weakness
352470	Mitochondrial DNA deletion syndrome with progressive myopathy
35698	Mitochondrial DNA depletion syndrome
1933	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria
255235	Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy
254871	Mitochondrial DNA depletion syndrome, hepatocerebral form
279934	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency
254875	Mitochondrial DNA depletion syndrome, myopathic form
352447	Mitochondrial DNA maintenance syndrome due to MGME1 deficiency
1194	Mitochondrial encephalo-cardio-myopathy due to ATP synthase deficiency
1194	Mitochondrial encephalo-cardio-myopathy due to F1Fo ATPase deficiency
1194	Mitochondrial encephalo-cardio-myopathy due to mitochondrial respiratory chain complex V deficiency
1194	Mitochondrial encephalo-cardio-myopathy due to TMEM70 deficiency
1933	Mitochondrial encephalomyopathy - aminoacidopathy
238329	Mitochondrial encephalomyopathy due to combined oxidative phosphorylation deficiency 6

ORPHA Number	Disease name
238329	Mitochondrial encephalomyopathy due to COXPD6
550	Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes
280288	Mitochondrial HSP60 chaperonopathy
314637	Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency
168609	Mitochondrial isolated neurosensory deafness with susceptibility to aminoglycoside exposure
168609	Mitochondrial isolated neurosensory hearing loss with susceptibility to aminoglycoside exposure
168609	Mitochondrial isolated sensorineural deafness with susceptibility to aminoglycoside exposure
168609	Mitochondrial isolated sensorineural hearing loss with susceptibility to aminoglycoside exposure
289560	Mitochondrial membrane protein associated neurodegeneration
2597	Mitochondrial myopathy - lactic acidosis - deafness
2597	Mitochondrial myopathy - lactic acidosis - hearing loss
2598	Mitochondrial myopathy and sideroblastic anemia
254864	Mitochondrial myopathy with reversible complex IV deficiency
254864	Mitochondrial myopathy with reversible COX deficiency
254864	Mitochondrial myopathy with reversible cytochrome C oxidase deficiency
550	Mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episodes
298	Mitochondrial neurogastrointestinal encephalomyopathy
90641	Mitochondrial nonsyndromic neurosensory deafness
168609	Mitochondrial nonsyndromic neurosensory deafness with susceptibility to aminoglycoside exposure
168609	Mitochondrial nonsyndromic neurosensory hearing loss with susceptibility to aminoglycoside exposure
90641	Mitochondrial nonsyndromic sensorineural deafness
168609	Mitochondrial nonsyndromic sensorineural deafness with susceptibility to aminoglycoside exposure
168609	Mitochondrial nonsyndromic sensorineural hearing loss with susceptibility to aminoglycoside exposure
254881	Mitochondrial spinocerebellar ataxia with epilepsy
746	Mitochondrial trifunctional protein deficiency
1205	Mitral atresia
3238	Mitral regurgitation - deafness - skeletal anomalies
99062	Mitral valve agenesis
295012	Mitten hand
90036	Mixed AIHA
98844	Mixed cellularity classical Hodgkin lymphoma
809	Mixed connective tissue disease
91138	Mixed cryoglobulinemia
93555	Mixed cryoglobulinemia type III
180234	Mixed germ cell tumor
252021	Mixed germ cell tumor of the central nervous system
86851	Mixed lineage acute leukemia
213610	Mixed müllerian cancer of the corpus uteri
251656	Mixed oligoastrocytoma
86851	Mixed phenotype acute leukemia
2785	Mixed renal tubular acidosis
2785	Mixed RTA
1879	Mixed sclerosing bone dystrophy
324364	Mixed sclerosing bone dystrophy with extra-skeletal manifestations
90036	Mixed-type autoimmune hemolytic anemia
45448	Miyoshi myopathy
2619	MJD*
98757	MJD*
565	MK
2598	MLASA
2478	MLC
512	MLD
309271	MLD, adult form
309263	MLD, juvenile form
309256	MLD, late infantile form
59306	MLS
2556	MLS syndrome
598	MmD
3434	MMEP syndrome
592	MMF
268249	MMF embryopathy
2241	MMIHS
137867	MMND
293181	MMPEI
293181	MMPSI
2479	MMR syndrome
1305	MMT
298	MNGIE
565	MNK
251656	MOA
77299	MOBA syndrome
570	Möbius syndrome
2560	Möbius syndrome - axonal neuropathy - hypogonadotropic hypogonadism
99732	MOCOD
308386	MOCOD type A
308393	MOCOD type B
308400	MOCOD type C
1305	MODED syndrome
90056	Moderate and severe traumatic brain injury
178145	Moderate multiminicore disease with hand involvement
169796	Moderately severe factor IX deficiency
169805	Moderately severe factor VIII deficiency
169805	Moderately severe hemophilia A
169796	Moderately severe hemophilia B
263335	Moderately-differentiated thymic neuroendocrine carcinoma
552	MODY syndrome
93111	MODY5
570	Moebius syndrome
1420	Moerman-Vandenberghe-Fryns syndrome
3198	Moersch-Woltman syndrome
2549	Moeschler-Clarren syndrome
2751	Mohr syndrome
52368	Mohr-Tranebjaerg syndrome
99927	Molar pregnancy
2650	Mollica-Pavone-Antener syndrome
1433	Moloney syndrome
2563	MOMO syndrome
573	Monilethrix
573	Moniliform hair syndrome
319254	Monkey disease
319254	Monkey fever
3057	Monoamine oxidase A deficiency
59	Monocarboxylate transporter 8 deficiency
91136	Monoclonal Ig light chain-associated Fanconi syndrome
91136	Monoclonal kappa Ig light chain-associated Fanconi syndrome
228423	Monocyte - B - natural killer - dendritic cell deficiency
228423	Monocytopenia and mycobacterial infection syndrome
228423	Monocytopenia with susceptibility to infections
228423	MonoMAC
65684	Monomelic amyotrophy
86870	Monomorphic NK-cell lymphoma
2565	Mononen-Karnes-Senac syndrome
2901	Mononeuritis multiplex with brachial predilection
293948	Monosomy 1p21.3
1606	Monosomy 1p36
1606	Monosomy 1pter
250989	Monosomy 1q21.1
250999	Monosomy 1q41q42
250999	Monosomy 1q41-q42
238769	Monosomy 1q44

*Caution: one same acronym may correspond to different diseases in medical terms. Please refer to the full name of the disease to get the correct Orpha code.

ORPHA Number	Disease name
36367	Monosomy 1qter
261349	Monosomy 2p15p16.1
261349	Monosomy 2p15-p16.1
163693	Monosomy 2p21
228402	Monosomy 2q23.1
1617	Monosomy 2q24
251014	Monosomy 2q31.1
251019	Monosomy 2q32
251019	Monosomy 2q32q33
251019	Monosomy 2q32-q33
251028	Monosomy 2q33.1
1001	Monosomy 2q37-qter
1620	Monosomy 3pter
1621	Monosomy 3q13
356947	Monosomy 3q26q27
356947	Monosomy 3q26-q27
65286	Monosomy 3q29
65286	Monosomy 3qter
1625	Monosomy 4q
238750	Monosomy 4q21
96145	Monosomy 4qter
281	Monosomy 5p
228384	Monosomy 5q14.3
314655	Monosomy 5q31.3
1627	Monosomy 5qter
251046	Monosomy 6p22
96125	Monosomy 6p25
171829	Monosomy 6q16
251056	Monosomy 6q25
96126	Monosomy 7pter
904	Monosomy 7q11.23
251061	Monosomy 7q31
1636	Monosomy 7qter
251066	Monosomy 8p11.2
251071	Monosomy 8p23.1
2496	Monosomy 8q13
284160	Monosomy 8q21.11
178303	Monosomy 8q22.1
502	Monosomy 8q24.1
261112	Monosomy 9p
324313	Monosomy 9p13
1642	Monosomy 9pter
77301	Monosomy 9q22.3
284169	Monosomy 10p11.21p12.31
1580	Monosomy 10pter
276413	Monosomy 10q22.3q23.3
96148	Monosomy 10qter
893	Monosomy 11p13
2308	Monosomy 11qter
313884	Monosomy 12p12.1
94063	Monosomy 12q14
289513	Monosomy 12q15q21.1
96149	Monosomy 12qter
1587	Monosomy 13q14
1590	Monosomy 13q32
96168	Monosomy 13q34
261120	Monosomy 14q11.2
261144	Monosomy 14q12
264200	Monosomy 14q22q23
264200	Monosomy 14q22-q23
261183	Monosomy 15q11.2
199318	Monosomy 15q13.3
261190	Monosomy 15q14
94065	Monosomy 15q24
1596	Monosomy 15q26
261211	Monosomy 16p11.2p12.2
261211	Monosomy 16p11.2-p12.2
261236	Monosomy 16p13.11
352629	Monosomy 16q24.1
261250	Monosomy 16q24.3
531	Monosomy 17p13.3
97685	Monosomy 17q11
261265	Monosomy 17q12
96169	Monosomy 17q21.31
261279	Monosomy 17q23.1q23.2
261279	Monosomy 17q23.1-q23.2
1597	Monosomy 17qter
1598	Monosomy 18p
1600	Monosomy 18q
254346	Monosomy 19p13.12
357001	Monosomy 19p13.13
217346	Monosomy 19q13.11
1611	Monosomy 20p
261295	Monosomy 20p12.3
313781	Monosomy 20p13
261311	Monosomy 20q13.33
96152	Monosomy 20qter
574	Monosomy 21
261323	Monosomy 21q22.11q22.12
261323	Monosomy 21q22.11-q22.12
268261	Monosomy 21q22.13q22.2
268261	Monosomy 21q22.13-q22.2
96123	Monosomy 22
567	Monosomy 22q11
48652	Monosomy 22q13
99226	Monosomy X
261476	Monosomy Xp21
93277	Monostotic fibrous dysplasia
158003	Montgomery syndrome
2569	Moore-Federman syndrome
2637	MOPD type II
2636	MOPD types I and III
52056	Morava-Mehes syndrome
77296	Morgagni-Stewart-Morel syndrome
3043	Morillo Cucci-Passarge syndrome
75858	MORM syndrome
35737	Morning glory syndrome
582	Morquio disease
309297	Morquio disease type A
309310	Morquio disease type B
754	Morris syndrome
2570	Morse-Rawnsley-Sargent syndrome
83467	Morvan syndrome
83467	Morvan's fibrillary chorea
329813	Mosaic genome-wide paternal uniparental disomy
329813	Mosaic genome-wide paternal UPD
99228	Mosaic monosomy X
96193	Mosaic paternal uniparental disomy of chromosome 11
1692	Mosaic trisomy 1
1723	Mosaic trisomy 2
100071	Mosaic trisomy 3
96059	Mosaic trisomy 4
96060	Mosaic trisomy 5
1747	Mosaic trisomy 7
96061	Mosaic trisomy 8
99776	Mosaic trisomy 9
96063	Mosaic trisomy 10
1698	Mosaic trisomy 12
1703	Mosaic trisomy 14
1706	Mosaic trisomy 15
1708	Mosaic trisomy 16
1711	Mosaic trisomy 17
1724	Mosaic trisomy 20
96068	Mosaic trisomy 22
1052	Mosaic variegated aneuploidy syndrome
54057	Moschcowitz disease
2717	MOTA syndrome
254516	Motor developmental delay due to 14q32.2 paternally expressed gene defect
3347	Mounier-Kuhn syndrome
83595	Mountain fever
83595	Mountain tick fever
2572	Mousa-Al Din-Al Nassar syndrome
324972	Mouth and genital ulcers with inflamed cartilage
2152	Mowat-Wilson syndrome
261537	Mowat-Wilson syndrome due to 2q22 microdeletion
261552	Mowat-Wilson syndrome due to a point mutation

ORPHA Number	Disease name
261537	Mowat-Wilson syndrome due to del(2) q(22)
261537	Mowat-Wilson syndrome due to monosomy 2q22
2573	Moyamoya disease
280679	Moyamoya disease - short stature - facial dysmorphism - hypergonadotropic hypogonadism
2574	Moynahan syndrome
352734	MP OCA type 1
727	MPA
289560	MPAN
59135	MPD1
79323	MPDU1-CDG syndrome
293181	MPEI
54370	MPGN
79319	MPI-CDG syndrome
79253	mPKU
3148	MPNST
252212	MPNST with rhabdomyosarcomatous differentiation
2587	MPO deficiency
231736	MPPC syndrome
83473	MPPH syndrome
581	MPS III
579	MPS1
93473	MPS1H
93476	MPS1H/S
93474	MPS1S
583	MPS6
276212	MPS6, rapidly progressing
276223	MPS6, slowly progressing
293181	MPSI
309297	MPSIVA
309310	MPSIVB
583	MPSVI
276212	MPSVI, rapidly progressing
276223	MPSVI, slowly progressing
99967	MRCLS
263347	MRCS syndrome
67045	MRGH
3109	MRKH syndrome
247775	MRKH syndrome type 1
2578	MRKH syndrome type 2
3059	MRX35
85274	MRXS7
85324	MRXS9
93952	MRXSH
2598	MSA
102	MSA
98932	MSA - urinary dysfunction
227510	MSA, cerebellar type
98933	MSA, parkinsonian type
227510	MSA-c
98933	MSA-p
1879	MSBD syndrome
254881	MSCAE
585	MSD
2619	Mseleni joint disease
1309	MSK
99898	MSMD due to complete IFNgammaR1 deficiency
319547	MSMD due to complete IFNgammaR2 deficiency
319558	MSMD due to complete IL12B deficiency
319552	MSMD due to complete IL12RB1 deficiency
99898	MSMD due to complete interferon gamma receptor 1 deficiency
319547	MSMD due to complete interferon gamma receptor 2 deficiency
319552	MSMD due to complete interleukin 12 receptor beta 1 deficiency
319558	MSMD due to complete interleukin 12B deficiency
319563	MSMD due to complete ISG15 deficiency
319600	MSMD due to partial interferon regulatory factor 8 deficiency
319600	MSMD due to partial IRF8 deficiency
319595	MSMD due to partial signal transducer and activator of transcription 1 deficiency
319595	MSMD due to partial STAT1 deficiency
157801	MSSD
65748	MSSE
511	MSUD
2505	MTBS
1332	MTC
352470	mtDNA deletion syndrome with progressive myopathy
352470	mtDNA deletion syndromesyndrome with limb-girdle weakness
35698	mtDNA depletion syndrome
1933	mtDNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria
255235	mtDNA depletion syndrome, encephalomyopathic form with renal tubulopathy
254871	mtDNA depletion syndrome, hepatocerebral form
254875	mtDNA depletion syndrome, myopathic form
352447	mtDNA maintenance syndrome due to MGME1 deficiency
395	MTHFR deficiency
252212	MTT
100024	Mu heavy-chain disease
319322	Mucinous tubular and spindle cell carcinoma
575	Muckle-Wells syndrome
2331	Mucocutaneous lymph node syndrome
2451	Mucocutaneous venous malformations
576	Mucolipidosis type 2
577	Mucolipidosis type 3
578	Mucolipidosis type 4
579	Mucopolysaccharidosis type 1
93473	Mucopolysaccharidosis type 1H
93476	Mucopolysaccharidosis type 1H/S
93474	Mucopolysaccharidosis type 1S
580	Mucopolysaccharidosis type 2
217093	Mucopolysaccharidosis type 2, attenuated form
217085	Mucopolysaccharidosis type 2, severe form
217085	Mucopolysaccharidosis type 2A
217093	Mucopolysaccharidosis type 2B
581	Mucopolysaccharidosis type 3
79269	Mucopolysaccharidosis type 3A
79270	Mucopolysaccharidosis type 3B
79271	Mucopolysaccharidosis type 3C
79272	Mucopolysaccharidosis type 3D
582	Mucopolysaccharidosis type 4
309297	Mucopolysaccharidosis type 4A
309310	Mucopolysaccharidosis type 4B
583	Mucopolysaccharidosis type 6
276212	Mucopolysaccharidosis type 6, rapidly progressing
276223	Mucopolysaccharidosis type 6, slowly progressing
584	Mucopolysaccharidosis type 7
67041	Mucopolysaccharidosis type 9
581	Mucopolysaccharidosis type III
583	Mucopolysaccharidosis type VI
276212	Mucopolysaccharidosis type VI, rapidly progressing
276223	Mucopolysaccharidosis type VI, slowly progressing
73263	Mucormycosis
52417	Mucosa-associated lymphatic tissue lymphoma
52417	Mucosa-associated lymphoid tissue lymphoma
46486	Mucosal pemphigoid
585	Mucosulfatidosis
46486	Mucosynechial pemphigoid
46486	Mucous membrane pemphigoid
586	Mucoviscidosis
53271	Muenke syndrome
100024	mu-HCD
444	MUHH
587	Muir-Torre syndrome

ORPHA Number	Disease name
2576	MULIBREY dwarfism
2576	MULIBREY nanism
73217	Mullerian aplasia
2578	Mullerian aplasia - renal aplasia - cervicothoracic somite dysplasia
1655	Mullerian derivatives - lymphangiectasia - polydactyly
2491	Mullerian duct anomalies - limb anomalies
73217	Mullerian duct failure
2774	Multicentric carpo-tarsal osteolysis with or without nephropathy
93686	Multicentric Castleman disease
93686	Multicentric giant lymph node hyperplasia
85196	Multicentric osteolysis - nodulosis - arthropathy
139436	Multicentric reticulohistiocytosis
168816	Multicystic mesothelioma
1851	Multicystic renal dysplasia
48162	Multifocal acquired demyelinating sensory and motor neuropathy
99873	Multifocal eosinophilic granuloma
641	Multifocal motor neuropathy with conduction block
2033	Multifocal muscular fibrosis - obstructed vessels
99003	Multifocal pattern dystrophy simulating fundus flavimaculatus
3286	Multifocal ventricular premature beats
319287	Multilocular clear cell adenocarcinoma
319287	Multilocular clear cell carcinoma
319287	Multilocular clear cell renal cell adenocarcinoma
319287	Multilocular clear cell renal cell carcinoma
97366	Multilocular cyst of the kidney
319287	Multilocular cystic renal cell adenocarcinoma
319287	Multilocular cystic renal cell carcinoma
168816	Multilocular peritoneal inclusion cyst
97366	Multilocular renal cyst
97366	Multiloculated renal cyst
598	Multiminicore disease
598	Multiminicore myopathy
2091	Multinodular goiter - cystic kidney - polydactyly
26791	Multiple acyl-CoA dehydrogenase deficiency
2505	Multiple benign circumferential skin creases on limbs
2678	Multiple café-au-lait spots
2678	Multiple café-au-lait syndrome
148	Multiple carboxylase deficiency
321	Multiple cartilaginous exostoses
280633	Multiple congenital anomalies - hypotonia - seizures syndrome
254519	Multiple congenital anomalies due to 14q32.2 maternally expressed gene defect
300496	Multiple congenital anomalies-hypotonia-seizures syndrome type 2
1486	Multiple contracture syndrome, Finnish type
137776	Multiple contracture syndrome, Israeli-Bedouin type
523	Multiple cutaneous and uterine leiomyomas
3453	Multiple endocrine deficiency - Addison's disease - candidiasis
3453	Multiple endocrine deficiency - Addison's disease - candidosis
652	Multiple endocrine neoplasia type 1
653	Multiple endocrine neoplasia type 2
247698	Multiple endocrine neoplasia type 2A
247709	Multiple endocrine neoplasia type 2B
247709	Multiple endocrine neoplasia type 3
276152	Multiple endocrine neoplasia type 4
251	Multiple epiphyseal dysplasia
166024	Multiple epiphyseal dysplasia - macrocephaly - distinctive facies
166011	Multiple epiphyseal dysplasia - myopia - deafness
166002	Multiple epiphyseal dysplasia due to collagen 9 anomaly
93308	Multiple epiphyseal dysplasia type 1
93307	Multiple epiphyseal dysplasia type 4
93311	Multiple epiphyseal dysplasia type 5
166016	Multiple epiphyseal dysplasia with Robin phenotype
166024	Multiple epiphyseal dysplasia, Al-Gazali type
166011	Multiple epiphyseal dysplasia, Beighton type
166016	Multiple epiphyseal dysplasia, Lowry type
93313	Multiple epiphyseal dysplasia, unclassified type
166032	Multiple epiphyseal dysplasia, with miniepiphyses
166029	Multiple epiphyseal dysplasia, with severe proximal femoral dysplasia
26791	Multiple FAD dehydrogenase deficiency
83454	Multiple glomus tumors
201	Multiple hamartoma syndrome
2300	Multiple intestinal atresia
284139	Multiple joint dislocations - short stature - craniofacial dysmorphism - congenital heart defects
493	Multiple keratoacanthoma
65748	Multiple keratoacanthoma, Ferguson-Smith type
587	Multiple keratoacanthoma, Muir-Torre type
79455	Multiple mastocytoma
29073	Multiple myeloma
2029	Multiple non-ossifying fibromatosis
321	Multiple osteochondromas
324299	Multiple paragangliomas associated with erythrocytosis
324299	Multiple paragangliomas associated with polycythemia
95494	Multiple pituitary hormone deficiencies, genetic forms
3151	Multiple sclerosis - ichthyosis - factor VIII deficiency
65748	Multiple self-healing squamous epithelioma
585	Multiple sulfatase deficiency
3237	Multiple synostoses syndrome
99156	Multiple syringomas
102	Multiple system atrophy
98932	Multiple system atrophy - urinary dysfunction
227510	Multiple system atrophy, cerebellar type
98933	Multiple system atrophy, parkinsonian type
99096	Multiple ventricular septal defects
2959	Mulvihill-Smith syndrome
2578	MURCS association
83315	Murine typhus
2028	Murray-Puretic-Drescher syndrome
99849	Muscle enolase deficiency
171445	Muscle filaminopathy
97234	Muscle phosphoglycerate mutase deficiency
588	Muscle-eye-brain disease
588	Muscle-eye-brain syndrome
2576	Muscle-liver-brain-eye nanism
2579	Muscular atrophy - ataxia - retinitis pigmentosa - diabetes mellitus
1877	Muscular dystrophy - white matter spongiosis
199340	Muscular dystrophy, Selcen type
99849	Muscular enolase deficiency
324416	Muscular hypertrophy - hepatomegaly - polyhydramnios
2349	Muscular pseudohypertrophy - hypothyroidism
3079	Mutchinick syndrome
494	Mutilating keratoderma of Vohwinkel
494	Mutilating keratoderma plus deafness
659	Mutilating palmoplantar hyperkeratosis with periorificial keratotic plaques
659	Mutilating palmoplantar keratoderma with periorificial keratotic plaques
247798	MUTYH-related AFAP
247798	MUTYH-related attenuated familial adenomatous polyposis
247798	MUTYH-related attenuated familial polyposis coli

ORPHA Number	Disease name
247798	MUTYH-related attenuated FAP
29	MVA
2582	Myalgia-eosinophilia syndrome associated with tryptophan
589	Myasthenia gravis
2583	Mycetoma
314946	Mycobacterium xenopi infection
268249	Mycophenolate mofetil embryopathy
83482	Mycoplasma encephalitis
2584	Mycosis fungoides, Alibert-Bazin type
178512	Mycosis fungoides-associated follicular mucinosis
183713	MyD88 deficiency
59298	Myelinoclastic diffuse sclerosis
135	Myelinosis centralis diffusa
2585	Myelocerebellar disorder
268813	Myelocystocele
86841	Myelodysplastic syndrome associated with isolated del(5q) chromosome abnormality
52688	Myelodysplastic syndromes
824	Myelofibrosis with myeloid metaplasia
168953	Myeloid neoplasm associated with FGFR1 rearrangement
168947	Myeloid neoplasm associated with PDGFRA rearrangement
168950	Myeloid neoplasm associated with PDGFRB rearrangement
86850	Myeloid sarcoma
91136	Myeloma-associated Fanconi syndrome
29073	Myelomatosis
2587	Myeloperoxidase deficiency
824	Myelosclerosis with myeloid metaplasia
2588	Myhre syndrome
109	Myhre-Riley-Smith syndrome
75110	Myiasis
45	Myoadenylate deaminase deficiency
1942	Myoclonic atonic epilepsy
36899	Myoclonic dystonia 11
210566	Myoclonic dystonia 15
86913	Myoclonic epilepsy in non-progressive encephalopathies
86909	Myoclonic epilepsy of infancy
1942	Myoclonic-astatic epilepsy in early childhood
2589	Myoclonus - cerebellar ataxia - deafness
3020	Myoclonus ataxia
551	Myoclonus epilepsy associated with ragged-red fibers
36899	Myoclonus-dystonia syndrome
1037	Myodysplasia
593	Myofibrillar myopathy
178464	Myofibrillar myopathy with early respiratory failure

ORPHA Number	Disease name
104077	Myopathic intestinal pseudoobstruction
2601	Myopathy - growth delay - intellectual deficit - hypospadias
1358	Myopathy - Moebius - Robin syndrome
2596	Myopathy and diabetes mellitus
88635	Myopathy due to calsequestrin and SERCA1 protein overload
97234	Myopathy due to phosphoglycerate mutase deficiency
43115	Myopathy with exercise intolerance, Swedish type
171889	Myopathy with hexagonally cross-linked tubular arrays
2598	Myopathy, lactic acidosis and sideroblastic anemia
289685	Myopericytoma
368	Myophosphorylase deficiency
178493	Myopic macular degeneration
178493	Myopic maculopathy
289380	Myosclerosis
337	Myositis ossificans progressiva
764	Myositis purulenta tropica
764	Myositis tropicans
306553	Myospherulosis
275534	Myostatin-related muscle hypertrophy
98911	Myotilin-related myofibrillar myopathy without spheroid body
3101	Myotonia - intellectual deficit - skeletal anomalies
99736	Myotonia - painful contractions
614	Myotonia congenita
99734	Myotonia fluctuans
99735	Myotonia permanens
800	Myotonic chondrodystrophy
273	Myotonic dystrophy type 1
606	Myotonic dystrophy type 2
800	Myotonic myopathy, dwarfism, chondrodystrophy, ocular and facial anomalies
596	Myotubular myopathy
79105	Myxofibrosarcoma
79105	Myxoid malignant fibrous histiocytoma
99967	Myxoid/round cell liposarcoma
1359	Myxoma - spotty pigmentation - endocrine overactivity
57782	Myxoma with fibrous dysplasia
251643	Myxopapillary ependymoma
2608	N syndrome
178303	Nablus mask-like facial syndrome
79270	N-acetyl-alpha-glucosaminidase deficiency
583	N-acetylgalactosamine 4-sulfatase deficiency
309297	N-acetylgalactosamine-6-sulfate sulfatase deficiency

ORPHA Number	Disease name
576	N-acetyl-glucosamine 1-phosphotransferase deficiency
79329	N-acetylglucosaminyltransferase 2 deficiency
137754	N-acyl-L-amino acid amidohydrolase deficiency
139373	NADH-cytochrome b5reductase deficiency type 1
139380	NADH-cytochrome b5reductase deficiency type 2
139373	NADH-diaphorase deficiency type 1
139380	NADH-diaphorase deficiency type 2
69087	Naegeli syndrome
69087	Naegeli-Franceschetti-Jadassohn syndrome
245	NAFD
3137	NAGA deficiency
79279	NAGA deficiency type 1
79280	NAGA deficiency type 2
79281	NAGA deficiency type 3
245	Nager acrofacial dysostosis
245	Nager syndrome
927	NAGS deficiency
2211	Naguib-Richieri-Costa syndrome
103908	Na-H exchange deficiency
2614	Nail-patella syndrome
2613	Nail-patella-like renal disease
158676	Nails-only DEB
853	NAIT
101	Naito-Oyanagi disease
2229	Najjar syndrome
1063	Nakagawa angioblastoma
2615	Nakajo-Nishimura syndrome
2822	Nakamura-Osame syndrome
44	NALD
383	Nance deafness
627	Nance-Horan syndrome
35612	Nanophthalmia
85196	NAO syndrome
247868	NAPS12
83465	Narcolepsy without cataplexy
2073	Narcolepsy-cataplexy
644	NARP syndrome
141103	Nasal dermoid cyst
141103	Nasal dermoid sinus cyst
141219	Nasal dorsum fistula/cyst
141118	Nasal encephalocele
141115	Nasal ganglioglioma
141112	Nasal glial heterotopia
141112	Nasal glioma
86879	Nasal T/natural killer-cell lymphoma
2662	Nasodigitoacoustic syndrome

ORPHA Number	Disease name
141083	Nasolacrimal duct cyst
2399	Nasopalpebral lipoma - coloboma - telecanthus
150	Nasopharyngeal carcinoma
141107	Nasopharyngeal teratoma
2770	Nasu-Hakola disease
1654	Natal teeth - intestinal pseudoobstruction - patent ductus
2663	Nathalie syndrome
168572	Native American myopathy
69739	Navajo brainstem syndrome
255229	Navajo neurohepatopathy
255229	Navajo neuropathy
34217	Naxos disease
377	NBCCS
289560	NBIA due to C19orf12 mutation
157850	NBIA1
216873	NBIA1, atypical form
216866	NBIA1, classic form
329284	NBIA4
289560	NBIA5
647	NBS
240760	NBSLD
240760	NBS-like disorder
95698	NCAH
217560	NCHI
216	NCL
1947	NCL, Northern epilepsy variant
2481	NCM
75327	NCMD
300337	NCRNA disease
224	NDM
158011	Necrobiotic xanthogranuloma
217560	NEHI
199244	Nelson syndrome
607	NEM
607	Nemaline myopathy
607	Nemaline rod myopathy
217563	Neonatal acute respiratory distress with surfactant metabolism deficiency
44	Neonatal adrenoleukodystrophy
137929	Neonatal brainstem dysfunction
314911	Neonatal Canavan disease
313906	Neonatal congenital pancreatic cyst
79118	Neonatal diabetes - congenital hypothyroidism - congenital glaucoma - hepatic fibrosis - polycystic kidneys
224	Neonatal diabetes mellitus
289857	Neonatal glycine encephalopathy
446	Neonatal hemochromatosis
137577	Neonatal hypoxic and ischemic brain injury
294023	Neonatal inflammatory skin and bowel disease
247598	Neonatal intrahepatic cholestasis caused by citrin deficiency
247598	Neonatal intrahepatic cholestasis due to citrin deficiency
238688	Neonatal iodine exposure
284979	Neonatal Marfan syndrome
97668	Neonatal membranous glomerulopathy with maternal NEP deficiency
284979	Neonatal MFS
79242	Neonatal multiple carboxylase deficiency
56304	Neonatal osseous dysplasia type 1
3455	Neonatal progeroid syndrome
70587	Neonatal respiratory distress syndrome
417	Neonatal severe primary hyperparathyroidism
1451	Neonatal-onset multisystem inflammatory disease
314950	Neoplastic hypereosinophilic syndrome
94058	Neovascular glaucoma
654	Nephroblastoma
2849	Nephroblastomatosis - fetal ascites - macrosomia - Wilms tumor
223	Nephrogenic diabetes insipidus
3145	Nephrogenic diabetes insipidus - intracranial calcification
137617	Nephrogenic fibrosing dermopathy
93606	Nephrogenic syndrome of inappropriate antidiuresis
137617	Nephrogenic systemic fibrosis
93622	Nephrolithiasis type 1
93623	Nephrolithiasis type 2
84081	Nephronophthisis - hepatic fibrosis - tapetoretinal degeneration - intellectual deficit
3156	Nephronophthisis with retinal dystrophy
2668	Nephropathy - deafness - hyperparathyroidism
2669	Nephrosis - deafness - urinary tract - digital malformations
2065	Nephrosis - neuronal dysmigration syndrome
300333	Nephrotic syndrome-deafness-pretibial epidermolysis bullosa syndrome
300333	Nephrotic syndrome-hearing loss-pretibial epidermolysis bullosa syndrome
2337	NEPPK
280576	Nestor-Guillermo progeria syndrome
634	Netherton syndrome
99078	Neuhauser anomaly
2479	Neuhäuser syndrome
3350	Neuhauser-Daly-Magnelli syndrome
2672	Neuhauser-Eichner-Opitz syndrome
2671	Neu-Laxova syndrome
635	Neural crest tumor
2901	Neuralgic amyotrophy
2901	Neuralgic shoulder amyotrophy
351	Neuraminidase deficiency with beta-galactosidase deficiency
268865	Neurenteric cyst
252164	Neurilemmoma
93921	Neurilemmomatosis
252164	Neurilemoma
2675	Neuroaxonal dystrophy - renal tubular acidosis
635	Neuroblastoma
2481	Neurocutaneous melanocytosis
2481	Neurocutaneous melanosis
35664	Neurocutaneous syndrome, Bicknell type
88639	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency
289560	Neurodegeneration with brain iron accumulation due to C19orf12 mutation
157850	Neurodegeneration with brain iron accumulation type 1
216873	Neurodegeneration with brain iron accumulation type 1, atypical form
216866	Neurodegeneration with brain iron accumulation type 1, classic form
329284	Neurodegeneration with brain iron accumulation type 4
289560	Neurodegeneration with brain iron accumulation type 5
217382	Neurodegenerative syndrome due to cerebral folate transport deficiency
3474	Neuroectodermal dysplasia, CHIME type
33445	Neuroectodermal melanolysosomal disease
2316	Neuroectodermal syndrome, Johnson type
3474	Neuroectodermal syndrome, Zunich type
2676	Neuroectodermal-endocrine syndrome
217560	Neuroendocrine cell hyperplasia of infancy
2677	Neuroepithelioma
2673	Neurofaciodigitorenal syndrome
157846	Neuroferritinopathy
252183	Neurofibroma
68388	Neurofibromatosis
636	Neurofibromatosis 1
137605	Neurofibromatosis 1-like syndrome
636	Neurofibromatosis type 1
97685	Neurofibromatosis type 1 microdeletion syndrome
637	Neurofibromatosis type 2
93921	Neurofibromatosis type 3
2678	Neurofibromatosis type 6
3148	Neurofibrosarcoma
970	Neurogenic acroosteolysis
1143	Neurogenic arthrogryposis multiplex congenita
100073	Neurogenic cervical rib syndrome

ORPHA Number	Disease name
100073	Neurogenic costoclavicular syndrome
178029	Neurogenic diabetes insipidus
644	Neurogenic muscle weakness - ataxia - retinitis pigmentosa
3148	Neurogenic sarcoma
85146	Neurogenic scapuloperoneal syndrome
100073	Neurogenic thoracic outlet compression syndrome
100073	Neurogenic thoracic outlet syndrome
100073	Neurogenic TOS
252164	Neurolemmoma
94093	Neuroleptic malignant syndrome
36397	Neurolipomatosis
163746	Neurologic Waardenburg-Shah syndrome
137754	Neurological conditions associated with aminoacylase 1 deficiency
206586	Neurolymphomatosis
71211	Neuromyelitis optica
216	Neuronal ceroid lipofuscinosis
1947	Neuronal ceroid lipofuscinosis, Northern epilepsy variant
99811	Neuronal intestinal pseudoobstruction
2289	Neuronal intranuclear inclusion disease
644	Neuropathy - ataxia - retinitis pigmentosa
139512	Neuropathy with hearing impairment
3228	Neurosensory deafness - pituitary dwarfism
217622	Neurosensory deafness with dilated cardiomyopathy
3228	Neurosensory hearing loss - pituitary dwarfism
217622	Neurosensory hearing loss with dilated cardiomyopathy
137596	Neurotrophic keratitis
137596	Neurotrophic keratopathy
165	Neutral lipid storage disease
98907	Neutral lipid storage disease with ichthyosis
98908	Neutral lipid storage disease with myopathy without ichthyosis
98908	Neutral lipid storage myopathy
2690	Neutropenia - monocytopenia - deafness
183707	Neutrophil immunodeficiency syndrome
169142	Neutrophil-specific granule deficiency
624	Nevi flammei
2691	Nevo syndrome
377	Nevoid basal cell carcinoma syndrome
228264	Nevus anelasticus
64754	Nevus comedonicus syndrome
228254	Nevus elasticus
263425	Nevus fusculocoeruleus ophthalmomaxillaris
263432	Nevus of Ito
263425	Nevus of Ota
2612	Nevus sebaceus of Jadassohn
2612	Nevus sebaceus syndrome
83471	Nezelof syndrome
636	NF1
97685	NF1 microdeletion syndrome
137605	NF1-like syndrome
637	NF2
93921	NF3
2678	NF6
69087	NFJ syndrome
91349	NFPA
289356	NGCO
280576	NGPS
2770	NHD
169079	NHEJ1 deficiency
642	NHSA4
64752	NHSA5
247598	NICCD
141179	NICH
3051	Nicolaides-Baraitser syndrome
77292	Niemann-Pick disease type A
77293	Niemann-Pick disease type B
646	Niemann-Pick disease type C
216986	Niemann-Pick disease type C, adult neurologic onset
216981	Niemann-Pick disease type C, classic form
216981	Niemann-Pick disease type C, juvenile neurologic onset
216978	Niemann-Pick disease type C, late infantile neurologic onset
216975	Niemann-Pick disease type C, severe early infantile neurologic onset
216972	Niemann-Pick disease type C, severe perinatal form
99022	Niemann-Pick disease type E
2633	Nievergelt syndrome
1390	Night blindness - skeletal anomalies - dysmorphism
98757	Nigro-spino-dentatal degeneration with nuclear ophthalmoplegia
432	nIHH
2322	Niikawa-Kuroki syndrome
647	Nijmegen breakage syndrome
240760	Nijmegen breakage syndrome-like disorder
99825	Nipah encephalitis
99825	Nipah fever
99825	Nipah virus disease
276608	NI-PHH
59303	NISCH syndrome
1422	Nivelon-Nivelon-Mabille syndrome
86879	NK/T-cell lymphoma
263665	NK-cell enteropathy
86873	NK-cell large granular lymphocyte leukemia
86873	NK-cell LGL leukemia
86879	NKTCL
86893	NLPHL
247868	NLRP12-associated hereditary periodic fever syndrome
98907	NLSDI
98908	NLSDM
607	NM
86867	NMZL
2615	NNS
1884	Noble-Bass-Sherman syndrome
31204	Nocardiosis
98784	Nocturnal frontal lobe epilepsy
86867	Nodal marginal zone B-cell lymphoma
137810	Nodular cutaneous amyloidosis
90393	Nodular lichen myxedematosus
86893	Nodular lymphocyte predominant Hodgkin lymphoma
2149	Nodular neuronal heterotopia
33577	Nodular non-suppurative panniculitis
48372	Nodular regenerative hyperplasia of the liver
98843	Nodular sclerosis classical Hodgkin lymphoma
158772	Nodular urticaria pigmentosa
85196	Nodulosis-arthropathy-osteolysis syndrome
2700	Noma
1451	NOMID syndrome
79450	Non hereditary congenital primary lymphedema
73267	Non-24-hour sleep-wake syndrome
467	Nonacquired combined pituitary hormone deficiency
231720	Nonacquired combined pituitary hormone deficiency with spine abnormalities
631	Non-acquired isolated growth hormone deficiency
602	Nonaka myopathy
97566	Non-amyloid fibrillary glomerulopathy
86861	Non-amyloid MIDD
86861	Non-amyloid monoclonal immunoglobulin deposition disease
79394	Non-bullous congenital ichthyosiform erythroderma
77259	Non-cerebral juvenile Gaucher disease
48372	Non-cirrhotic nodulation
95698	Non-classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency
325529	Non-classic congenital lipoid adrenal hyperplasia due to STAR deficency
216796	Non-deforming osteogenesis imperfecta
1581	Non-distal 10q deletion

ORPHA Number	Disease name
96136	Non-distal deletion 7p
96160	Non-distal deletion 12q
96164	Non-distal deletion 20q
96112	Non-distal duplication 9q
1695	Non-distal duplication 10q
1702	Non-distal duplication 13q
96136	Non-distal monosomy 7p
1581	Non-distal monosomy 10q
96160	Non-distal monosomy 12q
96164	Non-distal monosomy 20q
3306	Non-distal tetrasomy 15q
96112	Non-distal trisomy 9q
1695	Non-distal trisomy 10q
1702	Non-distal trisomy 13q
329469	Non-DS-AMKL
2337	Non-epidermolytic palmoplantar keratoderma
2972	Non-eruption of teeth - maxillary hypoplasia - genu valgum
217629	Non-familial dilated cardiomyopathy
217598	Non-familial hypertrophic cardiomyopathy
217071	Non-familial RCC
217071	Non-familial renal cell carcinoma
217720	Non-familial restrictive cardiomyopathy
357034	Non-familial retinoblastoma
100070	Non-fluent variant PPA
91349	Non-functioning pituitary adenoma
289356	Non-gestational ovarian choriocarcinoma
26137	Non-giant cell granulomatous temporal arteritis with eosinophilia
90185	Non-hereditary late-onset primary lymphedema
357034	Non-hereditary retinoblastoma
163924	Non-herpetic acute limbic encephalitis
329883	Non-hypoproteinemic hypertrophic gastropathy
329918	Non-Ig-mediated membranoproliferative glomerulonephritis
329918	Non-Ig-mediated MPGN
329918	Non-immunoglobulin-mediated membranoproliferative glomerulonephritis
329918	Non-immunoglobulin-mediated MPGN
141179	Non-involuting congenital hemangioma
407	Non-ketotic hyperglycinemia
98890	Non-Leber type optic atrophy with early-onset
79452	Nonne-Milroy lymphedema
84085	Non-neurogenic neurogenic bladder
209989	Non-papillary transitional cell carcinoma of the bladder
209989	Non-papillary urothelial carcinoma
238583	Non-phenylketonuric hyperphenylalaninemia
79651	Non-PKU HPA
99817	Non-polyposis Turcot syndrome
1766	Non-progressive cerebellar ataxia - intellectual deficit
314647	Non-progressive cerebellar ataxia with intellectual deficit
99648	Non-progressive congenital heart block
176	Non-rhizomelic chondrodysplasia punctata
101106	Non-secreting chemodectoma
94080	Non-secreting paraganglioma
169446	Non-skeletal hyper IgE syndrome
91364	Non-specific idiopathic interstitial pneumonia
91364	Non-specific interstitial pneumonia
90031	Non-spherocytic hemolytic anemia due to hexokinase deficiency
35099	Non-syndromic bicornal synostosis
30391	Non-syndromic biliary atresia
300337	Non-syndromic congenital retinal non-attachment
49042	Non-syndromic dentinogenesis imperfecta
49042	Non-syndromic DGI
3366	Non-syndromic metopic craniosynostosis
63440	Non-syndromic oxycephaly
35093	Non-syndromic sagittal synostosis
35098	Non-syndromic unicoronal synostosis
96136	Non-telomeric monosomy 7p
1581	Non-telomeric monosomy 10q
96160	Non-telomeric monosomy 12q
96164	Non-telomeric monosomy 20q
3306	Non-telomeric tetrasomy 15q
96112	Non-telomeric trisomy 9q
1695	Non-telomeric trisomy 10q
1702	Non-telomeric trisomy 13q
209919	Non-Wilsonian hepatic copper toxicosis of infancy and childhood
648	Noonan syndrome
2701	Noonan syndrome-like disorder with loose anagen hair
230	Noradrenaline deficiency
230	Norepinephrine deficiency
314928	Normal pressure hydrocephalus
2254	Norman disease
79255	Norman-Landing disease
306658	Normocalcemic tumoral calcinosis
812	Normomorphic sialidosis
432	Normosmic congenital hypogonadotropic hypogonadism
432	normosmic idiopathic hypogonadotropic hypogonadism
649	Norrie disease
649	Norrie-Warburg disease
75327	North Carolina macular dystrophy
75327	North Carolina macular dystrophy, retinal 1
1947	Northern epilepsy
79293	Norum disease
1134	Nose agenesis
77304	Not NOTCH3-related small vessel disease of the brain
2703	Nova syndrome
2005	Novak syndrome
314928	NPH
3032	NPHP3-related Meckel-like syndrome
634	NS
88616	NS-ARID
417	NSHPT
93606	NSIAD
91364	NSIP
100073	NTOS
98991	Nuclear cataract
314790	Null pituitary adenoma
280234	Null syndrome
54	OA1
1791	OAFNS
374	OAV dysplasia
141132	OAV spectrum
374	OAVS
97297	Oberklaid-Danks syndrome
88643	Obesity - colitis - hypothyroidism - cardiac hypertrophy - developmental delay
217031	Obesity due MC3R deficiency
66628	Obesity due to congenital leptin deficiency
179494	Obesity due to leptin receptor gene deficiency
71529	Obesity due to melanocortin-4 receptor deficiency
71528	Obesity due to prohormone convertase-I deficiency
71526	Obesity due to pro-opiomelanocortin deficiency
99704	Obesity, hyperphagia, and severe developmental delay due toTrkB gene deficiency
1303	Obliterative bronchiolitis
64743	Obliterative portal venopathy
3411	Obstructed hemivagina and ipsilateral renal anomaly
55	OCA
352731	OCA1
79431	OCA1A
79434	OCA1B
352734	OCA1-MP
352737	OCA1-TS
79432	OCA2
79433	OCA3
79435	OCA4
352745	OCA7

ORPHA Number	Disease name
217017	Occipital atretic cephalocele - unusual facies - large feet
268823	Occipital encephalocele
198	Occipital horn syndrome
280640	Occipital malformations of cortical development
280640	Occipital MCD
280640	Occipital pachygyria and polymicrogyria
353351	Occlusive idiopathic juxtafoveolar retinal telangiectasis
51608	Occlusive infantile arteriopathy
1647	OCCS
99889	Occult ectopic ACTH secretion
247834	Occult macular dystrophy
84085	Occult neuropathic bladder
99909	Occupational allergic alveolitis
2704	Ochoa syndrome
247834	OCMD
534	OCR
534	OCRL
664	OCT deficiency
54	Ocular albinism type 1
352740	Ocular albinism with congenital sensorineural deafness
1000	Ocular albinism with late-onset sensorineural deafness
54	Ocular albinism, Nettleship-Falls type
195	Ocular coloboma - imperforate anus
2788	Ocular form of osteogenesis imperfecta
1125	Ocular motor apraxia, Cogan type
99922	Ocular pemphigoid
157962	Oculoauricular syndrome, Schorderet type
1791	Oculo-auriculo-fronto-nasal syndrome
374	Oculoauriculovertebral dysplasia
374	Oculoauriculovertebral spectrum
141132	Oculo-auriculo-vertebral spectrum
2549	Oculoauriculovertebral spectrum with radial defects
374	Oculoauriculovertebral syndrome
2705	Oculocerebral dysplasia
2719	Oculocerebral hypopigmentation syndrome, Cross type
2720	Oculocerebral hypopigmentation syndrome, Preus type
2706	Oculo-cerebro-acral syndrome
1647	Oculocerebrocutaneous syndrome
2707	Oculocerebrofacial syndrome, Kaufman type
2708	Oculo-cerebro-osseous syndrome
534	Oculocerebrorenal dystrophy
534	Oculo-cerebro-renal dystrophy
534	Oculocerebrorenal syndrome
534	Oculo-cerebro-renal syndrome
55	Oculocutaneous albinism
352731	Oculocutaneous albinism type 1
79431	Oculocutaneous albinism type 1A
79434	Oculocutaneous albinism type 1B
79432	Oculocutaneous albinism type 2
79433	Oculocutaneous albinism type 3
79435	Oculocutaneous albinism type 4
352745	Oculocutaneous albinism type 7
79434	Oculocutaneous albinism type Amish
28378	Oculocutaneous tyrosinemia
2709	Oculodental syndrome, Rutherfurd type
2710	Oculodentodigital dysplasia
2710	Oculodentoosseous dysplasia
1305	Oculo-digito-esophageal-duodenal syndrome
3339	Oculoectodermal syndrome
2712	Oculofaciocardiodental syndrome
1876	Oculogastrointestinal muscular dystrophy
1794	Oculomaxillofacial dysostosis
1154	Oculomelic amyoplasia
2713	Oculoosteocutaneous syndrome
99806	Oculootodental syndrome
77302	Oculo-oto-facial dysplasia
2307	Oculo-oto-radial syndrome
2714	Oculo-palato-cerebral dwarfism
2714	Oculo-palato-cerebral syndrome
270	Oculopharyngeal muscular dystrophy
98897	Oculopharyngodistal myopathy
2715	Oculo-reno-cerebellar syndrome
2716	Oculo-skeletal-renal syndrome
2717	Oculotrichoanal syndrome
2718	Oculotrichodysplasia
166272	ODCD
2710	ODDD syndrome
1305	ODED syndrome
999	O'Doherty syndrome
2253	O'Donnell-Pappas syndrome
166272	Odontochondrodysplasia
247685	Odontohypophosphatasia
77295	Odontoleukodystrophy
2724	Odontomatosis - aortae esophagus stenosis
1811	Odontomicronychial dysplasia
2722	Odonto-onycho dysplasia - alopecia
2721	Odonto-onycho-dermal dysplasia
2723	Odontotrichomelic syndrome
69082	Odonto-tricho-ungual-digito-palmar syndrome
69082	Odonto-tricho-ungual-digito-palmar syndrome, Mendoza-Valiente type
1487	ODP
93929	OEIS complex
2676	Oerter-Friedman-Anderson syndrome
2792	OFC syndrome
2712	OFCD syndrome
2750	OFD1
2751	OFD2
2752	OFD3
2753	OFD4
2919	OFD5
2754	OFD6
2755	OFD8
141007	OFD9
2756	OFD10
141000	OFD11
141327	OFD12
141330	OFD13
2750	OFDI
2750	OFDSI
276432	Ogden syndrome
75382	Oguchi disease
75382	Oguchi syndrome
1186	Ohaha syndrome
2728	Ohdo syndrome
2728	Ohdo-Madokoro-Sonoda syndrome
64739	OHSS
1934	Ohtahara syndrome
3411	OHVIRA syndrome
666	OI
216796	OI type 1
216804	OI type 2
216812	OI type 3
216820	OI type 4
216828	OI type 5
2729	Okamoto syndrome
93293	Okihiro syndrome
261638	Okihiro syndrome due to 20q13 microdeletion
261647	Okihiro syndrome due to a point mutation
261638	Okihiro syndrome due to del(20)(q13)
261638	Okihiro syndrome due to monosomy 20q13
69088	OL-EDA-ID
79458	Oley syndrome
478	Olfacto-genital pathological sequence
1957	Olfactory neuroblastoma
85410	Oligoarticular juvenile arthritis
247839	Oligoarticular juvenile arthritis with anti-nuclear antibodies
247846	Oligoarticular juvenile arthritis without anti-nuclear antibodies
251656	Oligoastrocytoma
75378	Oligocone syndrome
75378	Oligocone trichromacy
46484	Oligodendroglial tumor
251627	Oligodendroglioma

ORPHA Number	Disease name
99798	Oligodontia
300576	Oligodontia - cancer predisposition syndrome
2260	Oligomeganephronia
2260	Oligomeganephronic renal hypoplasia
137831	Oligophrenin-1 syndrome
2920	Oliver syndrome
3363	Oliver-McFarlane syndrome
2732	Olivopontocerebellar atrophy - deafness
166063	Olivopontocerebellar hypoplasia
296	Ollier disease
659	Olmsted syndrome
247834	OMD
39041	Omenn syndrome
2741	OMM syndrome
2733	Omodysplasia
660	Omphalocele
93929	Omphalocele - cloacal exstrophy - imperforate anus - spinal defect
3164	Omphalocele syndrome, Shprintzen-Goldberg type
490	Omphalomesenteric cyst
210115	OMPP
319266	Omsk hemorrhagic fever
3191	Onat syndrome
2737	Onchocerciasis
137675	Oncocytic cardiomyopathy
352540	Oncogenic hypophosphatemic osteomalacia
352540	Oncogenic osteomalacia
661	Ondine curse
661	Ondine syndrome
99803	Ondine-Hirschsprung disease
99803	Ondine-Hirschsprung syndrome
2739	ONMR syndrome
300504	Onychocytic matricoma
238744	Onycho-digito-mammary syndrome
79153	Onychodystrophy totalis
300512	Onychomatricoma
2614	Onychoosteodysplasia
2739	Onycho-tricho-dysplasia - neutropenia
2786	OOCHS
99806	OOD
2721	OODD
98890	OPA2
67036	OPA3, autosomal dominant
49042	Opalescent teeth without OI
49042	Opalescent teeth without osteogenesis imperfecta
268363	Open iniencephaly
137831	OPHN1 syndrome
1106	Ophthalmoacromelic syndrome
2741	Ophthalmomandibulomelic dysplasia
1186	Ophthalmoplegia - hypotonia - ataxia - hypoacusis - athetosis
2743	Ophthalmoplegia - intellectual deficit - lingua scrotalis
2742	Ophthalmoplegia - myalgia - tubular aggregates
2745	Opitz G/BBB syndrome
2745	Opitz syndrome
1308	Opitz trigonocephaly syndrome
97297	Opitz trigonocephaly-like syndrome
1786	Opitz-Caltabiano syndrome
2745	Opitz-Frias syndrome
93932	Opitz-Kaveggia syndrome
2458	Opitz-Reynolds-FitzGerald syndrome
270	OPMD
256	Oppenheim dystonia
2788	OPPG
2746	Opsismodysplasia
1183	Opsoclonus-myoclonus syndrome
1215	Optic atrophy - deafness- polyneuropathy - myopathy
67047	Optic atrophy plus syndrome
98890	Optic atrophy type 2
313800	Optic nerve edema-splenomegaly syndrome
2086	Optic pathway glioma
31142	Oral erosive lichen
357154	Oral submucous fibrosis
2750	Oral-facial-digital syndrome type 1
2751	Oral-facial-digital syndrome type 2
2752	Oral-facial-digital syndrome type 3
2753	Oral-facial-digital syndrome type 4
2919	Oral-facial-digital syndrome type 5
2754	Oral-facial-digital syndrome type 6
2755	Oral-facial-digital syndrome type 8
141007	Oral-facial-digital syndrome type 9
2756	Oral-facial-digital syndrome type 10
141000	Oral-facial-digital syndrome type 11
141327	Oral-facial-digital syndrome type 12
141007	Oral-facial-digital syndrome with retinal abnormalities
2755	Oral-facial-digital syndrome, Edwards type
141000	Oral-facial-digital syndrome, Gabrielli type
1647	Orbital cyst with cerebral and focal dermal malformations
52994	Orbital leiomyoma
268139	Orbital medulloepithelioma
2612	Organoid nevus syndrome
166421	Orgasm-induced seizures
49041	Ormond disease
414	Ornithine aminotransferase deficiency
664	Ornithine carbamoyltransferase deficiency
664	Ornithine transcarbamylase deficiency
2749	Oroacral syndrome
2319	Orocraniodigital syndrome
2750	Orofaciodigital syndrome type 1
2751	Orofaciodigital syndrome type 2
2752	Orofaciodigital syndrome type 3
2753	Orofaciodigital syndrome type 4
2919	Orofaciodigital syndrome type 5
2754	Orofaciodigital syndrome type 6
2755	Orofaciodigital syndrome type 8
141007	Orofaciodigital syndrome type 9
2756	Orofaciodigital syndrome type 10
141000	Orofaciodigital syndrome type 11
141327	Orofaciodigital syndrome type 12
141330	Orofaciodigital syndrome type 13
2756	Orofaciodigital syndrome with fibular aplasia
141007	Orofaciodigital syndrome with retinal abnormalities
141000	Orofaciodigital syndrome, Gabrielli type
2919	Orofaciodigital syndrome, Thurston type
93958	Oromandibular dystonia
2749	Oro-mandibular-limb hypogenesis syndrome
141077	Oropharyngeal teratoma
30	Oroticaciduria
30	Orotidylic decarboxylase deficiency
64692	Oroya fever
93382	Osebold-Remondini syndrome
97335	Osgood-Schlatter disease
2760	OSLAM syndrome
729	Osler-Vaquez disease
1427	OSMED
357154	OSMF
140436	Osseous vascular malformation
73230	Ossification anomalies - psychomotor development delay
58040	Osteoblastoma
251262	Osteochondritis dissecans and short stature
3314	Osteochondritis of phalangeal epiphyses
2054	Osteochondritis of tarsal/metatarsal bone
2380	Osteochondritis of the capital femoral epiphysis
97332	Osteochondritis of the lunate bone
97335	Osteochondritis of the tibial tubercle
2653	Osteochondrodysplatic dwarfism - deafness - retinitis pigmentosa
2653	Osteochondrodysplatic nanism - deafness - retinitis pigmentosa
800	Osteochondromuscular dystrophy
2768	Osteochondrosis deformans tibiae
97337	Osteochondrosis of patella

ORPHA Number	Disease name
3314	Osteochondrosis of phalangeal epiphyses
2380	Osteochondrosis of the capital femoral epiphysis
97336	Osteochondrosis of the capital humerus
97332	Osteochondrosis of the lunate bone
2054	Osteochondrosis of the tarsal bone
97335	Osteochondrosis of the tibial tubercle
2763	Osteocraniosplenic syndrome
2763	Osteocraniostenosis
2484	Osteodysplasty, Melnick-Needles type
249	Osteofibrous dysplasia
666	Osteogenesis imperfecta
2771	Osteogenesis imperfecta - congenital joint contractures
2773	Osteogenesis imperfecta - retinopathy - seizures - intellectual deficit
216796	Osteogenesis imperfecta type 1
216804	Osteogenesis imperfecta type 2
216812	Osteogenesis imperfecta type 3
216820	Osteogenesis imperfecta type 4
216828	Osteogenesis imperfecta type 5
668	Osteogenic sarcoma
2645	Osteoglophonic dwarfism
2777	Osteomesopyknosis
2780	Osteopathia striata - cranial sclerosis
2779	Osteopathia striata - pigmentary dermopathy - white forelock
2324	Osteopenia - intellectual deficit - sparse hair
91133	Osteopenia - myopia - hearing loss - intellectual deficit - facial dysmorphism
178389	Osteopetrosis - hypogammaglobulinemia
53	Osteopetrosis autosomal dominant type 2
2785	Osteopetrosis with renal tubular acidosis
94063	Osteopoikilosis - short stature - intellectual deficit
2787	Osteoporosis - macrocephaly - blindness - joint hyperlaxity
2786	Osteoporosis - oculocutaneous hypopigmentation syndrome
2788	Osteoporosis - pseudoglioma
666	Osteopsathyrosis
668	Osteosarcoma
2760	Osteosarcoma - limb anomalies - erythroid macrocytosis
178377	Osteosclerosis - developmental delay - craniosynostosis
75325	Osteosclerosis - ichthyosis - premature ovarian failure
2905	Osteosclerotic myeloma
1338	Ostravik-Lindemann-Solberg syndrome
99965	O'Sullivan-McLeod syndrome
664	OTC deficency
166457	Other forms of non-paraneoplastic limbic encephalitis
206606	Other muscle weakness and/or chronic muscle pain
2791	Otodental dysplasia
2791	Otodental syndrome
2792	Otofaciocervical syndrome
141136	Otomandibular dysostosis
141136	Otomandibular syndrome
2793	Oto-onycho-peroneal syndrome
669	Otopalatodigital syndrome
90650	Otopalatodigital syndrome type 1
90652	Otopalatodigital syndrome type 2
1427	Otospondylomegaepiphyseal dysplasia
69082	OTUDP syndrome
1179	Ouvrier-Billson syndrome
213504	Ovarian adenocarcinoma
35808	Ovarian cancer of sex cord-stromal origin
213512	Ovarian carcinosarcoma
314473	Ovarian fibroma
314478	Ovarian fibrothecoma
35807	Ovarian germ cell cancer
35807	Ovarian germ cell malignant tumor
206484	Ovarian gonadoblastoma
64739	Ovarian hyperstimulation syndrome
99916	Ovarian malignant Sertoli-Leydig cell tumor
35808	Ovarian malignant tumor of sex cord-stromal origin
99916	Ovarian Sertoli-Leydig cell cancer
206473	Ovarian tumor of low malignant potential
99853	Ovarioleukodystrophy
137634	Overgrowth - macrocephaly - facial dysmorphism
3203	Overhydrated hereditary stomatocytosis
206572	Overlap myositis
326	Owren disease
31	Oxoglutaricaciduria
33572	Oxoprolinuria due to oxoprolinase deficiency
79302	Oxysterol 7-alpha-hydroxylase deficiency
36355	P2Y12 deficiency
35664	P5CS deficiency
35120	P5N deficiency
98971	PACD
2796	Pachydermoperiostosis
94084	Pachygyria - epilepsy - intellectual deficit - dysmorphism
2798	Pachygyria - intellectual deficit - epilepsy
2309	Pachyonychia congenita
1952	Pacman dysplasia
140989	PACNS
706	PAD
441	PAF
180275	Paget disease of the nipple
52430	Pagetoid amyotrophic lateral sclerosis
52430	Pagetoid neuroskeletal syndrome
178517	Pagetoid reticulosis, Woringer-Kolopp type
180275	Paget's disease of the nipple
357131	Paget-Schrotter disease
991	PAGOD syndrome
716	PAH deficiency
1993	Pai syndrome
37202	Painful bladder syndrome
324636	Painful bruising syndrome
99736	Painful congenital myotonia
99736	Painful myotonia
64686	Painful ophthalmoplegia
300501	Painful orbital and systemic neurofibromas-marfanoid habitus syndrome
90797	PAIS
1388	Palatodigital syndrome, Catel-Manzke type
171695	Pallidopyramidal syndrome
672	Pallister-Hall syndrome
884	Pallister-Killian syndrome
2804	Pallister-W syndrome
737	Palmar, plantar and disseminated porokeratosis
2184	Palmer-Pagon syndrome
659	Palmoplantar and periorificial keratoderma
50944	Palmoplantar hyperkeratosis - cystic eyelids - hypodontia - hypotrichosis
2342	Palmoplantar hyperkeratosis - periodontopathia - onychogryposis
85112	Palmoplantar hyperkeratosis - XX sex reversal - predisposition to squamous cell carcinoma
34217	Palmoplantar hyperkeratosis with arrythmogenic cardiomyopathy
140966	Palmoplantar hyperkeratosis, Nagashima type
2202	Palmoplantar hyperkeratosis-deafness syndrome
2198	Palmoplantar hyperkeratosis-esophageal carcinoma syndrome
2202	Palmoplantar hyperkeratosis-hearing loss syndrome
384	Palmoplantar hyperkeratosis-sclerodactyly syndrome
2201	Palmoplantar hyperkeratosis-spastic paralysis syndrome
86919	Palmoplantar keratoderma - clinodactyly
50944	Palmoplantar keratoderma - cystic eyelids - hypodontia - hypotrichosis
2342	Palmoplantar keratoderma - periodontopathia - onychogryposis
85112	Palmoplantar keratoderma - XX sex reversal - predisposition to squamous cell carcinoma

ORPHA Number	Disease name
1010	Palmoplantar keratoderma and congenital alopecia, Stevanovic type
1366	Palmoplantar keratoderma and congenital alopecia, Wallis type
34217	Palmoplantar keratoderma with arrythmogenic cardiomyopathy
140966	Palmoplantar keratoderma, Nagashima type
2202	Palmoplantar keratoderma-deafness syndrome
2198	Palmoplantar keratoderma-esophageal carcinoma syndrome
2202	Palmoplantar keratoderma-hearing loss syndrome
384	Palmoplantar keratoderma-sclerodactyly syndrome
2201	Palmoplantar keratoderma-spastic paralysis syndrome
736	Palmoplantar porokeratosis of Mantoux
163927	Palmoplantar pustulosis
612	PAM
93564	PAN, pediatric onset
98815	Panayiotopoulos syndrome
93292	Pancreatic adenoma
65288	Pancreatic and cerebellar agenesis
28455	Pancreatic beta cell agenesis with neonatal diabetes mellitus
217074	Pancreatic cancer
217074	Pancreatic carcinoma
97282	Pancreatic cholera
309108	Pancreatic colipase deficiency
97253	Pancreatic endocrine tumor
2255	Pancreatic hypoplasia - diabetes - congenital heart disease
199337	Pancreatic insufficiency - anemia - hyperostosis
97278	Pancreatic polypeptidoma
309031	Pancreatic triacylglycerol lipase deficiency
309031	Pancreatic triglyceride lipase deficiency
677	Pancreatoblastoma
317473	Pancytopenia due to IKZF1 mutations
66624	PANDAS
95513	Panhypophysitis
90695	Panhypopituitarism
97336	Panner disease
90159	Panniculitis and localized lipodystrophy
157850	Pantothenate-kinase-associated neurodegeneration
69126	PAPA syndrome
213817	Papillary carcinoma of the cervix uteri
213726	Papillary carcinoma of the corpus uteri
208600	Papillary fibroelastoma of the heart
251962	Papillary glioneuronal tumor
146	Papillary or follicular thyroid carcinoma
319298	Papillary renal cell adenocarcinoma
319298	Papillary renal cell carcinoma
251915	Papillary tumour of the pineal region
2807	Papilloma of choroid plexus
2750	Papillon-Leage-Psaume syndrome
678	Papillon-Lefèvre syndrome
1475	Papillo-renal syndrome
86819	Papular atrichia
228264	Papular elastorrhexis
313936	Papular epidermal nevi with skyline basal cell layers syndrome
86795	Papular mucinosis
90395	Papular mucinosis of infancy
158008	Papular xanthoma
679	Papulosis atrophican maligna
99056	Parachute tricuspid valve
73260	Paracoccidioidomycosis
324299	Paraganglioma - somatostatinoma - polycythemia
97286	Paraganglioma and gastric stromal sarcoma
326	Parahemophilia
141242	Paramedian nasal cleft
684	Paramyotonia congenita
684	Paramyotonia congenita of Von Eulenburg
2812	Parana hard-skin syndrome
99889	Paraneoplastic Cushing syndrome
63455	Paraneoplastic pemphigus
71505	Paraneoplastic retinopathy
231445	Paraparetic variant of GBS
231445	Paraparetic variant of Guillain-Barré syndrome
2823	Paraplegia - brachydactyly - cone-shaped epiphysis
2824	Paraplegia - intellectual deficit - hyperkeratosis
31827	Paraquat poisoning
2646	Parastremmatic dwarfism
143	Parathyroid carcinoma
99745	Paratyphoid fever
2825	PARC syndrome
268826	Parietal encephalocele
60015	Parietal foramina
251290	Parietal foramina with cleidocranial dysostosis
251290	Parietal foramina with cleidocranial dysplasia
851	Paris-Trousseau thrombocytopenia
306674	PARK9
90307	Parkes Weber syndrome
171695	Parkinsonian-pyramidal syndrome
314632	Parkinsonim due to ATP13A2 deficiency
178509	Parkinsonism with alveolar hypoventilation and mental depression
97355	Parkinsonism with dementia of Guadeloupe
90020	Parkinsonism-dementia-ALS complex
90035	Paroxysmal cold hemoglobinuria
1431	Paroxysmal dyskinesia
53583	Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity
98811	Paroxysmal exertion-induced dyskinesia
46348	Paroxysmal extreme pain disorder
157835	Paroxysmal hemicrania
98812	Paroxysmal hypnogenic dyskinesia
98809	Paroxysmal kinesigenic choreathetosis
98809	Paroxysmal kinesigenic dyskinesia
31709	Paroxysmal kinesigenic dyskinesia and infantile convulsions
447	Paroxysmal nocturnal hemoglobinuria
98810	Paroxysmal non-kinesigenic dyskinesia
3286	Paroxysmal ventricular fibrillation
1214	Parry-Romberg syndrome
574	Partial 21q monosomy
79087	Partial acquired lipodystrophy
2805	Partial agenesis of the pancreas
381	Partial albinism - immunodeficiency
90797	Partial androgen insensitivity syndrome
90797	Partial androgen resistance syndrome
1330	Partial atrioventricular canal
1646	Partial chromosome Y deletion
98992	Partial congenital cataract
98950	Partial cryptophthalmia
90076	Partial deep dermal and full thickness burns
79312	Partial deficiency of methylmalonyl-CoA mutase
2308	Partial deletion 11q
261318	Partial duplication of chromosome 20p
261318	Partial duplication of the short arm of chromosome 20
101046	Partial epilepsy with auditory aura
101046	Partial epilepsy with auditory features
2704	Partial facial palsy with urinary abnormalities
744	Partial gigantism - nevi - hemihypertrophy - macrocephaly
254693	Partial hydatidiform mole
79292	Partial LCAT deficiency
343	Partial mevalonate kinase deficiency
254693	Partial molar pregnancy
2805	Partial pancreatic agenesis
93178	Partial prune belly syndrome
157769	Partial situs inversus
261318	Partial trisomy 20p

ORPHA Number	Disease name
261318	Partial trisomy of chromosome 20p
261318	Partial trisomy of the short arm of chromosome 20
85453	Partington disease
94083	Partington syndrome
94083	Partington-Mulley syndrome
295	Parvovirus antenatal infection
1394	Pascual-Castroviejo syndrome type 1
42775	Pascual-Castroviejo syndrome type 2
289478	PASH syndrome
1252	Pashayan syndrome
2278	Passwell-Goodman-Siprkowski syndrome
3378	Patau syndrome
86789	Patella aplasia/hypoplasia
295041	Patella aplasia/hypoplasia, bilateral
295038	Patella aplasia/hypoplasia, unilateral
1428	Patellofemoral syndrome
706	Patent arterial duct
228190	Patent arterial duct - bicuspid aortic valve - hand anomalies
706	Patent ductus arteriosus
228190	Patent ductus arteriosus - bicuspid aortic valve - hand anomalies
46627	Patent ductus arteriosus with facial dysmorphism and abnormal fifth digits
99108	Patent foramen ovale
254531	Paternal 14q32.2 hypomethylation syndrome
254525	Paternal 14q32.2 microdeletion syndrome
261304	Paternal 20q13.2q13.3 microdeletion syndrome
261304	Paternal 20q13.2-q13.3 microdeletion syndrome
254525	Paternal del(14)(q32.2)
261304	Paternal del(20)(q13.2q13.3)
254525	Paternal monosomy 14q32.2
261304	Paternal monosomy 20q13.2q13.3
261304	Paternal monosomy 20q13.2-q13.3
251004	Paternal uniparental disomy of chromosome 1
96190	Paternal uniparental disomy of chromosome 5
96191	Paternal uniparental disomy of chromosome 6
96192	Paternal uniparental disomy of chromosome 7
99324	Paternal uniparental disomy of chromosome 13
96334	Paternal uniparental disomy of chromosome 14
96194	Paternal uniparental disomy of chromosome 20
96195	Paternal uniparental disomy of chromosome 21
261524	Paternal uniparental disomy of chromosome X
2439	Patterson-Stevenson syndrome
2439	Patterson-Stevenson-Fontaine syndrome
79136	PATX
85410	Pauciarticular chronic arthritis
247839	Pauciarticular chronic arthritis with anti-nuclear antibodies
247846	Pauciarticular chronic arthritis without anti-nuclear antibodies
93126	Pauci-immune glomerulonephritis
97563	Pauci-immune glomerulonephritis with ANCA
97564	Pauci-immune glomerulonephritis without ANCA
75373	PBCRA
289666	PBL
2309	PC
54247	PCA
88628	PCARP
231426	PCB variant of GBS
231426	PCB variant of Guillain-Barré syndrome
247198	PCCA
178544	PCDLBCL,LT
178540	PCFCL
90035	PCH
97249	PCH with optic atrophy
97249	PCH without dyskinesia
2254	PCH1
2524	PCH2
97249	PCH3
166063	PCH4
166068	PCH5
166073	PCH6
284339	PCH7
324569	PCH8
71528	PCI deficiency
2924	PCLD
178536	PCMZL
46135	PCNSL
101330	PCT
163746	PCWH
90020	PDALS
293462	PDCD
289157	PDDRI
765	PDH
79246	PDH phosphatase deficiency
79243	PDHAD
255138	PDHBD
765	PDHC
2796	PDP
85453	PDR
75496	PDS
699	Pearson syndrome
2835	Pectus excavatum - macrocephaly - dysplastic nails
98811	PED
66624	Pediatric autoimmune disorders associated with Streptococcus infections
66624	Pediatric autoimmune neuropsychiatric disorders associated with Streptococcus infections
93682	Pediatric Castleman disease
93564	Pediatric polyarteritis nodosa
93552	Pediatric systemic lupus erythematosus
93567	Pediatric systemic scleroderma
93567	Pediatric systemic sclerosis
817	Peeling skin syndrome
2836	PEHO syndrome
99807	PEHO-like syndrome
48686	PEL
702	Pelizaeus-Merzbacher brain sclerosis
702	Pelizaeus-Merzbacher disease
280229	Pelizaeus-Merzbacher disease in female carriers
280210	Pelizaeus-Merzbacher disease type II
280219	Pelizaeus-Merzbacher disease, classic form
280210	Pelizaeus-Merzbacher disease, connatal form
280234	Pelizaeus-Merzbacher disease, null syndrome
280224	Pelizaeus-Merzbacher disease, transitional form
280293	Pelizaeus-Merzbacher-like due to AIMP1 mutation
280282	Pelizaeus-Merzbacher-like due to GJC2 mutation
280288	Pelizaeus-Merzbacher-like due to HSPD1 mutation
97352	Pellagra
2837	Pellagra-like skin rash - neurological manifestations
137672	Pellucid marginal degeneration
2840	Pelvic dysplasia - arthrogryposis of lower limbs
83628	PELVIS syndrome
93333	Pelviscapular dysplasia
2839	Pelvis-shoulder dysplasia
63275	Pemphigoid gestationis
79480	Pemphigus erythematosus
79481	Pemphigus foliaceus
79479	Pemphigus vegetans
704	Pemphigus vulgaris
994	Pena-Shokeir syndrome type 1
1466	Pena-Shokeir syndrome type 2
705	Pendred syndrome

ORPHA Number	Disease name
49	Penile agenesis
49	Penis agenesis
2842	Penoscrotal transposition
313936	PENS syndrome
11	Pentasomy X
11	Penta-X
2843	Pentosuria
352447	PEO - myopathy - emaciation
2905	PEP syndrome
2880	PEPCK deficiency
79316	PEPCK1 deficiency
79317	PEPCK2 deficiency
2576	Perheentupa syndrome
767	Periarteritis nodosa
2847	Pericardial and diaphragmatic defect
2576	Pericardial constriction - growth failure
58208	Pericarditis
2848	Pericarditis - arthropathy - camptodactyly
137577	Perinatal asphyxia
137577	Perinatal hypoxia
313855	Perinatal lethal bent bone dysplasia
85212	Perinatal lethal Gaucher disease
247623	Perinatal lethal hypophosphatasia
247623	Perinatal lethal phosphoethanolaminuria
247623	Perinatal lethal Rathburn disease
83628	Perineal hemangioma - external genitalia malformations - lipomyelomeningocele - vesicorenal abnormalities - imperforate anus
95706	Perineal, scrotal or penoscrotal hypospadias
85102	Perineurioma
342	Periodic disease
42642	Periodic fever-aphtous stomatitis-pharyngitis-adenopathy syndrome
79136	Periodic vestibulocerebellar ataxia
139426	Perioral myoclonia with absences
563	Peripartum cardiomyopathy
163746	Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease
1795	Peripheral dysostosis
252164	Peripheral fibroblastoma
2400	Peripheral motor neuropathy - dysautonomia
84142	Peripheral nerve hyperexcitability
213812	Peripheral neuroectodermal cancer of the cervix uteri
213630	Peripheral neuroectodermal cancer of the corpus uteri
90120	Peripheral neuropathy and optic atrophy
171848	Peripheral neuropathy, Fiskerstrand type
178040	Peripheral precocious puberty
97927	Peripheral resistance to thyroid hormones
168816	Peritoneal cystic mesothelioma
171676	Periventricular leukomalacia
98892	Periventricular nodular heterotopia
2849	Perlman syndrome
99885	Permanent neonatal diabetes mellitus
65288	Permanent neonatal diabetes mellitus - pancreatic and cerebellar agenesis
97557	Permanent proteinuria with focal and segmental hyalinosis without nephrotic syndrome
2850	Perniola-Krajewska-Carnevale syndrome
2971	Peroxisomal acyl-CoA oxidase deficiency
93598	Peroxisomal alanine-glyoxylate aminotransferase deficiency
2855	Perrault syndrome
75374	PERRS
178509	Perry syndrome
99120	Persistent eustachian valve
91495	Persistent fetal vasculature syndrome
99076	Persistent fifth aortic arch
91495	Persistent hyperplastic primary vitreous
99109	Persistent left superior caval vein connecting to the left-sided atrium
99109	Persistent left superior vena cava connecting to the left-sided atrium
99109	Persistent left SVC connecting to the left-sided atrium
2856	Persistent Müllerian derivatives
2856	Persistent Müllerian duct syndrome
706	Persistent patency of the arterial duct
97341	Persistent placoid maculopathy
300324	Persistent polyclonal B-cell lymphocytosis
300324	Persistent polyclonal B-cell lymphocytosis with binucleated lymphocytes
2380	Perthes disease
1489	Pertussis
708	Peters anomaly
101033	Peters anomaly - cataract
709	Peters anomaly with short limb dwarfism
708	Peters congenital glaucoma
709	Peters-plus syndrome
2776	Petit-Fryns syndrome
2963	Petty-Laxova-Wiedemann syndrome
2869	Peutz-Jeghers syndrome
42642	PFAPA syndrome
90042	PFCP
710	Pfeiffer syndrome
93258	Pfeiffer syndrome type 1
93259	Pfeiffer syndrome type 2
93260	Pfeiffer syndrome type 3
3224	Pfeiffer-Kapferer syndrome
2921	Pfeiffer-Mayer syndrome
2871	Pfeiffer-Palm-Teller syndrome
2872	Pfeiffer-Singer-Zschiesche syndrome
33577	Pfeiffer-Weber-Christian syndrome
2019	PFFD
172	PFIC
79306	PFIC1
79304	PFIC2
79305	PFIC3
91495	PFVS
319646	PGM-CDG syndrome
251962	PGNT
756	PHA type 1
757	PHA2
88938	PHA2A
88939	PHA2B
88940	PHA2C
300525	PHA2D
300530	PHA2E
42775	PHACE syndrome
209959	Phaco-allergic endophthalmitis
209959	Phaco-anaphylactic uveitis
209959	Phacoanaphylaxis
209959	Phaco-anaphylaxis
209959	Phaco-antigenic endophthalmitis
757	PHAII
209959	Phako-anaphylactic endophthalmitis
209959	Phakoanaphylactic uveitis
209959	Phako-anaphylactic uveitis
79483	Phakomatosis cesioflammea
79484	Phakomatosis cesiomarmorata
2874	Phakomatosis pigmentokeratotica
2875	Phakomatosis pigmentovascularis
79483	Phakomatosis pigmentovascularis type 2
79485	Phakomatosis pigmentovascularis type 3
79484	Phakomatosis pigmentovascularis type 5
79485	Phakomatosis spilorosea
352636	Phalangeal acro-osteolysis
352636	Phalangeal microgeodic syndrome
73	Phantom bone disease
171848	PHARC syndrome
423	Pharmacogenetic myopathy of anesthesia
231426	Pharyngeal-cervical-brachial variant of Guillain-Barré syndrome
231426	Pharyngeal-cervical-brachial weakness
231426	Pharyngo-cervico-brachial variant of GBS
231426	Pharyngo-cervico-brachial variant of Guillain-Barré syndrome
2876	PHAVER syndrome
228410	PHD syndrome
48652	Phelan-McDermid syndrome
1919	Phenobarbital antenatal infection

ORPHA Number	Disease name
1919	Phenobarbital embryopathy
84064	Phenotypic diarrhea
716	Phenylalanine hydroxylase deficiency
716	Phenylketonuria
226	Phenylketonuria type 2
2209	Phenylketonuric embryopathy
1912	Phenytoin embryofetopathy
254723	PHID
69084	PHNED
294975	Phocomelia
2878	Phocomelia - ectrodactyly - deafness - sinus arrhythmia
3439	Phocomelia - thrombocytopenia - encephalocele - urogenital malformations
2879	Phocomelia, Schinzel type
534	Phosphatidylinositol 4,5-biphosphate 5-phosphatase deficiency
79316	Phosphoenolpyruvate carboxykinase 1 deficiency
79317	Phosphoenolpyruvate carboxykinase 2 deficiency
2880	Phosphoenolpyruvate carboxykinase deficiency
436	Phosphoethanolaminuria
711	Phosphoglucomutase 1 deficiency
79318	Phosphomannomutase 2 deficiency
79319	Phosphomannose isomerase deficiency
3222	Phosphoribosylpyrophosphate synthetase superactivity
284417	Phosphoserine aminotransferase deficiency
166409	Photosensitive epilepsy
91495	PHPV
180261	Phyllode tumor
180261	Phylloide tumor
773	Phytanic acid oxidase deficiency
2882	Phytosterolemia
670	PIBIDS syndrome
505	Piccardi-Lassueur-Little syndrome
2885	Piebald trait - neurologic defects
2884	Piebaldism
156723	Piepkorn dysplasia
2886	Pierre Robin sequence - congenital heart defect - talipes
2888	Pierre Robin sequence - faciodigital anomaly
3450	Pierre Robin sequence - fetal chondrodysplasia
1388	Pierre Robin sequence - hyperphalangy - clinodactyly
3104	Pierre Robin sequence - oligodactyly
2886	Pierre Robin syndrome - congenital heart defect - talipes
2888	Pierre Robin syndrome - faciodigital anomaly
3450	Pierre Robin syndrome - fetal chondrodysplasia
1388	Pierre Robin syndrome - hyperphalangy - clinodactyly
1388	Pierre Robin syndrome with hyperphalangy and clinodactyly
2670	Pierson syndrome
217557	PIG
99908	Pigeon-breeder lung disease
83639	PIGM-CDG syndrome
978	Pigment anomaly - ectrodactyly - hypodontia
999	Pigmentary disorder with hearing loss
64755	Pigmentary hairy epidermal nevus
435	Pigmentary mosaicism, Ito type
313808	Pigmentary orthochromatic leukodystrophy
254723	Pigmented hypertrichosis with insulin dependent diabetes mellitus syndrome
251295	Pigmented paravenous retinochoroidal atrophy
66627	Pigmented villonodular synovitis
169	Pili annulati
720	Pili bifurcati
719	Pili canulati
79492	Pili gemini
79492	Pili multigemini
2889	Pili torti
2891	Pili torti - developmental delay - neurological abnormalities
2890	Pili torti - onychodysplasia
1410	Pili trianguli et canaliculi
2741	Pillay syndrome
251612	Pilocytic astrocytoma
2892	Pilodental dysplasia - refractive errors
228379	Pilomatrix dysplasia
91414	Pilomatrixoma
251615	Pilomyxoid astrocytoma
2894	Pilotto syndrome
251919	Pineal parenchymal tumor of intermediate differenciation
251909	Pineoblastoma
251912	Pineocytoma
49382	Pingelapese blindness
3353	Pinheiro-Freire Maia-Miranda syndrome
247165	Pink disease
279904	PIOL
34	Pipecolic acidemia
2896	Pitt-Hopkins syndrome
221150	Pitt-Hopkins-like syndrome
93395	Pitt-Williams brachydactyly
251623	Pituicytoma
95613	Pituitary apoplexy
300385	Pituitary carcinoma
96253	Pituitary corticotroph micro-adenoma
91354	Pituitary deficiency due to empty sella turcica syndrome
91350	Pituitary deficiency due to Rathke's pouch cysts
96253	Pituitary dependent Cushing syndrome
91351	Pituitary dermoid and epidermoid cysts
2965	Pituitary lactotrophic adenoma
95496	Pituitary stalk interruption syndrome
91347	Pituitary thyrotrophic adenoma
2897	Pityriasis rubra pilaris
1078	Piussan-Lenaerts-Mathieu syndrome
2869	PJS
157850	PKAN
216873	PKAN, atypical form
216866	PKAN, classic form
238455	PKDYS
716	PKU
226	PKU type 2
99928	Placental site trophoblastic tumor
707	Plague
300359	PLAID
79141	Plamoplantar hyperkeratosis nummularis
79141	Plamoplantar keratoderma nummularis
199251	Plantar fibromatosis
251515	Plantar flexion contracture
158769	Plaque-form urticaria pigmentosa
29073	Plasma cell myeloma
329	Plasma thromboplastin antecedent deficiency
289666	Plasmablastic lymphoma
86855	Plasmacytoma
722	Plasminogen deficiency type 1
721	Platelet alpha-granule deficiency
99146	Platelet function disease associated with renal insufficiency
79434	Platinum oculocutaneous albinism
85166	Platyspondylic dysplasia, Torrance type
85166	Platyspondylic dysplasia, Torrance-Luton type
1417	Platyspondylic lethal chondrodysplasia
85166	Platyspondylic lethal skeletal dysplasia, Torrance type
2899	Platyspondyly - amelogenesis imperfecta
137807	PLCA
300359	PLCG2-associated antibody deficiency and immune dysregulation
137810	PLCNA
99969	Pleomorphic liposarcoma
293199	Pleomorphic rhabdomyosarcoma
293190	Pleomorphic undifferentiated sarcoma
251607	Pleomorphic xanthoastrocytoma
99131	Pleuro-pericardial cyst

ORPHA Number	Disease name
64742	Pleuropulmonary blastoma
284343	Pleuropulmonary blastoma family tumor susceptibility syndrome
284343	Pleuro-pulmonary blastoma family tumor susceptibility syndrome
99933	Pleuro-pulmonary blastoma type I
99934	Pleuro-pulmonary blastoma type II
99935	Pleuro-pulmonary blastoma type III
2770	PLOSL
2770	PLO-SL
2375	Plott syndrome
280234	PLP1 null syndrome
678	PLS*
35689	PLS*
99969	PLS*
85166	PLSD-T
2708	Plum syndrome
54028	Plummer-Vinson syndrome
732	PM*
764	PM*
702	PMD
2856	PMDS
352596	PMED
280282	PMLD1
79318	PMM2-CDG syndrome
26790	PMP
324977	PMSB8 deficiency
99885	PNDM
64741	Pneumoblastoma
55655	Pneumococcal meningitis
723	Pneumocystosis
90066	Pneumonia caused by Pseudomonas aeruginosa infection
447	PNH
760	PNP deficiency
79096	PNPO deficiency
79096	PNPO-related neonatal epileptic encephalopathy
246	POADS
2905	POEMS syndrome
2825	Poikiloderma - alopecia - retrognathism - cleft palate
2908	Poikiloderma of Kindler
2909	Poikiloderma of Rothmund-Thomson
221008	Poikiloderma of Rothmund-Thomson type 1
221016	Poikiloderma of Rothmund-Thomson type 2
221046	Poikiloderma with neutropenia
221046	Poikiloderma with neutropenia, Clericuzio type
279947	POIS
130	Pokkuri death syndrome
2911	Poland anomaly
2911	Poland sequence
2911	Poland syndrome
313808	POLD
2912	Poliomyelitis
330009	Poliomyelitis in patients with immunodeficiencies deemed at risk
29207	Polyarteritis enterica
767	Polyarteritis nodosa
85435	Polyarthritis with rheumatoid factor
85408	Polyarthritis without rheumatoid factor
247854	Polyarthritis without rheumatoid factor with anti-nuclear antibodies
247861	Polyarthritis without rheumatoid factor without anti-nuclear antibodies
2770	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy
2795	Polycystic ovaries - urethral sphincter dysfunction
729	Polycythemia rubra vera
729	Polycythemia vera
2754	Polydactyly - cleft lip/palate - psychomotor retardation
93339	Polydactyly of a biphalangeal thumb
93336	Polydactyly of a triphalangeal thumb
295150	Polydactyly of a triphalangeal thumb, bilateral
295148	Polydactyly of a triphalangeal thumb, unilateral
93337	Polydactyly of an index finger
295154	Polydactyly of an index finger, bilateral
295152	Polydactyly of an index finger, unilateral
295146	Polydactyly of the thumb, bilateral
295144	Polydactyly of the thumb, unilateral
2916	Polydactyly postaxial - dental and vertebral anomalies
2919	Polydactyly postaxial with median cleft of upper lip
2917	Polydactyly-myopia syndrome
180229	Polyembryoma
251	Polyepiphyseal dysplasia
93308	Polyepiphyseal dysplasia type 1
93307	Polyepiphyseal dysplasia type 4
93311	Polyepiphyseal dysplasia type 5
180182	Polymastia
35981	Polymicrogyria
2925	Polymicrogyria - turricephaly - hypogenitalism
300573	Polymicrogyria due to TUBB2B mutation
250972	Polymicrogyria with optic nerve hypoplasia
64745	Polymorphic eruption of pregnancy
732	Polymyositis
2905	Polyneuropathy - endocrinopathy - plasma cell dyscrasia
2926	Polyneuropathy - hand defect
171848	Polyneuropathy - hearing loss - ataxia - retinitis pigmentosa - cataract
2928	Polyneuropathy - intellectual deficit - acromicria - premature menopause
639	Polyneuropathy associated with IgM monoclonal gammopathy with anti-MAG
93276	Polyostotic fibrous dysplasia
96321	Polyploidy
160148	Polypoid prolapsing folds
208981	Polyradiculoneuropathy associated with IgG/IgA/IgM monoclonal gammopathy without known antibodies
141091	Polyrrhinia
93338	Polysyndactyly
2934	Polysyndactyly - cardiac malformation
295161	Polysyndactyly, bilateral
93405	Polysyndactyly, Haas type
295159	Polysyndactyly, unilateral
228410	Polyvalvular heart disease syndrome
11	Poly-X
139426	POMA
71526	POMC deficiency
365	Pompe disease
308604	Pompe disease, adult onset
308552	Pompe disease, infantile onset
308573	Pompe disease, juvenile onset
99748	Pontiac fever
269229	Pontine tegmental cap dysplasia
284339	Pontocerebellar hypoplasia - 46,XY disorder of sex development
324569	Pontocerebellar hypoplasia due to CHMP1A mutation
2254	Pontocerebellar hypoplasia type 1
2524	Pontocerebellar hypoplasia type 2
97249	Pontocerebellar hypoplasia type 3
166063	Pontocerebellar hypoplasia type 4
166068	Pontocerebellar hypoplasia type 5
166073	Pontocerebellar hypoplasia type 6
284339	Pontocerebellar hypoplasia type 7
324569	Pontocerebellar hypoplasia type 8
213777	Poorly differentiated endocrine carcinoma of the cervix uteri
213731	Poorly differentiated endocrine carcinoma of the corpus uteri
213731	Poorly differentiated endocrine carcinoma of the endometrium
213777	Poorly differentiated endocrine cervical carcinoma
284400	Poorly differentiated neuroendocrine carcinoma of the bladder
263339	Poorly-differentiated thymic neuroendocrine carcinoma

*Caution: one same acronym may correspond to different diseases in medical terms. Please refer to the full name of the disease to get the correct Orpha code.

ORPHA Number	Disease name
1300	Popliteal web syndrome
95699	POR deficiency
666	Porak and Durante disease
95699	PORD
2940	Porencephaly
2941	Porencephaly - cerebellar hypoplasia - internal malformations
306547	Porencephaly-microcephaly-bilateral congenital cataract syndrome
735	Porokeratosis of Mibelli
737	Porokeratosis plantaris palmaris et disseminata
166286	Porokeratotic eccrine nevus
166286	Porokeratotic eccrine ostial and dermal duct nevus
101330	Porphyria cutanea tarda
100924	Porphyria due to ALA dehydratase deficiency
100924	Porphyria due to ALAD deficiency
100924	Porphyria due to delta-aminolevulinate dehydratase deficiency
100924	Porphyria of Doss
79473	Porphyria variegata
854	Portal hypertension due to infrahepatic block
854	Portal vein thrombosis
2703	Port-wine nevi - mega cisterna magna - hydrocephalus
624	Port-wine stains
137839	Postanginal sepsis secondary to orophyngeal infection
246	Postaxial acrodysostosis
246	Postaxial acrofacial dysostosis
2920	Postaxial polydactyly - intellectual deficit
295008	Postaxial polydactyly of foot
295008	Postaxial polydactyly of toes
295181	Postaxial polydactyly of toes, bilateral
295179	Postaxial polydactyly of toes, unilateral
93334	Postaxial polydactyly type A
295165	Postaxial polydactyly type A, bilateral
295163	Postaxial polydactyly type A, unilateral
93335	Postaxial polydactyly type B
295169	Postaxial polydactyly type B, bilateral
295167	Postaxial polydactyly type B, unilateral
93406	Postaxial syndactyly with metacarpal synostosis
2730	Postaxial tetramelic oligodactyly
263352	Postcardiotomy right ventricular failure
97349	Postencephalitic parkinsonism
98971	Posterior amorphous corneal dystrophy
98971	Posterior amorphous stromal dystrophy
99141	Posterior choanal atresia - lymphedema syndrome
88628	Posterior column ataxia - retinitis pigmentosa
54247	Posterior cortical atrophy
99662	Posterior fossa tumors
2064	Posterior fusion of lumbosacral vertebrae - blepharoptosis
95706	Posterior hypospadias
268810	Posterior meningocele
98993	Posterior polar cataract
98973	Posterior polymorphous corneal dystrophy
98973	Posterior polymorphous dystrophy
98993	Posterior subcapsular cataract
93110	Posterior urethral valve
216452	Postlingual nonsyndromic genetic deafness
279947	Postorgasmic illness syndrome
563	Postpartum cardiomyopathy
2942	Post-polio sequelae
2942	Post-polio syndrome
2942	Post-poliomyelitic syndrome
2942	Post-poliomyelitis sequelae
2942	Post-poliomyelitis syndrome
98913	Postsynaptic congenital myasthenic syndromes
163921	Post-transplant acute limbic encephalitis
70568	Post-transplant lymphoproliferative disease
99859	Post-traumatic syringomyelia
238606	POT
612	Potassium-aggravated myotonia
640	Potato-grubbing palsy
1713	Potocki-Lupski syndrome
52022	Potocki-Shaffer syndrome
3316	Potter sequence - cleft lip/palate - cardiopathy
217067	Pouchitis
2876	Powell-Chandra-Saal syndrome
2201	Powell-Venencie-Gordon syndrome
95432	PPA
314566	PPAOS
284343	PPB family tumor susceptibility syndrome
284343	PPBFTDS
300324	PPBL
168829	PPC
98973	PPCD
93339	PPD1
93336	PPD2
93337	PPD3
93338	PPD4
494	PPK mutilans and deafness
79141	PPK nummularis
86923	PPK, Gamborg-Nielsen type
140966	PPK, Nagashima type
1010	PPK-CA, Stevanovic type
1366	PPK-CA, Wallis type
2202	PPK-deafness syndrome
79501	PPKP1
79502	PPKP2
38	PPKP3
308013	PPKP3 without elastoidosis
3077	PPM-X
189439	PPNAD
97278	PPoma
163927	PPP
308013	PPPK3 without elastoidosis
79502	PPPP
251295	PPRCA
739	Prader-Labhart-Willi syndrome
3409	Prader-Willi habitus - osteopenia - camptodactyly
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
177904	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2
177907	Prader-Willi syndrome due to translocation
171829	Prader-Willi-like syndrome due to deletion 6q16
2956	Prata-Liberal-Goncalves syndrome
245	Preaxial acrodysostosis
2957	Preaxial deficiency - postaxial polydactyly - hypospadias
2921	Preaxial polydactyly - colobomata - intellectual deficit
295006	Preaxial polydactyly of foot
295006	Preaxial polydactyly of toes
295177	Preaxial polydactyly of toes, bilateral
295175	Preaxial polydactyly of toes, unilateral
93339	Preaxial polydactyly type 1
295146	Preaxial polydactyly type 1, bilateral
295144	Preaxial polydactyly type 1, unilateral
93336	Preaxial polydactyly type 2
295150	Preaxial polydactyly type 2, bilateral
295148	Preaxial polydactyly type 2, unilateral
93337	Preaxial polydactyly type 3
295154	Preaxial polydactyly type 3, bilateral
295152	Preaxial polydactyly type 3, unilateral
93338	Preaxial polydactyly type 4
295161	Preaxial polydactyly type 4, bilateral

ORPHA Number	Disease name
295159	Preaxial polydactyly type 4, unilateral
1309	Precalicial canalicular ectasia
99860	Precursor B-cell acute lymphoblastic leukemia
99860	Precursor B-cell acute lymphoblastic leukemia/lymphoma
99860	Precursor B-cell acute lymphocytic leukemia
99860	Precursor B-cell acute lymphocytic leukemia/lymphoma
99861	Precursor T-cell acute lymphoblastic leukemia
99861	Precursor T-cell acute lymphoblastic leukemia/lymphoma
99861	Precursor T-cell acute lymphocytic leukemia
99861	Precursor T-cell acute lymphocytic leukemia/lymphoma
293462	Pre-Descemet corneal dystrophy
275555	Pre-eclampsia
2860	Preeyasombat-Varavithya syndrome
69665	Pregnancy-related cholestasis
216445	Prelingual nonsyndromic genetic deafness
276432	Premature ageing appearance-developmental delay-cardiac arrhythmia syndrome
52183	Premature chromosome condensation with microcephaly and intellectual deficit
95486	Premature closure of the arterial duct
95486	Premature closure of the patent ductus arteriosus
2114	Premature degenerative osteoarthropathy of the hip
247638	Prenatal benign hypophosphatasia
247638	Prenatal benign phosphoethanolaminuria
247638	Prenatal benign Rathburn disease
90160	Pressure-induced localized lipoatrophy
98914	Presynaptic congenital myasthenic syndromes
79410	Pretibial DEB
79410	Pretibial dystrophic epidermolysis bullosa
2958	Prieto-Badia-Mulas syndrome
1451	Prieur-Griscelli syndrome
930	Primary achalasia
75564	Primary acquired sideroblastic anemia
85138	Primary Addison's disease
85443	Primary amyloidosis
228272	Primary anetoderma
140989	Primary angiitis of the central nervous system
1572	Primary antibody deficiency
2285	Primary basilar impression
186	Primary biliary cirrhosis
93955	Primary blepharospasm
314684	Primary bone lymphoma
46135	Primary brain lymphoma
267	Primary calpainopathy
169464	Primary CD59 deficiency
46135	Primary central nervous system lymphoma
244	Primary ciliary dyskinesia
247522	Primary ciliary dyskinesia - retinitis pigmentosa
46135	Primary CNS lymphoma
90042	Primary congenital erythrocytosis
91138	Primary cryoglobulinemia
178528	Primary cutaneous aggressive epidermotropic CD8+ T-cell lymphoma
137807	Primary cutaneous amyloidosis
300865	Primary cutaneous anaplastic large cell lymphoma
541	Primary cutaneous CD30+ T-cell lymphoproliferative disease
178522	Primary cutaneous CD4+ small/medium-sized pleomorphic T-cell lymphoma
178544	Primary cutaneous diffuse large B-cell lymphoma, leg type
178540	Primary cutaneous follicle center lymphoma
178533	Primary cutaneous gamma/delta-positive T-cell lymphoma
541	Primary cutaneous Ki-1+ T-cell lymphoproliferative disease
178536	Primary cutaneous marginal zone B-cell lymphoma
86885	Primary cutaneous unspecified peripheral T-cell lymphoma
671	Primary cutis verticis gyrata
98807	Primary dystonia with mixed phenotype
99657	Primary dystonia, DYT2 type
98805	Primary dystonia, DYT4 type
98806	Primary dystonia, DYT6 type
98807	Primary dystonia, DYT13 type
306734	Primary dystonia, DYT21 type
48686	Primary effusion lymphoma
90026	Primary erythermalgia
357220	Primary essential cutis verticis gyrata
98957	Primary familial amyloidosis of the cornea
90042	Primary familial and congenital polycythemia
90042	Primary familial polycythemia
3337	Primary Fanconi renotubular syndrome
3337	Primary Fanconi syndrome
633	Primary GH insensitivity
633	Primary GH resistance
633	Primary growth hormone insensitivity
633	Primary growth hormone resistance
314950	Primary HES
314950	Primary hypereosinophilic syndrome
2232	Primary hypergonadotropic hypogonadism - partial alopecia
682	Primary hyperkalemic periodic paralysis
416	Primary hyperoxaluria
93598	Primary hyperoxaluria type 1
93599	Primary hyperoxaluria type 2
93600	Primary hyperoxaluria type 3
682	Primary hyperPP
33208	Primary hypersomnia
1572	Primary hypogammaglobulinemia
100049	Primary ILD specific to childhood due to pulmonary surfactant protein anomalies
90023	Primary immunodeficiency syndrome due to p14 deficiency
90023	Primary immunodeficiency syndrome with short stature
73272	Primary insulin-like growth factor deficiency
100049	Primary interstitial lung disease specific to childhood due to pulmonary surfactant protein anomalies
90362	Primary intestinal lymphangiectasia
279904	Primary intraocular lymphoma
279904	Primary intraocular non-Hodgkin's lymphoma
140436	Primary intraosseous vascular malformation
137926	Primary laryngeal lymphangioma
35689	Primary lateral sclerosis
314709	Primary localized amyloidosis
137807	Primary localized cutaneous amyloidosis
137810	Primary localized cutaneous nodular amyloidosis
319667	Primary lymphoid conjunctival tumor
319667	Primary lymphoma of the conjunctiva
228272	Primary macular atrophy
168811	Primary malignant peritoneal mesothelioma
98838	Primary mediastinal clear cell lymphoma of B-cell type
98838	Primary mediastinal large B-cell lymphoma
238642	Primary megaureter, adult-onset form
252050	Primary melanoma of the central nervous system
54370	Primary membranoproliferative glomerulonephritis
306558	Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome
824	Primary myelofibrosis
357225	Primary non-essential cutis verticis gyrata
279897	Primary oculocerebral lymphoma
279897	Primary oculocerebral non-Hodgkin's lymphoma
238606	Primary orthostatic tremor
99878	Primary parathyroids hyperplasia
168829	Primary peritoneal carcinoma
168829	Primary peritoneal serous carcinoma

ORPHA Number	Disease name
189439	Primary pigmented nodular adrenocortical disease
100021	Primary plasmacytoma of the bone
95432	Primary progressive aphasia
314566	Primary progressive apraxia of speech
75567	Primary progressive freezing gait
2420	Primary pulmonary lymphoma
314822	Primary renal tubular acidosis
358	Primary renal tubular hypokalemic hypomagnesemia with hypocalciuria
171	Primary sclerosing cholangitis
99856	Primary syringomyelia
268871	Primary syringomyelia/hydromyelia
314701	Primary systemic amyloidosis
158	Primary systemic carnitine deficiency
2284	Primary T cell immunodeficiency
268861	Primary tethered chord syndrome
268861	Primary tethered spinal cord syndrome
99867	Primary thymic epithelial neoplasm
263310	Primary thymic epithelial neoplasm type A
263324	Primary thymic epithelial neoplasm type AB
263317	Primary thymic epithelial neoplasm type B
99867	Primary thymic epithelial tumor
263310	Primary thymic epithelial tumor type A
263324	Primary thymic epithelial tumor type AB
263317	Primary thymic epithelial tumor type B
98807	Primary torsion dystonia with predominant craniocervical or upper limb onset
231580	Primary unilateral adrenal hyperplasia
140989	Primary vasculitis of the central nervous system
2542	Primitive anophthalmia
2636	Primordial microcephalic dwarfism, Crachami type
46658	Primordial short stature - microdontia - opalescent and rootless teeth
3042	Primrose syndrome
56970	Prion disease
2965	PRL-secreting pituitary adenoma
326	Proaccelerin deficiency
141099	Proboscis lateralis
740	Progeria
2959	Progeria - short stature - pigmented nevi
99706	Progeria-associated arthropathy
300382	Progeroid and marfanoid aspect-lipodystrophy syndrome
2962	Progeroid syndrome, De Barsy type
2963	Progeroid syndrome, Petty type
316	Progressiva symmetrica erythrokeratodermia
79094	Progressive arterial occlusive disease - hypertension - heart defects - bone fragility - brachysyndactyly
75373	Progressive bifocal chorioretinal atrophy
139447	Progressive cavitating leukoencephalopathy
79087	Progressive cephalothoracic lipodystrophy
247198	Progressive cerebello-cerebral atrophy
1871	Progressive cone dystrophy
220393	Progressive cutaneous systemic scleroderma
220393	Progressive cutaneous systemic sclerosis
3235	Progressive deafness with stapes fixation
216812	Progressive deforming osteogenesis imperfecta
217396	Progressive demyelinating neuropathy with bilateral striatal necrosis
1328	Progressive diaphyseal dysplasia
495	Progressive diffuse palmoplantar keratoderma
495	Progressive diffuse PPK
2836	Progressive encephalopathy - optic atrophy
2836	Progressive encephalopathy with edema, hypsarrhythmia and optic atrophy
1947	Progressive epilepsy - intellectual deficit, Finnish type
352447	Progressive external ophthalmoplegia - myopathy - emaciation
2744	Progressive external ophthalmoplegia and scoliosis
172	Progressive familial intrahepatic cholestasis
79306	Progressive familial intrahepatic cholestasis type 1
79304	Progressive familial intrahepatic cholestasis type 2
79305	Progressive familial intrahepatic cholestasis type 3
75327	Progressive foveal dystrophy
1214	Progressive hemifacial atrophy
73	Progressive massive osteolysis
217260	Progressive multifocal leukoencephalitis
217260	Progressive multifocal leukoencephalopathy
263516	Progressive myoclonic epilepsy due to KCTD7 deficiency
308	Progressive myoclonic epilepsy type 1
501	Progressive myoclonic epilepsy type 2
263516	Progressive myoclonic epilepsy type 3
280620	Progressive myoclonic epilepsy type 6
352596	Progressive myoclonic epilepsy with dystonia
726	Progressive neuronal degeneration of childhood with liver disease
228012	Progressive neurosensory deafness - hypertrophic cardiomyopathy
228012	Progressive neurosensory hearing loss - hypertrophic cardiomyopathy
158022	Progressive nodular histiocytosis
100070	Progressive non-fluent aphasia
2062	Progressive non-infectious anterior vertebral fusion
2762	Progressive osseous heteroplasia
3322	Progressive pancytopenia - immunodeficiency - cerebellar hypoplasia
1159	Progressive pseudorheumatoid arthropathy of childhood
352718	Progressive retinal dystrophy due to retinol transport defect
228012	Progressive sensorineural deafness - hypertrophic cardiomyopathy
228012	Progressive sensorineural hearing loss - hypertrophic cardiomyopathy
240112	Progressive supranuclear palsy - apraxia of speech
240103	Progressive supranuclear palsy - corticobasal syndrome
240085	Progressive supranuclear palsy - parkinsonism
240112	Progressive supranuclear palsy - progressive non fluent aphasia
240094	Progressive supranuclear palsy - pure akinesia with gait freezing
316	Progressive symmetric erythrokeratodermia
316	Progressive symmetric erythrokeratodermia, Gottron type
2965	Prolactinoma
2965	Prolactin-secreting pituitary adenoma
742	Prolidase deficiency
492	Proliferating trichilemmal cyst
86872	Proliferation of large granular lymphocytes
221126	Proliferative vasculopathy and hydranencephaly/hydrocephaly
419	Proline oxidase deficiency
75374	Prolonged electroretinal response supression
300878	Prolymphocytic variant of hairy cell leukemia
300878	Prolymphocytic variant of HCL
2083	Prominent glabella - microcephaly - hypogenitalism
2966	Properdin deficiency
35	Propionic acidemia
35	Propionyl-CoA carboxylase deficiency
324977	Proteasome disability syndrome
213	Protein defect of cystin transport
2967	Protein R deficiency
26349	Protein S acquired deficiency
744	Proteus syndrome
2969	Proteus-like syndrome
325	Prothrombin deficiency

ORPHA Number	Disease name
251598	Protoplasmic astrocytoma
79473	Protoporphyrinogen oxidase deficiency
2508	Proud-Levine-Carpenter syndrome
52022	Proximal 11p deletion syndrome
261197	Proximal 16p11.2 microdeletion syndrome
261197	Proximal del(16)(p11.2)
2019	Proximal focal femoral deficiency
261197	Proximal monosomy 16p11.2
606	Proximal myotonic dystrophy
606	Proximal myotonic myopathy
3269	Proximal radioulnar synostosis
47159	Proximal renal tubular acidosis
93607	Proximal renal tubular acidosis with ocular abnormalities and intellectual deficit
70	Proximal spinal muscular atrophy
83330	Proximal spinal muscular atrophy type 1
83418	Proximal spinal muscular atrophy type 2
83419	Proximal spinal muscular atrophy type 3
83420	Proximal spinal muscular atrophy type 4
3250	Proximal symphalangism
3390	Proximal tubulopathy - diabetes mellitus - cerebellar ataxia
47159	pRTA
2970	Prune belly syndrome
89843	Pruriginous dystrophic epidermolysis bullosa
64745	Pruritic urticarial papules and plaques of pregnancy
284417	PSAT deficiency
171	PSC
750	Pseudoachondroplasia
750	Pseudoachondroplastic dysplasia
750	Pseudoachondroplastic spondyloepiphyseal dysplasia
2971	Pseudoadrenoleukodystrophy
526	Pseudoaldosteronism
221120	Pseudoaminopterin syndrome
228402	Pseudo-Angelman syndrome
314459	Pseudo-Demons-Meigs syndrome
85174	Pseudodiastrophic dysplasia
2983	Pseudohermaphroditism - intellectual deficit
577	Pseudo-Hurler polydystrophy
526	Pseudohyperaldosteronism type 1
88660	Pseudohyperaldosteronism type 2
756	Pseudohypoaldosteronism type 1
757	Pseudohypoaldosteronism type 2
88938	Pseudohypoaldosteronism type 2A
88939	Pseudohypoaldosteronism type 2B
88940	Pseudohypoaldosteronism type 2C
300525	Pseudohypoaldosteronism type 2D
300530	Pseudohypoaldosteronism type 2E
79443	Pseudohypoparathyroidism type 1A
94089	Pseudohypoparathyroidism type 1B
79444	Pseudohypoparathyroidism type 1C
94090	Pseudohypoparathyroidism type 2
2976	Pseudoleprechaunism syndrome, Patterson type
314459	Pseudo-Meigs syndrome
263482	Pseudo-Morquio syndrome type 2
26790	Pseudomyxoma peritonei
2971	Pseudo-NALD
2971	Pseudo-neonatal adrenoleukodystrophy
251962	Pseudopapillary ganglioglioneurocytoma
251962	Pseudopapillary neurocytoma with glial differentiation
2980	Pseudopapilledema - blepharophimosis - hand anomalies
129	Pseudo-pelade of Brocq
2985	Pseudoprogeria syndrome
79445	Pseudopseudohypoparathyroidism
3103	Pseudothalidomide syndrome
1229	Pseudo-TORCH syndrome
2518	Pseudotoxoplasmosis syndrome
2166	Pseudo-trisomy 13 syndrome
238624	Pseudotumor cerebri
83316	Pseudotyphus of California
180079	Pseudo-unicornuate uterus
289157	Pseudovitamin D-deficient rickets
52530	Pseudo-Von Willebrand disease
52530	Pseudo-Von Willebrand disease type 2B
758	Pseudoxanthoma elasticum
228293	Pseudoxanthoma elasticum-like papillary dermal elastocytosis
91135	Pseudoxanthoma elasticum-like syndrome
228227	Pseudoxanthoma-like late-onset focal dermal elastosis
280794	Pseudoxanthomatous DCM
280794	Pseudoxanthomatous diffuse cutaneous mastocytosis
2981	Pseudo-Zellweger syndrome
95496	PSIS
240112	PSP-AOS
240103	PSP-CBS
240103	PSP-corticobasal syndrome
240085	PSP-p
240094	PSP-PAGF
240085	PSP-parkinsonism
240112	PSP-PNFA
240094	PSP-pure akinesia with gait freezing
99928	PSST
71519	Psychogenic dystonia
71519	Psychogenic movement disorders
324636	Psychogenic purpura
88618	Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency
329	PTA deficiency
247698	PTC syndrome
269229	PTCD
97290	PTC-RCC
2988	Pterygium colli - intellectual deficit - digital anomalies
2989	Pterygium of the conjunctiva, familial form
86789	PTLAH
70568	PTLD
2999	Ptosis - strabismus - ectopic pupils
238766	Ptosis - syndactyly - learning difficulties
228396	Ptosis - upper ocular movement limitation - absence of lacrimal punctum
2997	Ptosis - vocal cord paralysis
52530	PT-VWD
231580	PUAH
2038	Pulmonar arteriovenous aneurysm
984	Pulmonary agenesis
60025	Pulmonary alveolar microlithiasis
247257	Pulmonary anthrax
1137	Pulmonary aortic stenosis obstructive uropathy
2038	Pulmonary arteriovenous fistula
99049	Pulmonary artery coming from patent ductus arteriosus
99050	Pulmonary artery coming from the aorta
99083	Pulmonary artery hypoplasia
1208	Pulmonary atresia - intact ventricular septum
1207	Pulmonary atresia with ventricular septal defect
64741	Pulmonary blastoma
99084	Pulmonary branch stenosis
199241	Pulmonary capillary hemangiomatosis
210136	Pulmonary fibrosis - hepatic hyperplasia - bone marrow hypoplasia
217080	Pulmonary fungal infections in patients deemed at risk
99874	Pulmonary histiocytosis X
991	Pulmonary hypoplasia - agonadism - dextrocardia - diaphragmatic hernia syndrome
217557	Pulmonary interstitial glycogenosis
2414	Pulmonary lymphangiomatosis
60026	Pulmonary nodular lymphoid hyperplasia
60026	Pulmonary pseudolymphoma
982	Pulmonary valve agenesis
101206	Pulmonary valve agenesis - Fallot's tetralogy - absence of ductus arteriosus

ORPHA Number	Disease name
99048	Pulmonary valve agenesis - ventricular septal defect - persistent ductus arteriosus
31837	Pulmonary venoocclusive disease
85202	Pulmonic stenosis - brachytelephalangism - calcification of cartilages
3444	Pulmonic stenosis with 'cafe-au-lait' spots
98984	Pulverulent cataract
97353	Punch-drunk syndrome
99710	Punctate acrokeratoderma freckle like pigmentation
79502	Punctate palmoplantar hyperkeratosis type 2
38	Punctate palmoplantar hyperkeratosis type 3
308013	Punctate palmoplantar hyperkeratosis type 3 without elastoidosis
79501	Punctate palmoplantar keratoderma type 1
79502	Punctate palmoplantar keratoderma type 2
38	Punctate palmoplantar keratoderma type 3
308013	Punctate palmoplantar keratoderma type 3 without elastoidosis
231625	Pure aldosterone-producing adrenocortical carcinoma
231625	Pure aldosterone-secreting adrenocortical carcinoma
231625	Pure APAC
441	Pure autonomic failure
94148	Pure cerebellar syndrome - mild pyramidal signs
441	Pure dysautonomia
319465	Pure familial acute myeloid leukemia
319465	Pure familial AML
69084	Pure hair and nail ectodermal dysplasia
441	Pure idiopatic dysautonomia
475	Pure Joubert syndrome
254854	Pure mitochondrial myopathy
2028	Puretic syndrome
760	Purine nucleoside phosphorylase deficiency
49566	Purpura fulminans
761	Purpura rheumatica
2442	Purtilo syndrome
293190	PUS
293173	Pustular drug eruption
163927	Pustulosis palmaris et plantaris
48377	Pustulosis subcornealis
729	PV
982	PVA
101206	PVA/ADA, Fallot type
99048	PVA/PDA, non-Fallot type
251607	PXA
758	PXE
228227	PXE-like late-onset focal dermal elastosis
228293	PXE-like papillary dermal elastocytosis
91135	PXE-like syndrome
763	Pycnodysostosis
293633	PYCR1 deficiency
293633	PYCR1-related DeBarsy syndrome
3003	Pyknoachondrogenesis
763	Pyknodysostosis
64280	Pyknolepsy
3005	Pyle disease
48104	Pyoderma gangrenosum
289478	Pyoderma gangrenosum - acne - suppurative hidradenitis
69126	Pyogenic arthritis - pyoderma gangrenosum - acne
183713	Pyogenic bacterial infections due to MyD88 deficiency
764	Pyomyositis
2561	Pyramidal molar - glaucoma - upper abnormal lip
79096	Pyridoxal phosphate-responsive seizures
79096	Pyridoxamine 5'-oxidase deficiency
79096	Pyridoxamine 5'-phosphate oxidase deficiency
79096	Pyridoxa-phosphate dependent seizures
3006	Pyridoxine-dependent epilepsy
3006	Pyridoxine-responsive seizures
75563	Pyridoxine-responsive sideroblastic anemia
32	Pyroglutamicaciduria
293633	Pyrroline-5-carboxylate reductase 1 deficiency
3008	Pyruvate carboxylase deficiency
353308	Pyruvate carboxylase deficiency type A
353314	Pyruvate carboxylase deficiency type B
353320	Pyruvate carboxylase deficiency type C
353320	Pyruvate carboxylase deficiency, benign type
353308	Pyruvate carboxylase deficiency, infantile type
353314	Pyruvate carboxylase deficiency, severe neonatal type
79243	Pyruvate decarboxylase deficiency
79244	Pyruvate dehydrogenase complex component E2 deficiency
255182	Pyruvate dehydrogenase complex component E3 deficiency
765	Pyruvate dehydrogenase complex deficiency
79243	Pyruvate dehydrogenase complex E1 component subunit alpha deficiency
255138	Pyruvate dehydrogenase complex E1 component subunit beta deficiency
765	Pyruvate dehydrogenase deficiency
79243	Pyruvate dehydrogenase E1-alpha deficiency
255138	Pyruvate dehydrogenase E1-beta deficiency
79244	Pyruvate dehydrogenase E2 deficiency
2394	Pyruvate dehydrogenase E3 deficiency
255182	Pyruvate dehydrogenase E3-binding protein deficiency
79246	Pyruvate dehydrogenase phosphatase deficiency
255182	Pyruvate dehydrogenase protein X component deficiency
766	Pyruvate kinase deficiency of erythrocytes
781	Q fever
3010	Qazi-Markouizos syndrome
37553	QT long syndrome type 7
602	Quadriceps-sparing myopathy
9	Quadruple X
84142	Quantal squander syndrome
869	Quaternary A syndrome
220436	Quebec platelet disorder
137888	Question mark ear syndrome
346	Quinquaud's folliculitis decalvans
261529	r(Y)
100057	RAAS-blocker-induced angioedema
100057	RAAS-blocker-induced angioneurotic edema
770	Rabies
769	Rabson-Mendenhall syndrome
240760	RAD50 deficiency
93321	Radial clubhand
1121	Radial deficiency - tibial hypoplasia
93321	Radial hemimelia
295071	Radial hemimelia, bilateral
295069	Radial hemimelia, unilateral
2252	Radial hypoplasia - triphalangeal thumbs - hypospadias - maxillary diastema
295071	Radial longitidinal meromelia, bilateral
295069	Radial longitidinal meromelia, unilateral
93321	Radial ray agenesis
3026	Radial ray hypoplasia - choanal atresia
90021	Radiation myelitis
70475	Radiation proctitis
99789	Radicular dentin dysplasia
3015	Radio-renal syndrome
295219	Radio-ulnar fusion, bilateral
295217	Radio-ulnar fusion, unilateral
71289	Radio-ulnar synostosis - amegakaryocytic thrombocytopenia
3270	Radio-ulnar synostosis - intellectual deficit - hypotonia
295219	Radio-ulnar synostosis, bilateral
295217	Radio-ulnar synostosis, unilateral

ORPHA Number	Disease name
294979	Radio-ulnar terminal transverse meromelia
295095	Radio-ulnar terminal transverse meromelia, bilateral
295093	Radio-ulnar terminal transverse meromelia, unilateral
3016	Radius absent - anogenital anomalies
100057	RAE
100019	RAEB-1
100020	RAEB-2
168960	RAEB-t
1832	Raine syndrome
50811	Rajab-Spranger syndrome
268114	RALD
99843	Rambam-Hasharon syndrome
3018	Rambaud-Gallian syndrome
3018	Rambaud-Gallian-Touchard syndrome
3019	Ramon syndrome
1051	Ramos-Arroyo syndrome
3020	Ramsay-Hunt syndrome
86861	Randall disease
3021	RAPADILINO syndrome
141184	Rapidly involuting congenital hemangioma
280569	Rapidly progressive glomerulonephritis
293987	Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome
293987	Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation-neural tumors
71517	Rapid-onset dystonia-parkinsonism
178307	RAPK
213528	Rare adenocarcinoma of the breast
213574	Rare adenocarcinoma of the corpus uteri
97293	Rare benign ovarian tumor
213767	Rare cervical squamous cell carcinoma
68361	Rare deafness
137820	Rare endometriosis
98619	Rare isolated myopia
213767	Rare squamous cell carcinoma of the cervix uteri
75564	RARS
268114	RAS-associated autoimmune lymphoproliferative disease
1929	Rasmussen subacute encephalitis
1929	Rasmussen syndrome
3023	Rasmussen-Johnsen-Thomsen syndrome
31205	Rat-bite fever
436	Rathburn disease
99852	RAVINE syndrome
2840	Ray-Peterson-Scott syndrome
98961	RBCD
79127	RB-ILD
93111	RCAD syndrome
218432	RCM3
284388	RCVS
79408	RDEB generalisata gravis
89842	RDEB generalisata mitis
89841	RDEB, centripetalis
79408	RDEB, Hallopeau-Siemens type
89842	RDEB, non-Hallopeau-Siemens type
89841	RDEB-Ce
89842	RDEB-generalized other
79409	RDEB-I
89842	RDEB-O
79408	RDEB-sev gen
85445	Reactive amyloidosis
29207	Reactive arthritis
314962	Reactive hypereosinophilic syndrome
166433	Reading seizures
1188	Reardon-Baraitser syndrome
2631	Reardon-Hall-Slaney syndrome
96167	Rec(8) syndrome
96167	Rec8 syndrome
1115	Recessive aplasia cutis congenita of limbs
139485	Recessive cerebellar ataxia-2
139373	Recessive congenital methemoglobinemia type 1
139380	Recessive congenital methemoglobinemia type 2
79409	Recessive dystrophic epidermolysis bullosa inversa
89842	Recessive dystrophic epidermolysis bullosa, non-Hallopeau-Siemens type
89842	Recessive dystrophic epidermolysis bullosa-generalized other
139373	Recessive hereditary methemoglobinemia type 1
139380	Recessive hereditary methemoglobinemia type 2
280384	Recessive intellectual disability - motor dysfunction - multiple joint contractures
94125	Recessive mitochondrial ataxic syndrome
461	Recessive X-linked ichthyosis
96167	Recombinant 8 syndrome
96167	Recombinant chromosome 8 syndrome
171220	Rectal duplication
100081	Rectal endocrine tumor
51890	Rectus abdominis syndrome
88619	Recurrent acute necrotizing encephalopathy
64740	Recurrent acute pancreatitis
2672	Recurrent encephalophathy of childhood
90052	Recurrent hepatitis C virus induced liver disease in liver transplant recipients
293381	Recurrent hereditary corneal erosions
169142	Recurrent infection due to specific granule deficiency
251523	Recurrent infections - inflammatory syndrome due to zinc metabolism disorder
183675	Recurrent infections associated with rare immunoglobulin isotypes deficiency
69665	Recurrent intrahepatic cholestasis of pregnancy
169467	Recurrent Neisseria infections due to factor D deficiency
60032	Recurrent respiratory papillomatosis
199267	Recurring digital fibrous tumor of childhood
79433	Red oculocutaneous albinism
231031	Red palms disease
838	RED-M
97239	Reducing body myopathy
3221	Refetoff syndrome
310	Reflex epilepsy
99995	Reflex sympathetic dystrophy
98826	Refractory anemia
86839	Refractory anemia with excess blasts
168960	Refractory anemia with excess blasts in transformation
100019	Refractory anemia with excess blasts-1
100020	Refractory anemia with excess blasts-2
75564	Refractory anemia with ringed sideroblasts
86836	Refractory cytopenia with multilineage dysplasia
773	Refsum disease
1525	Reginato-Schiapachasse syndrome
83450	Regional odontodysplasia
1040	Regressive metaphyseal dysplasia
2634	Reinhardt-Pfeiffer mesomelic dysplasia
2634	Reinhardt-Pfeiffer syndrome
98961	Reis-Bücklers corneal dystrophy
29207	Reiter syndrome
99991	Relapsing epidemic thyphus
33577	Relapsing febrile nodular nonsuppurative panniculitis
33577	Relapsing febrile nodular panniculitis
91547	Relapsing fever
728	Relapsing polychondritis
2838	Renal caliceal diverticuli - deafness
319314	Renal cell carcinoma after neuroblastoma
319314	Renal cell carcinoma associated with neuroblastoma
1475	Renal coloboma syndrome
93111	Renal cysts - maturity-onset diabetes of the young
93111	Renal cysts and diabetes syndrome
93111	Renal dysfunction - early-onset diabetes
93108	Renal dysplasia

ORPHA Number	Disease name
3404	Renal dysplasia - limb defects
1850	Renal dysplasia - megalocystis - sirenomelia
3404	Renal dysplasia - mesomelia - radiohumeral fusion
3156	Renal dysplasia - retinal aplasia
140969	Renal dysplasia - retinal pigmentary dystrophy - cerebellar ataxia - skeletal dysplasia
654	Renal embryonic tumor
1652	Renal Fanconi syndrome with nephrocalcinosis and renal stones
69076	Renal glucosuria
34528	Renal hypomagnesemia type 2
93101	Renal hypoplasia
319319	Renal medullary carcinoma
71273	Renal nutcracker syndrome
171871	Renal pseudohypoaldosteronism type 1
18	Renal tubular acidosis type 1
93608	Renal tubular acidosis type 1a
93611	Renal tubular acidosis type 1b
93609	Renal tubular acidosis type 1c
47159	Renal tubular acidosis type 2
2785	Renal tubular acidosis type 3
89939	Renal tubular acidosis type 4
3033	Renal tubular dysgenesis
97368	Renal tubular dysgenesis drugs-related
97367	Renal tubular dysgenesis due to twin-twin transfusion
97369	Renal tubular dysgenesis of genetic origin
254902	Renal tubulopathy - encephalopathy - liver failure
1092	Renal-genital-middle ear anomalies
294415	Renal-hepatic-pancreatic dysplasia
3032	Renal-hepatic-pancreatic dysplasia - Dandy-Walker cysts
217330	REN-associated familial juvenile hyperuricemic nephropathy
217330	REN-associated FJHN
217330	REN-associated kidney disease
774	Rendu-Osler disease
774	Rendu-Osler-Weber disease
93975	Renier-Gabreels-Jasper syndrome
100057	Renin-angiotensin-aldosterone system-blocker-induced angioedema
100057	Renin-angiotensin-aldosterone system-blocker-induced angioneurotic edema
294415	Renohepaticopancreatic dysplasia
3242	Renpenning syndrome
73273	Resistance to IGF-1
424	Resistance to thyroid stimulating hormone
99832	Resistance to thyrotropin-releasing hormone syndrome
247257	Respiratory anthrax
79127	Respiratory bronchiolitis - interstitial lung disease
33355	Reticular dysgenesis
99002	Reticular dystrophy of the retinal pigment epithelium
100000	Reticular perineurioma
79145	Reticular pigment anomaly of flexures
178307	Reticulate acropigmentation of Kitamura
86900	Reticulum cell sarcoma
284247	Retinal arterial macroaneurysm and supravalvular pulmonic stenosis
75326	Retinal arterial tortuosity
75326	Retinal arteriolar tortuosity
36383	Retinal arteriolar tortuosity - infantile hemiparesis - autosomal dominant leukoencephalopathy
71213	Retinal cavernous hemangioma
1574	Retinal degeneration - nanophthalmos - glaucoma
1571	Retinal detachment - occipital encephalocele
71862	Retinal dystrophy
75326	Retinal hemorrhage with vascular tortuosity
3018	Retinal ischemic syndrome - digestive tract small vessel hyalinosis - diffuse cerebral calcifications
319640	Retinal macular dystrophy type 2
353356	Retinal vasoproliferative tumor
791	Retinitis pigmentosa
886	Retinitis pigmentosa - deafness
140976	Retinitis pigmentosa - hypopituitarism - nephronophthisis - skeletal dysplasia
3085	Retinitis pigmentosa - intellectual deficit - deafness - hypogenitalism
261512	Retinitis pigmentosa and intellectual deficit due to del(X)(p11.3)
261512	Retinitis pigmentosa and intellectual deficit due to monosomy Xp11.3
261512	Retinitis pigmentosa and intellectual deficit due to Xp11.3 microdeletion
52427	Retinitis punctata albescens
790	Retinoblastoma
838	Retinocochleocerebral vasculopathy
3087	Retinohepatoendocrinologic syndrome
2305	Retinoic acid embryopathy
2305	Retinoids embryopathy
352718	Retinol dystrophy-iris coloboma-comedogenic acne syndrome
3088	Retinopathy - anemia- central nervous system anomalies
90050	Retinopathy of prematurity
139455	Retinopathy, Burgess-Black type
838	Retinopathy, encephalopathy, deafness associated microangiopathy
53540	Retinoschisis with early nyctalopia
269200	Retrocerebellar cyst
49041	Retroperitoneal fibrosis
778	Rett syndrome
3095	Rett syndrome variant
99852	Reunion island - anorexia - vomiting which is irrepressible - neurological signs
294049	Reunion island's Larsen syndrome
284388	Reversible cerebral vasoconstriction syndrome
254864	Reversible infantile cytochrome c oxidase deficiency
254864	Reversible infantile respiratory chain deficiency
3088	Revesz-DeBuse syndrome
199267	Reye tumor
3096	Reye's syndrome
779	Reynolds syndrome
244310	RFT1-CDG syndrome
251975	RGNT
71275	Rh deficiency syndrome
69077	Rhabdoid tumor
3097	Rhabdomyomatous dysplasia - cardiopathy - genital anomalies
780	Rhabdomyosarcoma
213802	Rhabdomyosarcoma of the cervix uteri
213615	Rhabdomyosarcoma of the corpus uteri
3099	Rheumatic fever
761	Rheumatoid purpura
177	Rhizomelic chondrodysplasia punctata
309789	Rhizomelic chondrodysplasia punctata type 1
309796	Rhizomelic chondrodysplasia punctata type 2
309803	Rhizomelic chondrodysplasia punctata type 3
2831	Rhizomelic dysplasia, Patterson-Lowry type
93569	Rhizomelic pseudopolyarthritis
3098	Rhizomelic syndrome
71275	Rh-null syndrome
59315	Rhombencephalosynapsis
140976	RHYNS syndrome
97229	Riboflavin transporter deficiency
141184	RICH
2323	Richadson-Kirk syndrome
240071	Richardson syndrome
1399	Richards-Rundle syndrome
3101	Richieri Costa-da Silva syndrome
2649	Richieri Costa-Guion Almeida syndrome
2511	Richieri Costa-Guion Almeida-Ramos syndrome
3102	Richieri Costa-Pereira syndrome

ORPHA Number	Disease name
1784	Richieri-Costa-Colletto syndrome
1794	Richieri-Costa-Gorlin syndrome
28378	Richner-Hanhart syndrome
606	Ricker disease
606	Ricker syndrome
83312	Rickettsialpox
217055	RI-CMT type A
254334	RI-CMT type B
64744	Riedel thyroiditis
91483	Rieger anomaly
3163	Rieger anomaly - partial lipodystrophy
782	Rieger syndrome
319251	Rift valley fever
99081	Right aortic arch
99119	Right inferior caval vein connecting to left-sided atrium
99119	Right inferior vena cava connecting to left-sided atrium
99119	Right IVC connecting to left-sided atrium
99110	Right superior caval vein connecting to left-sided atrium
99110	Right superior vena cava connecting to left-sided atrium
99110	Right SVC connecting to left-sided atrium
293848	Right temporal lobar atrophy
439	Right ventricular hypoplasia
97244	Rigid spine syndrome
1764	Riley-Day syndrome
217335	RIN2 deficiency
217335	RIN2 syndrome
1437	Ring chromosome 1
96171	Ring chromosome 2
96172	Ring chromosome 3
1447	Ring chromosome 4
251043	Ring chromosome 5
1448	Ring chromosome 6
1449	Ring chromosome 7
1450	Ring chromosome 8
96173	Ring chromosome 9
1438	Ring chromosome 10
96175	Ring chromosome 11
1439	Ring chromosome 12
96176	Ring chromosome 13
1440	Ring chromosome 14
96177	Ring chromosome 15
96178	Ring chromosome 16
1441	Ring chromosome 17
1442	Ring chromosome 18
1443	Ring chromosome 19
1444	Ring chromosome 20
1445	Ring chromosome 21
1446	Ring chromosome 22
261529	Ring chromosome Y
91481	Ring dermoid of cornea
91481	Ring dermoid syndrome
169	Ringed hair disease
97238	Rippling muscle disease
206575	Rippling muscle disease with myasthenia gravis
7	Ritscher-Schinzel syndrome
2737	River blindness
1803	Rivera-Perez-Salas syndrome
137634	RNF135-related overgrowth syndrome
71273	RNS
3103	Roberts syndrome
3103	Roberts-SC phocomelia syndrome
3104	Robin sequence - oligodactyly
97360	Robinow dwarfism
97360	Robinow syndrome
3105	Robinow-like syndrome
97360	Robinow-Silverman-Smith syndrome
2780	Robinow-Unger syndrome
529	Roch-Leri mesosomatous lipomatosis
83311	Rocky Mountain spotted fever
49382	Rod monochromacy
49382	Rod monochromatism
1258	Rodini-Richieri Costa syndrome
49827	Rogers syndrome
293987	ROHHAD
293987	ROHHADNET
353298	Roifman syndrome
247775	Rokitansky sequence
3109	Rokitansky syndrome
163727	Rolandic epilepsy - paroxysmal exercise-induced dystonia - writer's cramp
163721	Rolandic epilepsy - speech dyspraxia
101016	Romano-Ward syndrome
3110	Rombo syndrome
1088	Rommen-Mueller-Sybert syndrome
158014	Rosaï-Dorfman disease
158014	Rosaï-Dorfman-Destombes disease
1837	Rosenberg-Lohr syndrome
329	Rosenthal factor deficiency
329	Rosenthal syndrome
251975	Rosette-forming glioneuronal tumour of the fourth ventricule
90339	Rosselli-Gulienetti syndrome
2909	Rothmund-Thomson syndrome
221008	Rothmund-Thomson syndrome type 1
221016	Rothmund-Thomson syndrome type 2
3111	Rotor syndrome
171709	Round-headed spermatozoa
3115	Roussy-Lévy syndrome
1323	Rozin-camptodactyly syndrome
1323	Rozin-Hertz-Goodman syndrome
280569	RPGN
1507	RRS
818	RSH syndrome
293848	RTLA
2909	RTS
221008	RTS1
221016	RTS2
83616	Rubella panencephalitis
783	Rubinstein-Taybi syndrome
353281	Rubinstein-Taybi syndrome due to 16p13.3 microdeletion
353277	Rubinstein-Taybi syndrome due to CREBBP mutations
353284	Rubinstein-Taybi syndrome due to EP300 haploinsufficiency
1768	Rudd-Klimek syndrome
3118	Rudiger syndrome
79433	Rufous oculocutaneous albinism
1834	Russell-Weaver-Bull syndrome
2709	Rutherfurd syndrome
3121	Ruvalcaba syndrome
293848	rvFTD
461	RXLI
16	S cone monochromacy
16	S cone monochromatism
3105	Saal-Greenstein syndrome
319239	Sabia hemorrhagic fever
3124	Saccharopine dehydrogenase deficiency
3124	Saccharopinuria
286	Sack-Barabas syndrome
3027	Sacral agenesis syndrome
2351	Sacral meningocele - conotroncal heart defects
3027	Sacral regression syndrome
1773	Sacrococcygeal dysgenesis association
85165	SADDAN
794	Saethre-Chotzen syndrome
2872	Sagittal craniostenosis with congenital heart disease, mental deficiency and mandibular ankylosis
300493	Sagliker syndrome
70472	Saguenay-Lac-St. Jean cytochrome oxidase deficiency
70472	Saguenay-Lac-St. Jean type Leigh syndrome
83484	Saint Louis encephalitis
2256	Saito-Kuba-Tsuruta syndrome
3128	Sakati syndrome
3128	Sakati-Nyhan syndrome
3128	Sakati-Nyhan-Tisdale syndrome
1409	Salamon syndrome
2613	Salcedo syndrome

ORPHA Number	Disease name
140969	Saldino-Mainzer syndrome
213557	Salivary gland type cancer of the breast
213557	Salivary gland type carcinoma of the breast
309334	Salla disease
795	Salmonellosis
2230	Salti-Salem syndrome
53721	SAMS 1-31
228123	San Joaquin valley fever
96167	San Luis Valley syndrome
796	Sandhoff disease
309169	Sandhoff disease, adult form
309155	Sandhoff disease, infantile form
309162	Sandhoff disease, juvenile form
71272	Sandifer syndrome
70595	SANDO
2378	Sandrow syndrome
581	Sanfilippo disease
79269	Sanfilippo syndrome type A
79270	Sanfilippo syndrome type B
79271	Sanfilippo syndrome type C
79272	Sanfilippo syndrome type D
2323	Sanjad-Sakati syndrome
588	Santavuori congenital muscular dystrophy
79263	Santavuori disease
79263	Santavuori-Haltia disease
2155	Santos-Mateus-Leal syndrome
247234	SAOA
793	SAPHO syndrome
54368	Sarcocystosis
797	Sarcoidosis
3129	Sarcosine dehydrogenase complex deficiency
3129	Sarcosinemia
54368	Sarcosporidiosis
140896	SARS
140896	SARS-associated corona virus
140896	SARS-CoV
3130	Satoyoshi syndrome
330015	Saturnism
3047	Say-Barber-Biesecker-Young-Simpson syndrome
2013	Say-Barber-Hobbs syndrome
3132	Say-Barber-Miller syndrome
3133	Say-Field-Coldwell syndrome
3369	Say-Meyer syndrome
79157	SBCAD deficiency
481	SBMA
3103	SC phocomelia
3103	SC pseudothalidomide syndrome
98755	SCA1
98756	SCA2
98757	SCA3
276238	SCA3, Joseph type
276244	SCA3, Machado type
98765	SCA4
98766	SCA5
98758	SCA6
94147	SCA7
98760	SCA8
98761	SCA10
98767	SCA11
98762	SCA12
98768	SCA13
98763	SCA14
98769	SCA15/16
98759	SCA17
98771	SCA18
98772	SCA19/22
101110	SCA20
98773	SCA21
101108	SCA23
101111	SCA25
101112	SCA26
98764	SCA27
101109	SCA28
208513	SCA29
211017	SCA30
217012	SCA31
276183	SCA32
276193	SCA35
276198	SCA36
26792	SCAD deficiency
26792	SCADD
254881	SCAE
1003	Scalp defects - postaxial polydactyly
2036	Scalp-ear-nipple syndrome
64753	SCAN 2
94124	SCAN1
168624	Scaphocephaly - macrocephaly - maxillary retrusion - intellectual deficit
2839	Scapuloiliac dysostosis
85146	Scapuloperoneal amyotrophy
64753	SCAR1
1170	SCAR2
95433	SCAR3
95434	SCAR4
83472	SCAR5
284332	SCAR6
284324	SCAR7
88644	SCAR8
139485	SCAR9
284289	SCAR10
284271	SCAR11
284282	SCAR12
3134	SCARF syndrome
90080	Scarring in glaucoma filtration surgical procedures
95434	SCASI
85297	SCAX3
85292	SCAX4
284400	SCCB
98967	SCCD
91365	SCD*
98967	SCD*
1383	Schaap-Taylor-Baraitser syndrome
35123	SCHAD deficiency
93474	Scheie syndrome
2353	Schilbach-Rott syndrome
59298	Schilder disease
59298	Schilder's disease
1830	Schimke immuno-osseous dysplasia
1830	Schimke syndrome
2612	Schimmelpenning syndrome
3137	Schindler disease
79279	Schindler disease type 1
79280	Schindler disease type 2
79281	Schindler disease type 3
3138	Schinzel syndrome
798	Schinzel-Giedion syndrome
63862	Schisis association
1247	Schistosomiasis
799	Schizencephaly
98973	Schlichting dystrophy
3143	Schmidt syndrome
2252	Schmitt-Gillenwater-Kelly syndrome
3144	Schneckenbecken dysplasia
37748	Schnitzler syndrome
98967	Schnyder corneal dystrophy
98967	Schnyder crystalline corneal dystrophy
98967	Schnyder crystalline dystrophy sine crystals
3145	Schofer-Beetz-Bohl syndrome
3041	Scholte-Begeer-van Essen syndrome
50944	Schopf-Schulz-Passarge syndrome
93921	Schwannomatosis
800	Schwartz-Jampel syndrome
800	Schwartz-Jampel syndrome type 1
800	Schwartz-Jampel-Aberfeld syndrome
183660	SCID
277	SCID due to adenosine deaminase deficiency
275	SCID due to artemis deficiency
357237	SCID due to CARD11 deficiency
331206	SCID due to complete RAG1/2 deficiency

*Caution: one same acronym may correspond to different diseases in medical terms. Please refer to the full name of the disease to get the correct Orpha code.

ORPHA Number	Disease name
228003	SCID due to CORO1A deficiency
228003	SCID due to coronin-1A deficiency
275	SCID due to DCLRE1C deficiency
317425	SCID due to DNA-PKcs deficiency
280142	SCID due to LCK deficiency
280142	SCID due to lymphocyte-specific protein tyrosine kinase deficiency
33355	SCID with leukopenia
275	SCID, Athabaskan type
276	SCIDX1
185	Scimitar syndrome
70573	SCLC
352763	Scleredema
75840	Scleroatonic muscular dystrophy
384	Scleroatrophic syndrome
801	Scleroderma
167635	Scleromyxedema
90400	Scleromyxedema without monoclonal gammopathy
75325	Sclerosing dysplasia of bone - ichthyosis - premature ovarian failure
63999	Sclerosing mediastinitis
238593	Sclerosing mesenteritis
100001	Sclerosing perineurioma
3152	Sclerosteosis
384	Sclerotylosis
188	SCLS
331176	SCN4
832	SCOT deficiency
1514	Scott craniodigital syndrome
806	Scott syndrome
1514	Scott-Bryant-Graham syndrome
1509	Scott-Taor syndrome
86813	SCRA
83317	Scrub typhus
84064	SD/THE
295193	SD1, Castilla type
295189	SD1, Lueken type
295191	SD1, Montagu type
295187	SD1, Weidenreich type
295187	SD1a
295189	SD1b
295191	SD1c
295193	SD1d
295197	SD2, Debeer type
295199	SD2, Malik type
295195	SD2, Vordingborg type
295195	SD2a
295197	SD2b
295199	SD2c
93404	SD3
93406	SD5
263463	SDCD, CHST3 type
168577	sdCHC
29072	SDHx-related paraganglioma-pheochromocytoma
300869	SDRPL
98932	SDS
373	SDYS
158029	Sea-blue histiocytosis
1778	Seaver-Cassidy syndrome
841	Sebocystomatosis
168606	Seborrhea-like dermatitis with psoriasiform elements
79480	Seborrheic pemphigus
98873	SEC23B-CDG syndrome
808	Seckel syndrome
141022	Second branchial cleft anomaly
141022	Second branchial cleft cyst
141022	Second branchial cleft fistula
139420	Secondary acute transverse myelitis
85445	Secondary amyloidosis
169618	Secondary central precocious puberty
91365	Secondary ciliary dyskinesia
314962	Secondary HES
314962	Secondary hypereosinophilic syndrome
2615	Secondary hypertrophic osteoperiostosis with pernio
90363	Secondary intestinal lymphangiectasia
3452	Secondary non-tropical sprue
99930	Secondary pulmonary hemosiderosis
1077	Secondary retention of permanent molars
95427	Secondary short bowel syndrome
99857	Secondary syringomyelia
163654	SED-BDS
94068	SEDC
567	Sedlackova syndrome
647	Seemanova syndrome type 2
2528	Seemanova-Lesny syndrome
251618	SEGA
255	Segawa syndrome
2759	Seghers syndrome
67039	Segmental odontomaxillary dysplasia
137608	Segmental outgrowth - lipomatosis - arteriovenous malformation - epidermal nevus
314662	Segmental progressive overgrowth syndrome with fibroadipose hyperplasia
455	SEI
35069	Seitelberger disease
79156	Seizures - intellectual deficit due to hydroxylysinuria
199343	Seizures - sensorineural deafness - ataxia - intellectual deficit - electrolyte imbalance
35858	Selective cobalamin malabsorption with proteinuria
183675	Selective IgG subclass deficiency
331235	Selective IgM deficiency
331235	Selective immunoglobulin M deficiency
165994	Selective pituitary resistance to thyroid hormone
99798	Selective tooth agenesis
281122	Self-healing collodion baby
90397	Self-healing papular mucinosis
65748	Self-healing squamous epithelioma type 1
1850	Selig-Benacerraf-Greene syndrome
3232	Sellars-Beighton syndrome
100069	Semantic dementia
100069	Semantic primary progressive aphasia
100069	Semantic variant PPA
93356	SEMD type 2
93351	SEMD type Irapa
171866	SEMD, aggrecan type
93351	SEMD, Irapa type
156728	SEMD, MATN3-related
156728	SEMD, matrilin-3 type
93356	SEMD, Missouri type
93352	SEMD, Shohat type
93359	SEMDJL
93359	SEMDJL1
93360	SEMDJL2
93360	SEMD-MD
220386	Semilobar holoprosencephaly
329284	SENDA
79480	Senear-Usher syndrome
1369	Sengers syndrome
2183	Sengers-Hamel-Otten syndrome
330001	Senile systemic amyloidosis
1292	Senior syndrome
84081	Senior-Boichis syndrome
3156	Senior-Loken syndrome
1515	Sensenbrenner syndrome
217622	Sensorineural deafness with dilated cardiomyopathy
66633	Sensorineural hearing loss - early greying - essential tremor
97229	Sensorineural hearing loss - pontobulbar palsy
217622	Sensorineural hearing loss with dilated cardiomyopathy
70595	Sensory ataxic neuropathy - dysarthria - ophthalmoparesis
477	Senter syndrome
90118	SEOAN due to MFN2 deficiency
90051	Sepsis in premature infants
180154	Septate vagina

ORPHA Number	Disease name
137839	Septic phlebitis of the internal jugular vein
3157	Septo-optic dysplasia
3157	Septo-optic dysplasia spectrum
280195	Septopreoptic holoprosencephaly
280195	Septopreoptic HPE
139466	SERKAL syndrome
43116	Serotonergic syndrome
43116	Serotonin storm
43116	Serotonin syndrome
43116	Serotonin toxicity
43116	Serotonin toxidrome
206470	Serous or mucinous cystadenoma of childhood
168829	Serous surface papillary carcinoma
35686	Serpiginous choroiditis
2901	Serum neuritis
75508	Servelle-Martorell syndrome
199343	SeSAME syndrome
85165	Severe achondroplasia - developmental delay - acanthosis nigricans
140896	Severe acute respiratory syndrome
314911	Severe Canavan disease
183660	Severe combined immunodeficiency
277	Severe combined immunodeficiency due to adenosine deaminase deficiency
275	Severe combined immunodeficiency due to artemis deficiency
357237	Severe combined immunodeficiency due to CARD11 deficiency
331206	Severe combined immunodeficiency due to complete RAG1/2 deficiency
228003	Severe combined immunodeficiency due to CORO1A deficiency
228003	Severe combined immunodeficiency due to coronin-1A deficiency
275	Severe combined immunodeficiency due to DCLRE1C deficiency
317425	Severe combined immunodeficiency due to DNA-PKcs deficiency
280142	Severe combined immunodeficiency due to LCK deficiency
280142	Severe combined immunodeficiency due to lymphocyte-specific protein tyrosine kinase deficiency
33355	Severe combined immunodeficiency with leukopenia
275	Severe combined immunodeficiency, Athabaskan type
209370	Severe congenital encephalopathy due to MECP2 mutation
300298	Severe congenital hypochromic anemia with ringed sideroblasts
300298	Severe congenital hypochromic sideroblastic anemia
171430	Severe congenital nemaline myopathy
42738	Severe congenital neutropenia
331176	Severe congenital neutropenia - pulmonary hypertension - superficial venous angiectasis
331176	Severe congenital neutropenia 4
262	Severe dystrophinopathy, Duchenne and Becker type
98896	Severe dystrophinopathy, Duchenne type
228374	Severe early-onset axonal neuropathy due to light neurofilament subunit deficiency
90118	Severe early-onset axonal neuropathy due to MFN2 deficiency
228374	Severe early-onset axonal neuropathy due to NEFL deficiency
329249	Severe early-onset obesity-insulin resistance syndrome due to SH2B1 deficiency
169793	Severe factor IX deficiency
169802	Severe factor VIII deficiency
352577	Severe feeding difficulties - failure to thrive - microcephaly due to ASXL3 deficiency
79408	Severe generalized RDEB
79408	Severe generalized recessive dystrophic epidermolysis bullosa
2109	Severe Hallermann-Streiff-François syndrome
169802	Severe hemophilia A
169793	Severe hemophilia B
94075	Severe immune-mediated enteropathy
98920	Severe infantile axonal neuropathy with respiratory failure
94066	Severe intellectual deficit - epilepsy - anal anomalies - distal phalangeal hypoplasia
280763	Severe intellectual deficit and progressive spastic paraplegia
324307	Severe lateral tibial bowing with short stature
2879	Severe limb deficit
33069	Severe myoclonic epilepsy of infancy
209370	Severe neonatal-onset encephalopathy with microcephaly
216812	Severe osteogenesis imperfecta
280210	Severe PMD
163703	Severe refractory status epilepticus owing to presumed encephalitis
169095	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy
3078	Severe X-linked intellectual deficit, Gustavson type
238329	Severe X-linked mitochondrial encephalomyopathy
139466	Sex reversion - kidneys, adrenal and lung dysgenesis
3162	Sézary lymphoma
3162	Sézary syndrome
373	SGBS
373	SGBS1
79022	SGBS2
35710	SGLT1 deficiency
69076	SGLT2 deficiency
2462	SGS
2407	Shabbir syndrome
897	Shah-Waardenburg syndrome
29822	Shapiro syndrome
1506	Sharma-Kapoor-Ramji syndrome
809	Sharp syndrome
281122	SHCB
91355	Sheehan syndrome
1147	Sheldon-Hall syndrome
3329	SHFLD syndrome
2440	SHFM
3329	SHFM associated with aplasia of long bones
90038	Shiga-like toxin-associated HUS
810	Shigellosis
158014	SHML
1008	Shokeir syndrome
99063	Shone complex
1738	Short arm of chromosome 4 trisomy
1742	Short arm of chromosome 5 trisomy
236	Short arm of chromosome 9 duplication
236	Short arm of chromosome 9 trisomy
1715	Short arm of chromosome 18 duplication
1715	Short arm of chromosome 18 trisomy
95427	Short bowel syndrome due to necrotizing enterocolitis
95427	Short bowel syndrome due to surgical resection
95427	Short bowel syndrome due to thrombosis
95427	Short bowel syndrome due to total or sub-total aganglionosis
95427	Short bowel syndrome due to trauma
95427	Short bowel syndrome due to volvulus
35123	Short chain 3-hydroxyacyl-CoA dehydrogenase deficiency
26792	Short chain acyl-CoA dehydrogenase deficiency
66518	Short fifth metacarpals - insulin resistance
294996	Short fingers
295130	Short fingers, bilateral
295128	Short fingers, unilateral
1505	Short rib-polydactyly syndrome
93270	Short rib-polydactyly syndrome type 1
93269	Short rib-polydactyly syndrome type 2
93271	Short rib-polydactyly syndrome type 3
93268	Short rib-polydactyly syndrome type 4
93268	Short rib-polydactyly syndrome, Beemer-Langer type
93269	Short rib-polydactyly syndrome, Majewski type

ORPHA Number	Disease name
93270	Short rib-polydactyly syndrome, Saldino-Noonan type
93271	Short rib-polydactyly syndrome, Verma-Naumoff type
156723	Short ribs - craniosynostosis - polysyndactyly
2994	Short stature - craniofacial anomalies - genital hypoplasia
2866	Short stature - deafness - neutrophil dysfunction - dysmorphism
2332	Short stature - facial and skeletal anomalies - intellectual deficit - macrodontia
1088	Short stature - heart defect - craniofacial anomalies
2860	Short stature - hyperkaliemia - acidosis
2649	Short stature - intellectual deficit - eye anomalies - cleft lip/palate
1937	Short stature - locking fingers
2861	Short stature - microcephaly - heart defect
3102	Short stature - Pierre Robin sequence - cleft mandible - hand anomalies clubfoot
3102	Short stature - Pierre Robin syndrome - cleft mandible - hand anomalies clubfoot
85442	Short stature - pituitary and cerebellar defects - small sella turcica
2864	Short stature - prognathism - short femoral necks
2868	Short stature - valvular heart disease - characteristic facies
2865	Short stature - webbed neck - heart disease
2863	Short stature - wormian bones - dextrocardia
314811	Short stature due to GHSR deficiency
629	Short stature due to growth hormone qualitative anomaly
633	Short stature due to growth hormone resistance
314811	Short stature due to growth hormone secretagogue receptor deficiency
632	Short stature due to isolated growth hormone deficiency with X-linked hypogammaglobulinemia
314802	Short stature due to partial GHR deficiency
314802	Short stature due to partial growth hormone receptor deficiency
140941	Short stature due to primary acid-labile subunit deficiency
220465	Short stature due to STAT5b deficiency
2867	Short stature, Brussels type
171706	Short stature-delayed bone age due to thyroid hormone metabolism deficiency
314394	Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome
3163	SHORT syndrome
2832	Short tarsus - absence of lower eyelashes
251515	Short tendo calcaneus
294998	Short toes
295134	Short toes, bilateral
295132	Short toes, unilateral
357175	Short ulna - dysmorphism - hypotonia - intellectual deficit
79157	Short/branched-chain acyl-coA dehydrogenase deficiency
57145	Short-lasting unilateral neuralgiform headache attacks with conjunctival injection and tearing
935	Short-limb skeletal dysplasia with SCID
935	Short-limb skeletal dysplasia with severe combined immunodeficiency
2580	Shoulder and girdle defects - familial intellectual deficit
1940	Shoulder and thorax deformity - congenital heart disease
314795	Shox-related short stature
567	Shprintzen syndrome
2462	Shprintzen-Goldberg syndrome
3165	Shulman syndrome
811	Shwachman-Diamond syndrome
98932	Shy-Drager syndrome
812	Sialidosis type 1
87876	Sialidosis type 2
3166	Sialuria
3166	Sialuria, French type
98920	SIANRF
75789	SIBIDS syndrome
611	sIBM
166282	Sick sinus syndrome
251359	Sickle cell - beta-thalassemia disease
251365	Sickle cell - hemoglobin C disease
251370	Sickle cell - hemoglobin D disease
251375	Sickle cell - hemoglobin E disease
232	Sickle cell anemia
232	Sickle cell disease
210272	Sickness of disembarkment
838	SICRET syndrome
168593	SIDDT
54028	Sideropenic dysphagia
2267	Sidransky-Feinstein-Goodman syndrome
3167	Siegler-Brewer-Carey syndrome
314786	Silent pituitary adenoma
71276	Silent sinus syndrome
3168	Sillence syndrome
60014	Silver staining
100998	Silver Syndrome
813	Silver-Russell dwarfism
813	Silver-Russell syndrome
231144	Silver-Russell syndrome due to 11p15 microduplication
231137	Silver-Russell syndrome due to 7p11.2p13 microduplication
231137	Silver-Russell syndrome due to 7p11.2-p13 microduplication
231137	Silver-Russell syndrome due to dup(7)(p11.2p13)
231140	Silver-Russell syndrome due to imprinting defect of 11p15
231147	Silver-Russell syndrome due to maternal uniparental disomy of chromosome 11
96182	Silver-Russell syndrome due to maternal uniparental disomy of chromosome 7
231137	Silver-Russell syndrome due to trisomy 7p11.2p13
231137	Silver-Russell syndrome due to trisomy 7p11.2-p13
1968	Simosa-Penchaszadeh-Bustos syndrome
91139	Simple cryoglobulinemia
373	Simpson dysmorphia syndrome
373	Simpson-Golabi-Behmel syndrome
373	Simpson-Golabi-Behmel syndrome type 1
79022	Simpson-Golabi-Behmel syndrome type 2
97337	Sinding-Larsen-Johansson disease
50809	Singh-Williams-McAlister syndrome
2286	Single upper central incisor
99097	Single ventricular septal defect
85191	Singleton-Merten dysplasia
85191	Singleton-Merten syndrome
324321	Sinoatrial node dysfunction and deafness
1260	Sino-auricular heart block
158014	Sinus histiocytosis with massive lymphadenopathy
3122	Sinus node disease - myopia
166282	Sinus node dysfunction
890	Sinusoidal obstruction syndrome
247698	Sipple syndrome
3169	Sirenomelia
2882	Sitosterolemia
157769	Situs ambiguous
157769	Situs ambiguus
101063	Situs inversus
101063	Situs inversus totalis
816	Sjögren-Larsson syndrome
800	SJS
800	SJS1
95455	SJS-TEN
2565	Skeletal dysplasia - brachydactyly
1858	Skeletal dysplasia - epilepsy - short stature
1436	Skeletal dysplasia - intellectual deficit
166277	Skeletal dysplasia with wormian bone - multiple fractures - dentin abnormality
1426	Skeletal dysplasia, Greenberg type
1860	Skeletal dysplasia, San Diego type

ORPHA Number	Disease name
293165	Skin fragility-woolly hair-palmoplantar hyperkeratosis syndrome
293165	Skin fragility-woolly hair-palmoplantar keratoderma syndrome
178475	Skin infectious botulism
178475	Skin toxin-mediated botulism
238459	SLC35A1-CDG syndrome
356961	SLC35A2-CDG syndrome
99843	SLC35C1-CDG syndrome
3144	SLC35D1-CDG syndrome
93552	SLE, pediatric onset
3385	Sleeping sickness
88633	SLK
818	SLOS
70472	SLSJ-COX deficiency
3156	SLSN
584	Sly disease
70	SMA
83419	SMA type 3
83330	SMA1
83418	SMA2
83419	SMA3
83420	SMA4
83330	SMA-I
83418	SMA-II
83419	SMA-III
83420	SMA-IV
104075	Small bowel adenocarcinoma
104076	Small bowel leiomyosarcoma
284400	Small cell bladder cancer
284400	Small cell bladder carcinoma
284400	Small cell carcinoma of the bladder
284400	Small cell carcinoma of the urinary bladder
70573	Small cell lung cancer
838	Small infarctions of cochlear, retinal and encephalic tissue
1201	Small intestinal atresia
67038	Small lymphocytic lymphoma
543	Small non-cleaved cell lymphoma
1509	Small patella syndrome
98920	SMARD
98920	SMARD1
1145	SMAX2
98959	SMCD
85167	SMD-CRD
33069	SMEI
93974	Smith-Fineman-Myers syndrome
818	Smith-Lemli-Opitz syndrome
819	Smith-Magenis syndrome
178355	Smith-McCort dysplasia
2286	SMMCI
158775	Smouldering systemic mastocytosis
86854	SMZL
820	Sneddon syndrome
48377	Sneddon-Wilkinson disease
91496	Snowflake vitreoretinal degeneration
3063	Snyder-Robinson syndrome
3157	SOD*
67039	SOD*
306577	Sodium channelopathy-related small fiber neuropathy
99903	Sodoku
99772	Soft cleft palate
314394	SOFT syndrome
100002	Soft tissu perineurioma
2234	Sohval-Soffer syndrome
137608	SOLAMEN syndrome
97230	Solar urticaria
83468	Solitary bone cyst
2126	Solitary fibrous tumor
79455	Solitary mastocytoma
2286	Solitary median maxillary central incisor syndrome
100035	Solitary necrotic tumor of the liver
86855	Solitary plasmacytoma
209964	Solitary rectal ulcer syndrome
2612	Solomon syndrome
314769	Somatomammotropinoma
97283	Somatostatinoma
2564	Sommer-Hines syndrome
1064	Sommer-Rathbun-Battles syndrome
1529	Sommer-Young-Wee-Frye syndrome
1355	Sonoda syndrome
1471	Sorsby syndrome
59181	Sorsby's fundus dystrophy
821	Sotos syndrome
98868	Southeast asian ovalocytosis
352403	SPARCA
352403	SPARCA1
79132	Sparse hair - short stature - skin anomalies
93961	Spasmodic dysphonia
93962	Spasmodic torticollis
279882	Spasmus nutans
2572	Spastic ataxia - corneal dystrophy
2572	Spastic ataxia - ocular anomalies
1182	Spastic ataxia with congenital miosis
1680	Spastic diplegia, infantile type
99015	Spastic gait type 2
100990	Spastic paraparesis - amyopathy - cataracts - gastroesophageal reflux
2815	Spastic paraparesis - deafness
101003	Spastic paraparesis - vitiligo - premature graying - characteristic facies
99015	Spastic paraparesis type 2
2816	Spastic paraplegia - epilepsy - intellectual deficit
2819	Spastic paraplegia - facial-cutaneous lesions
2818	Spastic paraplegia - glaucoma - intellectual deficit
2822	Spastic paraplegia - intellectual deficit - thin corpus callosum
2820	Spastic paraplegia - nephritis - deafness
2821	Spastic paraplegia - neuropathy - poikiloderma
329475	Spastic paraplegia - Paget disease of bone
2826	Spastic paraplegia - precocious puberty
100996	Spastic paraplegia - retinal degeneration
139480	Spastic paraplegia due to neuropathy target esterase mutation
139480	Spastic paraplegia due to NTE mutation
99015	Spastic paraplegia type 2
100998	Spastic paraplegia-amyotrophy of hands and feet
320406	Spastic paraplegia-optic atrophy-neuropathy syndrome
3011	Spastic quadriplegia - retinitis pigmentosa - intellectual deficit
3011	Spastic tetraplegia - retinitis pigmentosa - intellectual deficit
3175	Spasticity - intellectual deficit - X-linked epilepsy
251282	SPAX1
314603	SPAX3
254343	SPAX4
313772	SPAX5
295197	SPD, Debeer type
295199	SPD, Malik type
295195	SPD, Vordingborg type
295195	SPD1
295197	SPD2
295199	SPD3
169443	Specific antibody deficiency with normal immunoglobulin concentrations and normal numbers of B cells
352403	Spectrin-associated autosomal recessive cerebellar ataxia
352403	Spectrin-associated autosomal recessive cerebellar ataxia type 1
209908	Speech and language disorder with orofacial dyspraxia
209908	Speech-language disorder type 1
1855	SPENCD
2816	SPERM
99865	Spermatocytic seminoma
685	SPG
306617	SPG1

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ORPHA Number	Disease name
99015	SPG2
100985	SPG4
100986	SPG5A
100988	SPG6
99013	SPG7
100989	SPG8
100990	SPG9
100991	SPG10
2822	SPG11
100993	SPG12
100994	SPG13
100995	SPG14
100996	SPG15
100997	SPG16
100998	SPG17
209951	SPG18
100999	SPG19
101000	SPG20
101001	SPG21
101003	SPG23
101004	SPG24
101005	SPG25
101006	SPG26
101007	SPG27
101008	SPG28
101009	SPG29
101010	SPG30
101011	SPG31
171622	SPG32
171607	SPG34
171629	SPG35
320365	SPG36
171612	SPG37
171617	SPG38
139480	SPG39
320355	SPG41
171863	SPG42
320370	SPG43
320401	SPG44
320396	SPG45
320391	SPG46
306511	SPG48
320385	SPG49
319199	SPG53
320380	SPG54
320375	SPG55
320411	SPG56
98866	Spherocytic elliptocytosis
268129	Spheroid body myopathy
3449	Spherophakia - brachymorphia
306553	Spherulocytosis
79264	Spielmeyer-Vogt disease
314432	Spigelian hernia-cryptorchidism syndrome
3176	Spina bifida - hypospadias
268369	Spina bifida aperta
268744	Spina bifida cystica
481	Spinal and bulbar muscular atrophy
79093	Spinal arteriovenous malformation
53721	Spinal arteriovenous metameric syndrome
1217	Spinal atrophy - ophthalmoplegia - pyramidal syndrome
90058	Spinal cord injury
73245	Spinal muscular atrophy - Dandy-Walker complex - cataracts
1145	Spinal muscular atrophy with arthrogryposis
98920	Spinal muscular atrophy with respiratory distress
83420	Spinal muscular atrophy, adult form
210584	Spindle cell hemangioendothelioma
210584	Spindle cell hemangioma
481	Spinobulbar muscular atrophy
2074	Spinocerebellar ataxia - amyotrophy - deafness
1185	Spinocerebellar ataxia - dysmorphism
98755	Spinocerebellar ataxia type 1
94124	Spinocerebellar ataxia type 1 with axonal neuropathy
98756	Spinocerebellar ataxia type 2
98757	Spinocerebellar ataxia type 3
98765	Spinocerebellar ataxia type 4
98766	Spinocerebellar ataxia type 5
98758	Spinocerebellar ataxia type 6
94147	Spinocerebellar ataxia type 7
98760	Spinocerebellar ataxia type 8
98761	Spinocerebellar ataxia type 10
98767	Spinocerebellar ataxia type 11
98762	Spinocerebellar ataxia type 12
98768	Spinocerebellar ataxia type 13
98763	Spinocerebellar ataxia type 14
98769	Spinocerebellar ataxia type 15/16
98759	Spinocerebellar ataxia type 17
98771	Spinocerebellar ataxia type 18
98772	Spinocerebellar ataxia type 19/22
101110	Spinocerebellar ataxia type 20
98773	Spinocerebellar ataxia type 21
101108	Spinocerebellar ataxia type 23
101111	Spinocerebellar ataxia type 25
101112	Spinocerebellar ataxia type 26
98764	Spinocerebellar ataxia type 27
101109	Spinocerebellar ataxia type 28
208513	Spinocerebellar ataxia type 29
211017	Spinocerebellar ataxia type 30
217012	Spinocerebellar ataxia type 31
276183	Spinocerebellar ataxia type 32
276193	Spinocerebellar ataxia type 35
276198	Spinocerebellar ataxia type 36
64753	Spinocerebellar ataxia with axonal neuropathy type 2
254881	Spinocerebellar ataxia with epilepsy
3177	Spinocerebellar degeneration - corneal dystrophy
99903	Spirillary rat-bite fever
757	Spitzer-Weinstein syndrome
300869	Splenic diffuse red pulp B-cell lymphoma
300869	Splenic diffuse red pulp lymphoma
86854	Splenic marginal zone lymphoma
2063	Splenogonadal fusion - limb defects - micrognathia
294994	Split foot
2439	Split foot deformity - mandibulofacial dysostosis
295126	Split foot, bilateral
295124	Split foot, unilateral
294992	Split hand
71271	Split hand - split foot - deafness
2437	Split hand - urinary anomalies - spina bifida
2440	Split hand foot malformation
295122	Split hand, bilateral
295120	Split hand, unilateral
3329	Split hand/foot malformation with long bone deficiency
958	Split hand/split foot - mandibular hypoplasia
2329	Split hand/split foot - nystagmus
2440	Split hand-split foot malformation
1756	Split notochord syndrome
3329	Split-hand/foot malformation associated with aplasia of long bones
320406	SPOAN
93357	SPONASTRIME dysplasia
3180	Spondylocamptodactyly syndrome
3275	Spondylocarpotarsal synostosis
94095	Spondylocostal dysostosis - anal and genitourinary malformations
329252	Spondylocostal dysostosis - hypospadias - intellectual deficit
1855	Spondyloenchondrodysplasia
1855	Spondyloenchondromatosis
168451	Spondyloepimetaphyseal dysplasia - abnormal dentition
168443	Spondyloepimetaphyseal dysplasia - hypotrichosis
93358	Spondyloepimetaphyseal dysplasia - short limb - abnormal calcification
93346	Spondyloepimetaphyseal dysplasia congenita, Strudwick type

ORPHA Number	Disease name
93356	Spondyloepimetaphyseal dysplasia type 2
93360	Spondyloepimetaphyseal dysplasia with joint laxicity, Hall type
93359	Spondyloepimetaphyseal dysplasia with joint laxity
93359	Spondyloepimetaphyseal dysplasia with joint laxity type 1
93360	Spondyloepimetaphyseal dysplasia with joint laxity type 2
93360	Spondyloepimetaphyseal dysplasia with joint laxity, leptodactylic type
93360	Spondyloepimetaphyseal dysplasia with multiple dislocations
93360	Spondyloepimetaphyseal dysplasia with multiple dislocations, Hall type
171866	Spondyloepimetaphyseal dysplasia, aggrecan type
93347	Spondyloepimetaphyseal dysplasia, anauxetic type
168448	Spondyloepimetaphyseal dysplasia, Bieganski type
168454	Spondyloepimetaphyseal dysplasia, Geneviève type
99642	Spondyloepimetaphyseal dysplasia, Handigodu type
93351	Spondyloepimetaphyseal dysplasia, Irapa type
156728	Spondyloepimetaphyseal dysplasia, matrilin-3 type
93347	Spondyloepimetaphyseal dysplasia, Menger type
93356	Spondyloepimetaphyseal dysplasia, Missouri type
93282	Spondyloepimetaphyseal dysplasia, Pakistani type
93352	Spondyloepimetaphyseal dysplasia, Shohat type
93357	Spondyloepimetaphyseal dysplasia, Sponastrime type
163654	Spondyloepiphyseal dysplasia - brachydactyly - speech disorder
163649	Spondyloepiphyseal dysplasia - craniosynostosis - cleft palate - cataract - intellectual deficit
163668	Spondyloepiphyseal dysplasia - myopia - sensorineural deafness
1830	Spondyloepiphyseal dysplasia - nephrotic syndrome
353298	Spondyloepiphyseal dysplasia - retinal dystrophy - immunodeficiency
94068	Spondyloepiphyseal dysplasia congenita
93284	Spondyloepiphyseal dysplasia tarda
1159	Spondyloepiphyseal dysplasia tarda - progressive arthropathy
163665	Spondyloepiphyseal dysplasia tarda, Kohn type
263463	Spondyloepiphyseal dysplasia with congenital joint dyslocations, CHST3 type
163654	Spondyloepiphyseal dysplasia, Cantu type

ORPHA Number	Disease name
93283	Spondyloepiphyseal dysplasia, Kimberley type
163668	Spondyloepiphyseal dysplasia, MacDermot type
263482	Spondyloepiphyseal dysplasia, Maroteaux type
163649	Spondyloepiphyseal dysplasia, Nishimura type
163662	Spondyloepiphyseal dysplasia, Reardon type
1190	Spondylo-humero-femoral dysplasia
228387	Spondylo-megaepiphyseal-metaphyseal dysplasia
168552	Spondylometaphyseal dysplasia - bowed forearms - facial dysmorphism
85167	Spondylometaphyseal dysplasia - cone-rod dystrophy
1855	Spondylometaphyseal dysplasia with enchondromatous changes
93316	Spondylometaphyseal dysplasia with severe genu valgum
168555	Spondylometaphyseal dysplasia, A4 type
93316	Spondylometaphyseal dysplasia, Algerian type
93315	Spondylometaphyseal dysplasia, 'corner fracture' type
168544	Spondylometaphyseal dysplasia, Golden type
93314	Spondylometaphyseal dysplasia, Kozlowski type
93316	Spondylometaphyseal dysplasia, Schmidt type
93317	Spondylometaphyseal dysplasia, Sedaghatian type
93315	Spondylometaphyseal dysplasia, Sutcliffe type
85194	Spondylo-ocular syndrome
1856	Spondyloperipheral dysplasia - short ulna
141	Spongy degeneration of the brain
54260	Spongy myocardium
29822	Spontaneous periodic hypothermia
99722	Sporadic achalasia
247234	Sporadic adult-onset ataxia of unknown etiology
225147	Sporadic IBSN
84271	Sporadic idiopathic nephrosis
84271	Sporadic idiopathic steroid-resistant nephrotic syndrome
97555	Sporadic idiopathic steroid-resistant nephrotic syndrome with collapsing glomerulopathy
93222	Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial proliferation
93220	Sporadic idiopathic steroid-resistant nephrotic syndrome with diffuse mesangial sclerosis

ORPHA Number	Disease name
93218	Sporadic idiopathic steroid-resistant nephrotic syndrome with focal segmental glomerulosclerosis
93218	Sporadic idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis
93221	Sporadic idiopathic steroid-resistant nephrotic syndrome with minimal changes
611	Sporadic inclusion body myositis
225147	Sporadic infantile bilateral striatal necrosis
225147	Sporadic infantile striatonigral necrosis
255199	Sporadic infantile subacute necrotizing encephalopathy
255199	Sporadic Leigh disease
255199	Sporadic Leigh syndrome
227510	Sporadic olivopontocerebellar atrophy type 1
227510	Sporadic OPCA type 1
276624	Sporadic pheochromocytoma
276621	Sporadic pheochromocytoma/secreting paraganglioma
276627	Sporadic secreting paraganglioma
826	Sporotrichosis
94068	Spranger-Wiedemann disease
3181	Sprengel deformity
70476	Spring catarrh
234	Sprinz-Nelson syndrome
3198	SPS*
1509	SPS*
86884	SPTCL
51083	SQTS
67037	Squamous cell carcinoma of head and neck
213716	Squamous cell carcinoma of the corpus uteri
324737	SRD5A3-CDG syndrome
83601	SREAT
2806	SSPE
50944	SSPS
2323	SSS
36236	SSSS
83484	St. Louis encephalitis
171714	ST3GAL5-CDG syndrome
2454	Stalker-Chitayat syndrome
1798	Stanescu osteosclerosis
3235	Stapedo-vestibular ankylosis
140917	Stapes ankylosis with broad thumbs and toes
36238	Staphylococcal necrotizing pneumonia
36236	Staphylococcal scalded skin syndrome
36235	Staphylococcal scarlet fever
99919	Staphylococcal toxic-shock syndrome
99919	Staphylococcal TSS

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ORPHA Number	Disease name
140952	STAR syndrome
827	Stargardt disease
85146	Stark-Kaeser syndrome
166427	Startle epilepsy
329284	Static encephalopathy of childhood with neurdegeneration in adulthood
841	Steatocystoma multiplex
3184	Steatocystoma multiplex - natal teeth
240071	Steele-Richardson-Olszewski disease
565	Steely hair disease
565	Steely hair syndrome
273	Steinert disease
273	Steinert myotonic dystrophy
3186	Steinfeld syndrome
168953	Stem cell leukemia/lymphoma
99087	Stenosis or atrophy of the coronary ostium
210115	Sterile multifocal osteomyelitis with periostitis and pustulosis
2017	Sternal cleft
3195	Sternal malformation - vascular dysplasia
3194	Stern-Lubinsky-Durrie syndrome
2017	Sternum bifidum
3196	Steroid dehydrogenase deficiency - dental anomalies
461	Steroid sulfatase deficiency
83601	Steroid-responsive encephalopathy associated with autoimmune thyroiditis
93207	Steroid-sensitive MCNS
97552	Steroid-sensitive nephrotic syndrome without renal biopsy
909	Sterol 27-hydroxylase deficiency
46059	Sterol C5-desaturase deficiency
36426	Stevens-Johnson syndrome
828	Stickler syndrome
90653	Stickler syndrome type 1
90654	Stickler syndrome type 2
166100	Stickler syndrome type 3
166100	Stickler syndrome, nonocular type
3197	Stiff baby syndrome
3198	Stiff man syndrome
3198	Stiff person syndrome
2833	Stiff skin syndrome
85414	Still disease
3199	Stimmler syndrome
2972	Stoelinga-de Koomen-Davis syndrome
3200	Stoll-Alembik-Finck syndrome
3074	Stoll-Géraudel-Chauvin syndrome
3201	Stoll-Kieny-Dott syndrome
2878	Stoll-Lévy-Francfort syndrome
63443	Stomach cancer
168577	Stomatin-deficient cryohydrocytosis
3204	Stormorken-Sjaastad-Langslet syndrome
99064	Straddling and/or overriding mitral valve
95461	Straddling or overriding tricuspid valve
1277	Stratton-Garcia-Young syndrome
2863	Stratton-Parker syndrome
99905	Streptobacillary rat-bite fever
99918	Streptococcal toxic-shock syndrome
99918	Streptococcal TSS
66529	Stress cardiomyopathy
90041	Stress erythrocytosis
90041	Stress polycythemia
50942	Striate palmoplantar keratoderma
98933	Striatonigral degeneration
137599	Stromal keratitis
213711	Stromal sarcoma of the corpus uteri
76	Strongyloidiasis
100984	Strümpell disease
685	Strümpell-Lorrain disease
328	Stuart-Prower factor deficiency
830	Stuccokeratosis
3205	Sturge-Weber syndrome
3206	Stüve-Wiedemann dysplasia
3206	Stüve-Wiedemann syndrome
166277	Suarez-Stickler syndrome
79093	Subacute angiohypertrophic myelomalacia
79093	Subacute ascending necrotizing myelitis
163525	Subacute cutaneous lupus erythematosus
2806	Subacute inclusion body encephalitis
206594	Subacute inflammatory demyelinating polyneuropathy
206594	Subacute inflammatory demyelinating polyradiculoneuropathy
98824	Subacute myeloid leukemia
79093	Subacute necrotizing myelitis
2806	Subacute sclerosing leukoencephalitis
2806	Subacute sclerosing panencephalitis
356	Subacute spongiform encephalopathy, Gerstmann-Straussler type
99113	Subaortic course of brachiocephalic vein
99113	Subaortic course of innominate vein
3191	Subaortic stenosis - short stature
48377	Subcorneal pustular dermatitis
48377	Subcorneal pustular dermatosis
99796	Subcortical band heterotopia
99796	Subcortical laminar heterotopia
101029	Sub-cortical nodular heterotopia
86884	Subcutaneous panniculitic T-cell lymphoma
86884	Subcutaneous panniculitis-like T-cell lymphoma
251618	Subependymal giant cell astrocytoma
101030	Subependymal nodular heterotopia
251639	Subependymoma
98957	Subepithelial amyloidosis of the cornea
98959	Subepithelial mucinous corneal dystrophy
155878	Submucosal cleft palate
3190	Subpulmonary stenosis
1606	Subtelomeric 1p36 deletion
96168	Subtelomeric deletion 13q34
180129	Subtotal septate uterus
22	Succinic semialdehyde dehydrogenase deficiency
832	Succinyl-CoA acetoacetate transferase deficiency
702	Sudanophilic leukodystrophy, Paelizeus-Merzbacher type
168593	Sudden infant death - dysgenesis of the testes
130	Sudden unexplained nocturnal death syndrome
2752	Sugarman syndrome
3412	Sujansky-Leonard syndrome
99732	Sulfite oxidase deficiency due to molybdenum cofactor deficiency
308386	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A
308393	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type B
308400	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type C
99731	Sulfocysteinuria
65682	Summerskill-Walshe-Tygstrup syndrome
254395	Summertime actinic lichenoid eruption
3210	Summitt syndrome
57145	SUNCT syndrome
130	SUNDS
455	Superficial epidermolytic ichthyosis
98961	Superficial granular corneal dystrophy
79490	Superficial lymphangioma
79490	Superficial lymphatic malformation
46485	Superficial pemphigus
247245	Superficial siderosis
88633	Superior limbic keratoconjunctivitis
180182	Supernumerary breasts
96170	Supernumerary der(22) syndrome
141096	Supernumerary nostril
295002	Supernumerary phalanges
295002	Supernumerary phalanx
295142	Supernumerary phallanges, bilateral
295140	Supernumerary phallanges, unilateral
295142	Supernumerary phallanx, bilateral
295140	Supernumerary phallanx, unilateral
764	Suppurative myositis
3193	Supravalvar aortic stenosis
3193	Supravalvular aortic stenosis

ORPHA Number	Disease name
3192	Supravalvular pulmonary stenosis
838	Susac syndrome
2566	Susceptibility to chronic infection by Epstein-Barr virus
169085	Susceptibility to respiratory infections associated with CD8alpha chain mutation
3193	SVAS
86813	Sveinsson chorioretinal atrophy
3243	Sweet syndrome
247165	Swift disease
247165	Swift-Feer disease
242	Swyer syndrome
90038	Sxt-HUS
306731	Sydenham chorea
295138	Symbrachydactyly of hand and foot, bilateral
295136	Symbrachydactyly of hand and foot, unilateral
1570	Symbrachydactyly of hands and feet
1314	Symmetrical thalamic calcifications
79098	Sympathetic ophthalmia
79098	Sympathetic uveitis
635	Sympathoblastoma
3237	Symphalangism - brachydactyly
3246	Symphalangism with multiple anomalies of hands and feet
3250	Symphalangism, Cushing type
276630	Symptomatic form of Coffin-Lowry syndrome in female carriers
177926	Symptomatic form of hemophilia A in female carriers
177929	Symptomatic form of hemophilia B in female carriers
206546	Symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers
357332	Synactyly - camptodactyly and clinodactyly of fifth fingers - bifid halluces
98915	Synaptic congenital myasthenic syndromes
3286	Syncopal paroxysmal tachycardia
3286	Syncopal tachyarythmia
357332	Syndactyly - camptodactyly and clinodactyly of fifth fingers - bifid toes
3253	Syndactyly - ectodermal dysplasia - cleft/lip palate
85203	Syndactyly - preaxial polydactyly - sternal deformity
140952	Syndactyly - telecanthus - anogenital and renal malformations
93404	Syndactyly of fingers 4 and 5
93402	Syndactyly type 1
3255	Syndactyly type 1 - microcephaly - intellectual deficit
295193	Syndactyly type 1, Castilla type
295189	Syndactyly type 1, Lueken type
295191	Syndactyly type 1, Montagu type
295187	Syndactyly type 1, Weidenreich type
295187	Syndactyly type 1a
295189	Syndactyly type 1b
295191	Syndactyly type 1c
295193	Syndactyly type 1d
93403	Syndactyly type 2
93404	Syndactyly type 3
93405	Syndactyly type 4
93406	Syndactyly type 5
295012	Syndactyly type 6
3258	Syndactyly type 7
2498	Syndactyly type 8
157801	Syndactyly type 9
157801	Syndactyly, Malik-Percin type
295012	Syndactyly, mitten type
3259	Syndactyly-polydactyly-ear lobe syndrome
84064	Syndromatic diarrhea
109	Syndrome de Myhre-Rikey-Smith
52	Syndromic bile duct paucity
261619	Syndromic bile duct paucity due to a JAG1 point mutation
261629	Syndromic bile duct paucity due to a NOTCH2 point mutation
261600	Syndromic bile duct paucity due to monosomy 20p12
84064	Syndromic diarrhea
84064	Syndromic diarrhea/Tricho-hepato-enteric syndrome
77298	Syndromic microphthalmia type 3
85275	Syndromic microphthalmia type 4
178364	Syndromic microphthalmia type 5
139471	Syndromic microphthalmia type 6
2556	Syndromic microphthalmia type 7
3434	Syndromic microphthalmia type 8
2470	Syndromic microphthalmia type 9
77299	Syndromic microphthalmia type 10
178364	Syndromic microphthalmia/anophthalmia due to OTX2 mutation
280679	Syndromic Moyamoya disease
228426	Syndromic multisystem autoimmune disease due to Itch deficiency
137905	Syndromic optic nerve hypoplasia
98606	Syndromic orbital border hypoplasia
281090	Syndromic recessive X-linked ichthyosis
281090	Syndromic RXLI
281090	Syndromic X-linked ichthyosis
85274	Syndromic X-linked intellectual deficit 7
85279	Syndromic X-linked intellectual deficit due to JARID1C mutation
85295	Syndromic X-linked intellectual deficit type 10
85286	Syndromic X-linked intellectual deficit type 11
319332	SYNE1-related AMC
319332	SYNE1-related arthrogryposis multiplex congenita
3263	Syngnathia - cleft palate
3262	Syngnathia multiple anomalies
3268	Synostosis - microcephaly - scoliosis
35098	Synostotic plagiocephaly
3273	Synovial sarcoma
793	Synovitis-acne-pustulosis-hyperostosis-osteitis syndrome
93403	Synpolydactyly
295195	Synpolydactyly type 1
295197	Synpolydactyly type 2
295199	Synpolydactyly type 3
295197	Synpolydactyly, Debeer type
295199	Synpolydactyly, Malik type
295195	Synpolydactyly, Vordingborg type
3275	Synspondylism
93926	Syntelencephaly
840	Syringocystadenoma papilliferum
3280	Syringomyelia
314701	Systemic AL amyloidosis
2039	Systemic arteriovenous fistula
188	Systemic capillary leak syndrome
314701	Systemic immunoglobulinic amyloidosis
2467	Systemic mastocytosis
98849	Systemic mastocytosis with an associated clonal hematologic non-mast cell lineage disease
90069	Systemic monochloroacetate poisoning
85414	Systemic polyarthritis
90291	Systemic scleroderma
90291	Systemic sclerosis
220407	Systemic sclerosis sine scleroderma
85414	Systemic-onset juvenile idiopathic arthritis
1350	Tabatznik syndrome
3384	TAC
3284	Tachycardia - hypertension - microphthalmos - hyperglycinuria
99974	TACI-related selective deficiency of IgA
567	Takao syndrome
2905	Takatsuki syndrome
3287	Takayasu arteritis
66529	Takotsubo cardiomyopathy
66529	Tako-Tsubo cardiomyopathy
66529	Takotsubo syndrome
66529	Tako-tsubo syndrome
101028	TALDO

ORPHA Number	Disease name
2886	Talipes equinovarus - atrial septal defect - Robin sequence - Persistence of the left superior vena cava
99861	T-ALL
329191	Tall stature - scoliosis - macrodactyly of the great toes
329191	Tall stature - scoliosis - macrodactyly of the halluces
50809	Talo-patello-scaphoid osteolysis
31150	Tangier disease
180	Tapetochoroidal dystrophy
98839	Tappeiner-Pfleger disease
3320	TAR syndrome
609	Tardive tibial muscular dystrophy
2886	TARP syndrome
99170	Tarsal kink syndrome
1412	Tarsal-carpal coalition syndrome
371	Tarui disease
163654	Tattoo dysplasia
2731	Taurodontia - absent teeth - sparse hair
3289	Taurodontism
101042	Taussig-Bing syndrome
453	Tay syndrome
669	Taybi syndrome
2636	Taybi-Linder syndrome
845	Tay-Sachs disease
309192	Tay-Sachs disease, B variant, adult form
309178	Tay-Sachs disease, B variant, infantile form
309185	Tay-Sachs disease, B variant, juvenile form
309239	Tay-Sachs disease, B1 variant
169160	T-B+ SCID due to CD3delta/CD3epsilon/ CD3zeta
169157	T-B+ SCID due to CD45 deficiency
276	T-B+ SCID due to gamma chain deficiency
169154	T-B+ SCID due to IL-7Ralpha deficiency
35078	T-B+ SCID due to JAK3 deficiency
169160	T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta
169157	T-B+ severe combined immunodeficiency due to CD45 deficiency
276	T-B+ severe combined immunodeficiency due to gamma chain deficiency
169154	T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency
35078	T-B+ severe combined immunodeficiency due to JAK3 deficiency
276	T-B+ severe combined immunodeficiency, X-linked
98960	TBCD
86871	T-cell chronic lymphocytic leukemia
324294	T-cell immunodeficiency due to RHOH deficiency
324294	T-cell immunodeficiency with epidermodysplasia verruciformis
86872	T-cell large granular lymphocyte leukemia
86872	T-cell LGL leukemia
86886	T-cell lymphoma, AILD type
86871	T-cell prolymphocytic leukemia
300857	T-cell/histiocyte rich large B cell lymphoma
103918	TCP
3352	TDO syndrome
1519	Teebi hypertelorism syndrome
1519	Teebi syndrome
2432	Teebi-Al Saleh-Hassoon syndrome
1094	Teebi-Kaurah syndrome
1974	Teebi-Naguib-Alawadi syndrome
3291	Teebi-Shaltout syndrome
3292	Tel Hashomer camptodactyly syndrome
284227	Telangiectasia - erythrocytosis - monoclonal gammopathy - perinephric-fluid collections - intrapulmonary shunting
90389	Telangiectasia macularis eruptiva perstans
3293	Telecanthus - hypertelorism - strabismus - pes cavus
2885	Telfer-Sugar-Jaeger syndrome
1590	Telomeric 13q deletion
1596	Telomeric 15q deletion
1597	Telomeric 17q deletion
1627	Telomeric 5q deletion
36367	Telomeric deletion 1q
280	Telomeric deletion 4p
96145	Telomeric deletion 4q
96126	Telomeric deletion 7p
1636	Telomeric deletion 7q36
1642	Telomeric deletion 9p
1580	Telomeric deletion 10p
96148	Telomeric deletion 10q
2308	Telomeric deletion 11q
96149	Telomeric deletion 12q
96150	Telomeric deletion 14q
531	Telomeric deletion 17p
96129	Telomeric deletion 19p
96152	Telomeric deletion 20q
96069	Telomeric duplication 1p36
96070	Telomeric duplication 2p
96094	Telomeric duplication 2q
96071	Telomeric duplication 3p
96072	Telomeric duplication 4p
96096	Telomeric duplication 4q
96097	Telomeric duplication 5q
1745	Telomeric duplication 6p
96098	Telomeric duplication 6q
96074	Telomeric duplication 7p
96100	Telomeric duplication 8q
96101	Telomeric duplication 9q
96102	Telomeric duplication 10q
96103	Telomeric duplication 11q
96105	Telomeric duplication 13q
1705	Telomeric duplication 14q
1707	Telomeric duplication 15q
96078	Telomeric duplication 16p
96106	Telomeric duplication 16q
3379	Telomeric duplication 17q
1716	Telomeric duplication 18q
1717	Telomeric duplication 19q
96107	Telomeric duplication 20q
96109	Telomeric duplication 22q
1762	Telomeric duplication Xq
1620	Telomeric monosomy 3p
75565	TEMF
352737	Temperature-sensitive oculocutaneous albinism type 1
284227	TEMPI syndrome
397	Temporal arteritis
1777	Temtamy syndrome
1777	Temtamy-Shalash syndrome
3398	TEN
137834	Ter Haar syndrome
252018	Teratoma of the central nervous system
141107	Teratoma of the nasopharynx
88630	Terminal osseous dysplasia - pigmentary defects
93937	Terminal transverse defects of arm
141242	Tessier number 1 cleft
141258	Tessier number 4 facial cleft
141261	Tessier number 5 facial cleft
141265	Tessier number 6 facial cleft
325124	Testicular agenesis
754	Testicular feminization syndrome
983	Testicular regression syndrome
842	Testicular seminoma
3000	Testotoxicosis
3299	Tetanus
9	Tetra X
294971	Tetraamelia
3301	Tetraamelia - multiple malformations
199310	Tetragametic chimerism
293284	Tetrahydrobiopterin-responsive HPA/PKU
293284	Tetrahydrobiopterin-responsive hyperphenylalaninemia/phenylketonuria
3303	Tetralogy of Fallot
2564	Tetramelic monodactyly
3305	Tetraploidy

ORPHA Number	Disease name
3309	Tetrasomy 5p
3310	Tetrasomy 9p
289522	Tetrasomy 11q24.1
884	Tetrasomy 12p
314588	Tetrasomy 15(q25-qter)
314588	Tetrasomy 15q26
3307	Tetrasomy 18p
96055	Tetrasomy 21
9	Tetrasomy X
140917	Teunissen-Cremers syndrome
225123	TFR2-related hemochromatosis
216729	TGA with cardiac malformation
99042	TGA with coarctation
1780	Thakker-Donnai syndrome
3312	Thalidomide embryopathy
2655	Thanatophoric dwarfism
93274	Thanatophoric dwarfism - cloverleaf skull
1860	Thanatophoric dwarfism type I
93274	Thanatophoric dwarfism type II
2655	Thanatophoric dysplasia
93275	Thanatophoric dysplasia, Glasgow variant
99917	Theca (steroid-producing) cell cancer, not further specified
99917	Theca steroid-producing cell malignant tumor, not further specified
88633	Theodore's superior limbic keratoconjunctivitis
88633	Theodore's syndrome
199348	Thiamine-responsive encephalopathy
49827	Thiamine-responsive megaloblastic anemia syndrome
49827	Thiamine-responsive megaloblastic anemia with diabetes mellitus and sensorineural deafness
268184	Thiamin-responsive BCKD deficiency
268184	Thiamin-responsive branched-chain ketoacid dehydrogenase deficiency
268184	Thiamin-responsive branched-chain ketoaciduria
268184	Thiamin-responsive leucinosis
268184	Thiamin-responsive maple syrup urine disease
268184	Thiamin-responsive MSUD
2405	Thickened earlobes - conductive deafness
98960	Thiel-Behnke corneal dystrophy
3313	Thiele syndrome
3314	Thiemann disease, familial form
3235	Thies-Reis syndrome
1506	Thin ribs - tubular bones - dysmorphism
166424	Thinking seizures
2981	Thiolase deficiency
3315	Thiopurine S-methyltransferase deficiency
141030	Third branchial cleft anomaly
141030	Third branchial cleft cyst
141030	Third branchial cleft fistula
3316	Thomas syndrome
276241	Thomas type SCA3
2547	Thomas-Jewett-Raines syndrome
2031	Thompson-Baraitser syndrome
614	Thomsen and Becker disease
2866	Thong-Douglas-Ferrante syndrome
1861	Thoracic dysplasia-hydrocephalus syndrome
97330	Thoracic outlet compression syndrome
97330	Thoracic outlet syndrome
1759	Thoraco-abdominal enteric duplication
1335	Thoraco-abdominal syndrome
3317	Thoracolaryngopelvic dysplasia
268384	Thoracolumbosacral spina bifida aperta
268752	Thoracolumbosacral spina bifida cystica
1803	Thoracomelic dysplasia
99832	THR resistance syndrome
300857	THRLBCL
36258	Thromboangiitis obliterans
3204	Thrombocytopathy - asplenia - miosis
3320	Thrombocytopenia - absent radius
3323	Thrombocytopenia - Robin sequence
67044	Thrombocytopenia with congenital dyserythropoietic anemia
3002	Thrombocytopenic purpura, autoimmune
54057	Thrombotic thrombocytopenic purpura
3329	TH-SHFM
2251	Thumb deformity - alopecia - pigmentation anomaly
294988	Thumb hypodactyly
295112	Thumb hypodactyly, bilateral
295110	Thumb hypodactyly, unilateral
294988	Thumb oligodactyly
295112	Thumb oligodactyly, bilateral
295110	Thumb oligodactyly, unilateral
1078	Thumb stiffness - brachydactyly - intellectual deficit
2919	Thurston syndrome
83471	Thymic aplasia
99868	Thymic carcinoma
97289	Thymic endocrine tumor
3398	Thymic epithelial neoplasm
3398	Thymic epithelial tumor
99869	Thymic neuroendocrine carcinoma
97289	Thymic neuroendocrine tumor
3326	Thymic-renal-anal-lung dysplasia
99867	Thymoma
263310	Thymoma type A
263324	Thymoma type AB
263317	Thymoma type B
169105	Thymoma-immunodeficiency
3327	Thyrocerebrorenal syndrome
489	Thyroglossal duct cyst
489	Thyroglossal tract cyst
95716	Thyroid dyshormonogenesis
95712	Thyroid ectopia
95719	Thyroid hemiagenesis
95720	Thyroid hypoplasia
97285	Thyroid lymphoma
95721	Thyroid pyramidal lobe
91347	Thyroid stimulating hormone-secreting pituitary adenoma
2091	Thyroid-renal-digital anomalies
79102	Thyrotoxic hypokalemic periodic paralysis
79102	Thyrotoxic periodic paralysis
91347	Thyrotroph adenoma
2768	Tibia vara Blount
3329	Tibial aplasia - ectrodactyly
93322	Tibial hemimelia
3329	Tibial hemimelia with split hand/foot malformation
295079	Tibial hemimelia, bilateral
295077	Tibial hemimelia, unilateral
3329	Tibial hemimelia-ectrodactyly syndrome
295079	Tibial longitudinal meomelia, bilateral
295077	Tibial longitudinal meomelia, unilateral
609	Tibial muscular dystrophy
295028	Tibio-fibular fusion
295028	Tibio-fibular synostosis
294981	Tibiofibular terminal transverse meromelia
295099	Tibiofibular terminal transverse meromelia, bilateral
295097	Tibiofibular terminal transverse meromelia, unilateral
221091	Tic douloureux
297	Tick-borne encephalitis
42665	Tietz syndrome
65283	Timothy syndrome
91500	TINU syndrome
352540	TIO
86872	T-LGL
228407	TMCO1 defect syndrome
314667	TMEM165-CDG syndrome
99886	TNDM
32960	TNF receptor 1 associated periodic syndrome
295118	Toes absent, bilateral
295116	Toes absent, unilateral
64686	Tolosa-Hunt syndrome
1920	Toluene antenatal infection

ORPHA Number	Disease name
1920	Toluene embryopathy
640	Tomaculous neuropathy
3336	Tomé-Brunet-Fardeau syndrome
1547	Tonoki-Ohura-Niikawa syndrome
2228	Tooth and nail syndrome
3460	Torg-Winchester syndrome
1827	Toriello syndrome
3338	Toriello-Carey syndrome
79347	Toriello-Higgins-Miller syndrome
3339	Toriello-Lacassie-Droste syndrome
3340	Torres-Aybar syndrome
51084	Torsade-de-pointes syndrome with short coupling interval
3341	Torticollis - keloids - cryptorchidism - renal dysplasia
75326	Tortuosity of retinal arteries
97330	TOS
294971	Total amelia
49382	Total color blindness
98994	Total congenital cataract
180126	Total septate uterus
268377	Total spina bifida aperta
268748	Total spina bifida cystica
2796	Touraine-Solente-Gole syndrome
857	Townes-Brocks syndrome
95455	Toxic epidermal necrolysis
95455	Toxic epidermolysis
279894	Toxic maculopathy due to antimalarial drugs
227972	Toxic oil syndrome
206619	Toxic or/and iatrogenic neuropathy
293173	Toxic pustuloderma
3343	Toxocariasis
858	Toxoplasma embryofetopathy
858	Toxoplasma embryopathy
93164	TPHA
86871	T-PLL
2950	TPT-PS syndrome
3346	Tracheal agenesis
3347	Tracheobronchomegaly
3348	Tracheobronchopathia osteochondroplastica
2042	Tracheo-esophageal fistula - hypospadias
1250	Tracheo-esophageal fistula - symphalangism
3348	Tracheopathia osteoplastica
3052	Tranebjaerg-Svejgaard syndrome
101028	Transaldolase deficiency
859	Transcobalamin II deficiency
199247	Transcortin deficiency
495	Transgrediens et progrediens palmoplantar keratoderma
495	Transgrediens et progrediens PPK
87503	Transgrediens palmoplantar keratoderma of Siemens
98871	Transient acquired pure red cell aplasia
79411	Transient bullous dermolysis of the newborn
178045	Transient congenital hypothyroidism
98871	Transient erythroblastopenia of childhood
2312	Transient familial neonatal hyperbilirubinemia
289877	Transient hyperammonemia of the newborn
169139	Transient hypogammaglobulinemia of infancy
66529	Transient left ventricular apical ballooning syndrome
1153	Transient neonatal arthrogryposis
280615	Transient neonatal cyanosis and anemia due to Toms River Hemoglobin
99886	Transient neonatal diabetes mellitus
329942	Transient neonatal glutaric aciduria type 2
329942	Transient neonatal MAD deficiency
329942	Transient neonatal MADD
329942	Transient neonatal multiple acyl-CoA dehydrogenase deficiency
93164	Transient pseudohypoaldosteronism
3402	Transient tyrosinemia of the neonate
3402	Transient tyrosinemia of the newborn
213746	Transitional cell carcinoma of the corpus uteri
280224	Transitional PMD
319308	Translocation carcinoma
319308	Translocation renal cell carcinoma
56970	Transmissible spongiform encephalopathy
85451	Transthyretin amyloid cardiopathy
85447	Transthyretin amyloid neuropathy
85447	Transthyretin amyloid polyneuropathy
85451	Transthyretin-related familial amyloid cardiomyopathy
2486	Transverse limb deficiency - hemangioma
180160	Transverse vaginal septum
32960	TRAPS syndrome
861	Treacher-Collins syndrome
3350	Tremor - nystagmus - duodenal ulcer
64694	Trench fever
1822	Trevor disease
85170	Triangular tibia - fibular aplasia
1463	Triatrial heart
863	Trichinellosis
863	Trichinosis
3351	Trichodental syndrome
3352	Tricho-dento-osseous syndrome
3360	Trichodermal syndrome - intellectual deficit
3353	Trichodermodysplasia - dental alterations
79129	Trichodysplasia - amelogenesis imperfecta
3361	Trichodysplasia - xeroderma
228379	Trichodysplasia spinulosa
864	Trichofolliculoma
84064	Tricho-hepato-enteric syndrome
3362	Trichomegaly - cataract - hereditary spherocytosis
3363	Trichomegaly - retina pigmentary degeneration - dwarfism
3354	Tricho-oculo-dermo-vertebral syndrome
3355	Tricho-odonto-onychial dysplasia
3355	Tricho-odonto-onychial dysplasia with bone deficiency in frontoparietal region
3357	Tricho-odonto-onychodysplasia - dominant syndactyly
565	Trichopoliodystrophy
1264	Tricho-retino-dento-digital syndrome
77258	Trichorhinophalangeal syndrome type 1 and 3
502	Trichorhinophalangeal syndrome type 2
33364	Trichothiodystrophy
75789	Trichothiodystrophy - osteosclerosis
670	Trichothiodystrophy - sun sensitivity
1245	Trichothiodystrophy type D
453	Trichothiodystrophy type E
670	Trichothiodystrophy type F
2739	Trichothiodystrophy type G
453	Trichothiodystrophy with congenital ichthyosis
1209	Tricuspid atresia
95457	Tricuspid valve agenesis
95458	Tricuspid valve prolapse
157843	Trigeminal autonomic cephalalgia
221091	Trigeminal neuralgia
3368	Trigonocephaly - bifid nose - acral anomalies
3365	Trigonocephaly - broad thumbs
3369	Trigonocephaly - short stature - developmental delay
1308	Trigonocephaly C syndrome
1913	Trimethadione antenatal infection
35056	Trimethylaminuria
3374	Triopia
868	Triose phosphate-isomerase deficiency
2950	Triphalangeal thumb - polysyndactyly syndrome
2947	Triphalangeal thumbs - brachyectrodactyly
3133	Triphalangeal thumbs - dislocation of patella
869	Triple A syndrome
415	Triple H syndrome
3375	Triple X

ORPHA Number	Disease name
3376	Triploidy
3375	Triplo-X
3377	Trismus - pseudocamptodactyly
96069	Trisomy 1pter
261344	Trisomy 1q
250994	Trisomy 1q21.1
96070	Trisomy 2pter
313947	Trisomy 2q23.1
294026	Trisomy 2q31.1
96094	Trisomy 2qter
96071	Trisomy 3pter
96095	Trisomy 3q26
251038	Trisomy 3q29
1738	Trisomy 4p
96072	Trisomy 4pter
1739	Trisomy 4q
96096	Trisomy 4qter
1742	Trisomy 5p
329802	Trisomy 5p13
228415	Trisomy 5q35
96097	Trisomy 5qter
1745	Trisomy 6pter
96098	Trisomy 6qter
314034	Trisomy 7p22.1
96074	Trisomy 7pter
96121	Trisomy 7q11.23
264450	Trisomy 8p
251076	Trisomy 8p23.1
1752	Trisomy 8q
228399	Trisomy 8q12
96100	Trisomy 8qter
236	Trisomy 9p
96101	Trisomy 9qter
171929	Trisomy 10p
276422	Trisomy 10q22.3q23.3
96102	Trisomy 10qter
300305	Trisomy 11p15.4
96103	Trisomy 11qter
1699	Trisomy 12p
3378	Trisomy 13
96105	Trisomy 13qter
261229	Trisomy 14q11.2
1705	Trisomy 14qter
238446	Trisomy 15q11q13
238446	Trisomy 15q11-q13
1707	Trisomy 15qter
261204	Trisomy 16p11.2p12.2
261243	Trisomy 16p13.11
96078	Trisomy 16pter
96106	Trisomy 16qter
261290	Trisomy 17p
1713	Trisomy 17p11.2
217385	Trisomy 17p13.3
139474	Trisomy 17q11.2
261272	Trisomy 17q12
217340	Trisomy 17q21.31
3379	Trisomy 17qter
3380	Trisomy 18
1715	Trisomy 18p
1716	Trisomy 18qter
1717	Trisomy 19qter
261318	Trisomy 20p
96107	Trisomy 20qter
870	Trisomy 21
1727	Trisomy 22q11.2
96109	Trisomy 22qter
3375	Trisomy X
217377	Trisomy Xp11.22-p11.23
261483	Trisomy Xq27.3q28
261483	Trisomy Xq27.3-q28
1762	Trisomy Xq28
88629	Tritan colour blindness
88629	Tritanopia
49827	TRMA
1349	tRNA-LYS-related cardiomyopathy - hearing loss
1863	Trochlear dysplasia
99664	Trochlear nerve palsy
103918	Tropical calcific chronic pancreatitis
75565	Tropical endomyocardial fibrosis
103918	Tropical pancreatitis
764	Tropical pyomyositis
289326	Tropical spastic paraparesis
101000	Troyer syndrome
983	TRS
313906	True congenital pancreatic cyst
2138	True hermaphroditism
2512	True microcephaly
180074	True unicornuate uterus
100072	True vascular thoracic outlet syndrome
3357	Trueb-Burg-Bottani syndrome
93956	Truncal dystonia
3384	Truncus arteriosus
228379	TS
352737	TS OCA type 1
3173	Tsao-Ellingson syndrome
91347	TSH-oma
91347	TSH-secreting pituitary adenoma
289326	TSP
3268	Tsukahara syndrome
3387	Tsukahara-Kajii syndrome
83317	Tsutsugamushi disease
83317	Tsutsugamushi fever
54057	TTP
85447	TTR amyloid neuropathy
85451	TTR-related amyloid cardiomyopathy
85451	TTR-related cardiac amyloidosis
3389	Tuberculosis
805	Tuberous sclerosis
805	Tuberous sclerosis complex
88924	Tuberous sclerosis/polycystic kidney disease contiguous gene syndrome
2593	Tubular aggregate myopathy
100048	Tubular duplication of the esophagus
73224	Tubular renal disease - cardiomyopathy
319325	Tubulocystic carcinoma
91500	Tubulointerstitial nephritis and uveitis syndrome
2997	Tucker syndrome
1063	Tufted angioma
92050	Tufting enteropathy
3392	Tularemia
640	Tulip-bulb digger's palsy
32960	Tumor necrosis factor receptor 1 associated periodic syndrome
289539	Tumor susceptibility linked to germline BAP1 mutations
53715	Tumoral calcinosis
352540	Tumor-induced osteomalacia
879	Tungiasis
3225	Tungland-Bellman syndrome
99053	Tunnel subaortic stenosis
211	Turban tumor syndrome
99818	Turcot syndrome with polyposis
881	Turner syndrome
99413	Turner syndrome due to structural X chromosome anomalies
2614	Turner-Kieser syndrome
63440	Turricephaly
79153	Twenty-nail dystrophy
95431	Twin to twin transfusion syndrome
1461	Twisted atrioventricular connections
2889	Twisted hair
2198	Tylosis - oesophageal carcinoma
93554	Type II mixed cryoglobulinemia
99745	Typhoid
99745	Typhoid fever
99745	Typhoidal salmonellosis
90038	Typical hemolytic uremic syndrome
90038	Typical HUS
171436	Typical nemaline myopathy
158766	Typical urticaria pigmentosa

ORPHA Number	Disease name
79431	Tyrosinase-negative oculocutaneous albinism
101150	Tyrosine hydroxylase deficiency
69723	Tyrosinemia due to 4-hydroxyphenylpyruvate dioxygenase deficiency
69723	Tyrosinemia due to 4-hydroxyphenylpyruvic acid oxidase deficiency
69723	Tyrosinemia due to HPD deficiency
28378	Tyrosinemia due to TAT deficiency
28378	Tyrosinemia due to tyrosine aminotransferase deficiency
882	Tyrosinemia type 1
28378	Tyrosinemia type 2
69723	Tyrosinemia type 3
28378	Tyrosinemia type II
69723	Tyrosinemia type III
75840	UCMD
90002	UCTD
79238	UDP-galactose-4-epimerase deficiency
178315	UES
205	UGT deficiency
79234	UGT deficiency type 1
79235	UGT deficiency type 2
3403	Uhl anomaly
2032	UIP
3404	Ulbright-Hodes syndrome
308	ULD
3406	Ulerythema ophryogenesis
320	Ulick syndrome
75840	Ullrich disease
2497	Ulna hypoplasia
2249	Ulna hypoplasia - intellectual deficit
1837	Ulna metaphyseal dysplasia syndrome
93320	Ulnar clubhand
93320	Ulnar hemimelia
295073	Ulnar hemimelia, bilateral
295075	Ulnar hemimelia, unilateral
1122	Ulnar hypoplasia - lobster-claw deformity of feet
295073	Ulnar longitudinal meromelia, bilateral
295075	Ulnar longitudinal meromelia, unilateral
52056	Ulnar/fibula ray defect - brachydactyly
3138	Ulnar-mammary syndrome
3138	Ulnar-mammary syndrome of Pallister
3405	Umbilical cord ulceration - intestinal atresia
209886	UMOD-associated familial juvenile hyperuricemic nephropathy
209886	UMOD-associated FJHN
35120	UMPH1 deficiency
3138	UMS

ORPHA Number	Disease name
86830	Unclassified chronic myeloproliferative disease
97569	Unclassified glomerulonephritis
104078	Unclassified intestinal pseudoobstruction
91140	Unclassified juvenile idiopathic arthritis
90345	Unclassified metaphyseal chondrodysplasia
98825	Unclassified mixed myelodysplastic/ myeloproliferatic syndrome
98827	Unclassified myelodysplastic syndrome
98825	Unclassified myelodysplastic/ myeloproliferative disease
1410	Uncombable hair syndrome
103920	Undetermined colitis
213721	Undifferentiated carcinoma of the corpus uteri
90002	Undifferentiated connective tissue syndrome
178315	Undifferentiated embryonal sarcoma of the liver
86830	Undifferentiated myeloproliferative disease
178315	Undifferentiated sarcoma of the liver
319658	Unexplained intellectual deficit
83468	Unicameral bone cyst
180114	Unicervical bicornuate uterus
180079	Unicornuate uterus with rudimentary horn
180074	Unicornuate uterus without rudimentary horn
137917	Unilateral choanal atresia
93176	Unilateral congenital megacalycosis
268947	Unilateral focal polymicrogyria
101071	Unilateral hemispheric polymicrogyria
99802	Unilateral megalencephaly
97363	Unilateral multicystic renal dysplasia
141132	Unilateral or bilateral and asymmetric otomandibular dysplasia
268943	Unilateral polymicrogyria
295148	Unilateral PPD2
93100	Unilateral renal agenesis
93172	Unilateral renal dysplasia
97361	Unilateral renal hypoplasia
357034	Unilateral retinoblastoma
295012	Unilateral syndactyly of digits 2-5
95483	Univentricular cardiopathy
1464	Univentricular heart
99069	Univentricular heart with single atrio-ventricular valve
79146	Universal melanosis
84096	Unknown leukodystrophy
99104	Unroofed coronary sinus
308	Unverricht-Lundborg disease
251009	UPD(1)mat

ORPHA Number	Disease name
251004	UPD(1)pat
96179	UPD(2)mat
96180	UPD(4)mat
96190	UPD(5)pat
96181	UPD(6)mat
96191	UPD(6)pat
96182	UPD(7)mat
96192	UPD(7)pat
96183	UPD(9)mat
231147	UPD(11)mat
96193	UPD(11)pat
97678	UPD(13)mat
99324	UPD(13)pat
96184	UPD(14)mat
96334	UPD(14)pat
98754	UPD(15)mat
98795	UPD(15)pat
96185	UPD(16)mat
96186	UPD(20)mat
96194	UPD(20)pat
96187	UPD(21)mat
96195	UPD(21)pat
96188	UPD(22)mat
261519	UPD(X)mat
261524	UPD(X)pat
3408	Upington disease
2489	Upper limb defect - eye and ear abnormalities
295049	Upper limb hypertrophy
2497	Upper limb mesomelic dysplasia
268740	Upper thoracic spina bifida aperta
268770	Upper thoracic spina bifida cystica
93583	Upshaw-Schulman syndrome
488	Urachal cyst
530	Urbach-Wiethe disease
221145	Urban-Rifkin-Davis syndrome
3409	Urban-Rogers-Meyer syndrome
1839	Urban-Schosser-Spohn syndrome
94059	Uremic pruritus
2970	Urethral obstruction sequence
35120	Uridine 5' monophosphate hydrolase deficiency
79238	Uridine diphosphate galactose-4-epimerase deficiency
30	Uridine monophosphate synthetase deficiency
210128	Urocanic aciduria
2704	Urofacial syndrome
83628	Urorectal septum malformation sequence
98606	Urrets-Zavalia syndrome
575	Urticaria - deafness - amyloidosis

ORPHA Number	Disease name
79457	Urticaria pigmentosa
886	USH
231169	USH1
231178	USH2
231183	USH3
886	Usher syndrome
231169	Usher syndrome type 1
231178	Usher syndrome type 2
231183	Usher syndrome type 3
2032	Usual interstitial pneumonia
180145	Uterine cervical aplasia and agenesis
180139	Uterine hypoplasia
180118	Uterus arcuatus
180118	Uterus cordiformis
1473	Uveal coloboma - cleft lip and palate - intellectual deficit
39044	Uveal melanoma
3437	Uveomenigitic syndrome
178338	UV-sensitive syndrome
99771	Uvular cleft
887	VACTERL association
3412	VACTERL with hydrocephalus
887	VACTERL/VATER association
25980	Vacuolar myopathy
2478	Vacuolating megalencephalic leukoencephalopathy with subcortical cysts
65681	Vaginal atresia
180247	Vaginal carcinoma
206489	Vaginal germ cell cancer
206489	Vaginal germ cell malignant tumor
180247	Vaginal malignant epithelial tumor
228123	Valley fever
1906	Valproate antenatal infection
1906	Valproic acid antenatal infection
99054	Valvular pulmonary stenosis
1548	Van Benthem-Driessen-Hanveld syndrome
1789	Van Biervliet-Hendrickx-van Ertbruggen syndrome
2806	Van Bogaert disease
2806	Van Bogaert encephalitis
3416	Van Buchem disease
3417	Van den Bosch syndrome
2460	Van den Ende-Gupta syndrome
216796	Van der Hoeve syndrome
2478	Van der Knaap syndrome
888	Van der Woude syndrome
314679	Van Maldergem syndrome
3419	Van Regemorter-Pierquin-Vamos syndrome
73	Vanishing bone disease
983	Vanishing testes syndrome
983	Vanishing testis syndrome

ORPHA Number	Disease name
729	Vaquez disease
2754	Váradi syndrome
2754	Váradi-Papp syndrome
79253	Variant phenylketonuria
79253	Variant PKU
291	Varicella virus antenatal infection
79473	Variegate porphyria
3160	Vascular disruption sequence
2452	Vascular malposition
353356	Vasoproliferative tumor of the ocular fundus
353356	Vasoproliferative tumor of the retina
3423	Vasquez-Hurst-Sotos syndrome
85128	Västerbotten dystrophy
887	VATER association
52047	Vater-like syndrome with pulmonary hypertension, abnormal ears and growth deficiency
228379	VATS
898	VCAN-related vitreoretinopathy
289157	VDDI
93160	VDDR II
289157	VDDR-I
2460	VDEGS
93160	VDRR II
1053	Vein of Galen aneurysm
1053	Vein of Galen arteriovenous malformations
567	Velocardiofacial syndrome
3424	Velo-facial-skeletal syndrome
29207	Venereal arthritis
319234	Venezuelan hemorrhagic fever
357131	Venous cervical rib syndrome
357131	Venous costoclavicular syndrome
357131	Venous hyperabduction syndrome
83454	Venous malformations with glomus cells
357131	Venous scalenus anticus syndrome
357131	Venous thoracic outlet compression syndrome
357131	Venous thoracic outlet syndrome
357131	Venous TOS
3201	Ventricular extrasystoles with syncopal episodes - perodactyly - Robin sequence
216694	Ventricular inversion
1480	Ventricular septal defect
99094	Ventricular septal defect with aortic insufficiency
216694	Ventriculoarterial and atrioventricular discordance
2899	Verloes-Bourguignon syndrome
2496	Verloes-David syndrome
50817	Verloes-Deprez syndrome
2983	Verloes-Gillerot-Fryns syndrome

ORPHA Number	Disease name
2551	Verloes-Van Maldergem-de Marneffe syndrome
3429	Verloove Vanhorick-Brubakk syndrome
70476	Vernal keratoconjunctivitis
97282	Verner-Morrison syndrome
79467	Verrucous nevus
26793	Very long chain acyl-CoA dehydrogenase deficiency
252175	Vestibular schwannoma
99663	Vestibular torticollis
892	VHL
1493	Vici syndrome
3433	Viljoen-Kallis-Voges syndrome
3434	Viljoen-Smart syndrome
97282	VIPoma
97282	VIP-secreting tumor
341	Viral hemorrhagic fever
180176	Virginal breast hypertrophy
99916	Virilizing ovarian tumor
228379	Virus-associated trichodysplasia spinulosa
280068	Visceral calciphylaxis
450	Visceral heterotaxy
1876	Visceral myopathy - familial external ophthalmoplegia
73246	Visceral neuropathy - brain anomalies - facial dysmorphism - developmental delay
353344	Visible and exudative idiopathic juxtafoveolar retinal telangiectasis
28	Vitamin B12-responsive methylmalonic acidemia
79310	Vitamin B12-responsive methylmalonic acidemia type cblA
79311	Vitamin B12-responsive methylmalonic acidemia type cblB
308442	Vitamin B12-responsive methylmalonic acidemia, type cblDv2
28	Vitamin B12-responsive methylmalonic aciduria
79310	Vitamin B12-responsive methylmalonic aciduria type cblA
79311	Vitamin B12-responsive methylmalonic aciduria, type cblB
308442	Vitamin B12-responsive methylmalonic aciduria, type cblDv2
27	Vitamin B12-unresponsive methylmalonic acidemia
79312	Vitamin B12-unresponsive methylmalonic acidemia type mut-
289916	Vitamin B12-unresponsive methylmalonic acidemia type mut0
27	Vitamin B12-unresponsive methylmalonic aciduria
79312	Vitamin B12-unresponsive methylmalonic aciduria type mut-
289916	Vitamin B12-unresponsive methylmalonic aciduria type mut0

ORPHA Number	Disease name
3006	Vitamin B6-responsive seizures
289157	Vitamin D dependent rickets type I
289157	Vitamin D-dependency type I
93160	Vitamin D-dependent rickets type II
93160	Vitamin D-resistant rickets type II
1914	Vitamin K antagonists embryofetopathy
1243	Vitelliform macular dystrophy
247871	Vitiligo-associated autoimmune disease
898	Vitreoretinal degeneration, Wagner type
26793	VLCAD deficiency
26793	VLCADD
2451	VMCM
83454	VMGLOM
79124	VODI syndrome
3437	Vogt-Koyanagi-Harada disease
494	Vohwinkel syndrome
79395	Vohwinkel syndrome - ichthyosis
2427	Volcke-Soekarman syndrome
35737	Volubilis syndrome
364	Von Gierke disease
98941	Von Hippel anomaly
892	Von Hippel-Lindau disease
892	Von Hippel-Lindau syndrome
238557	Von Hippel-Lindau-dependent polycythemia
636	Von Recklinghausen disease
3439	Von Voss-Cherstvoy syndrome
903	Von Willebrand disease
166078	Von Willebrand disease type 1
166081	Von Willebrand disease type 2
166084	Von Willebrand disease type 2A
166087	Von Willebrand disease type 2B
166090	Von Willebrand disease type 2M
166093	Von Willebrand disease type 2N
166096	Von Willebrand disease type 3
52530	Von Willebrand disease, platelet type
353356	VPTR
1480	VSD
99094	VSD with aortic insufficiency
357131	VTOS
137583	Vulvar intraepithelial neoplasia
137583	Vulvar intraepithelial tumor
83453	Vulvovaginal gingival syndrome
206492	Vulvovaginal rhabdomyosarcoma
53696	Vuopala disease
888	VWS
2804	W syndrome
2180	Waaler-Aarskog syndrome
1106	Waardenburg anophthalmia syndrome
3440	Waardenburg syndrome
894	Waardenburg syndrome type 1
895	Waardenburg syndrome type 2
352740	Waardenburg syndrome type 2 with ocular albinism
896	Waardenburg syndrome type 3
897	Waardenburg syndrome type 4
896	Waardenburg syndrome with limb anomalies
897	Waardenburg-Hirschsprung syndrome
98960	Waardenburg-Jonker corneal dystrophy
897	Waardenburg-Shah syndrome
280558	WABS
247709	Wagenmann-Froboese syndrome
898	Wagner disease
898	Wagner syndrome
893	WAGR syndrome
90033	wAHA
357332	Wahab syndrome
90033	wAIHA
2379	Waisman syndrome
33226	Waldenström macroglobulinemia
90362	Waldmann disease
1068	Walker-Dyson syndrome
899	Walker-Warburg syndrome
1453	Wallis-Zieff-Goldblatt syndrome
2078	Walt Disney dwarfism
2510	WARBM1
2510	Warburg Micro syndrome
3214	Warburg-Thomsen syndrome
1052	Warburton-Anyane-Yeboa syndrome
96061	Warkany syndrome
90033	Warm AIHA
1541	Warman-Mulliken-Hayward syndrome
280558	Warsaw breakage syndrome
51636	Warts-hypogammaglobulinemia-infections-myelokathexis
69745	Warty dyskeratoma
906	WAS
100067	Waterhouse-Friderichsen syndrome
1046	Water-West syndrome
97282	Watery diarrhea - hypokalemia - achlorhydria
97282	Watery diarrhea - hypokalemic alkalosis
3444	Watson syndrome
33577	WCD
284395	WDFA
97282	WDHA syndrome
99971	WDLS
3447	Weaver syndrome
3446	Weaver-like syndrome
3448	Weaver-Williams syndrome
33577	Weber-Christian disease
33577	Weber-Christian panniculitis
1521	Webster-Deming syndrome
900	Wegener granulomatosis
228254	Weidman juvenile elastoma
3449	Weill-Marchesani syndrome
3344	Weismann-Netter syndrome
3450	Weissenbacher- Zweymuller syndrome
213736	Well-differentiated endocrine neoplasm of the corpus uteri
213736	Well-differentiated endocrine neoplasm of the endometrium
213736	Well-differentiated endocrine tumor of the corpus uteri
213736	Well-differentiated endocrine tumor of the endometrium
284395	Well-differentiated fetal adenocarcinoma of the lung
99971	Well-differentiated liposarcoma
263331	Well-differentiated thymic neuroendocrine carcinoma
146	Well-differentiated thyroid carcinoma
1373	Wellesley-Carman-French syndrome
901	Wells syndrome
2815	Wells-Jankovic syndrome
83330	Werdnig-Hoffmann disease
652	Wermer syndrome
3332	Werner mesomelic syndrome
902	Werner syndrome
1979	Werner-like syndrome due to combined growth factor deficiency
3451	West syndrome
2435	Westerhof-Beemer-Cormane syndrome
83593	Western equine encephalitis
83593	Western equine encephalomyelitis
83476	West-Nile encephalitis
83476	West-Nile fever
681	Westphall disease
952	Weyers acrodental dysostosis
952	Weyers acrofacial dysostosis
51636	WHIM syndrome
3452	Whipple disease
2053	Whistling face syndrome
228290	White fibrous papulosis of the neck
2475	White forelock with malformations
3207	White matter hypoplasia - corpus callosum agenesis - intellectual deficit
171723	White sponge nevus
171723	White sponge nevus of Cannon
1489	Whooping cough
2779	Whyte-Murphy syndrome
3454	Wieacker-Wolff syndrome
116	Wiedemann-Beckwith syndrome

ORPHA Number	Disease name
2156	Wiedemann-Oldigs-Oppermann syndrome
3455	Wiedemann-Rautenstrauch syndrome
319182	Wiedemann-Steiner syndrome
3456	Wildervanck syndrome
903	Willebrand disease
166078	Willebrand disease type 1
166081	Willebrand disease type 2
166096	Willebrand disease type 3
904	Williams syndrome
904	Williams-Beuren syndrome
739	Willi-Prader syndrome
654	Wilms tumor
893	Wilms tumor - aniridia - genitourinary anomalies - intellectual deficit
220	Wilms tumor and pseudohermaphroditism
905	Wilson disease
3459	Wilson-Turner syndrome
3460	Winchester syndrome
169095	Winged helix deficiency
2901	Winged scapula
94087	Winkelmann cytophagic panniculitis
3228	Winkelmann-Bethge-Pfeiffer syndrome
2515	Winship-Viljoen-Leary syndrome
906	Wiskott-Aldrich syndrome
829	Wissler-Fanconi syndrome
2228	Witkop syndrome
101068	Witschel dystrophy
85291	Wittwer syndrome
3237	WL syndrome
247768	WNT4 deficiency
1667	Wolcott-Rallison syndrome
3080	Wolff-Zimmermann syndrome
280	Wolf-Hirschhorn syndrome
3463	Wolfram syndrome
75233	Wolman disease
3464	Woodhouse-Sakati syndrome
2571	Woods-Black-Norbury syndrome
137658	Woods-Crouchman-Huson syndrome
170	Woolly hair
1409	Woolly hair - hypotrichosis - everted lower lip - outstanding ears
79414	Woolly hair nevus
65282	Woolly hair-palmoplantar hyperkeratosis-dilated cardiomyopathy syndrome
65282	Woolly hair-palmoplantar keratoderma-dilated cardiomyopathy syndrome
170	Wooly hair
1409	Wooly hair - hypotrichosis - everted lower lip - outstanding ears
65282	Wooly hair - palmoplantar keratoderma - dilated cardiomyopathy
79414	Wooly hair nevus
65282	Wooly hair-palmoplantar hyperkeratosis-dilated cardiomyopathy syndrome
3465	Worster-Drought syndrome
2790	Worth syndrome
178475	Wound botulism
165955	Wound myiasis
2834	Wrinkled skin syndrome
2834	Wrinkly skin syndrome
1667	WRS
902	WS
897	WS4
163746	WS4 plus
2834	WSS
3466	WT limb-blood syndrome
3411	Wunderlich syndrome
899	WWS
53719	Wyburn-Mason syndrome
96201	X small rings
43	X-ALD
3467	Xanthic urolithiasis
93602	Xanthine dehydrogenase and xanthine aldehyde oxidase dual deficiency
93601	Xanthine dehydrogenase deficiency
93601	Xanthine oxidase deficiency
93601	Xanthine oxidoreductase deficiency
3467	Xanthine stone disease
93601	Xanthinuria type I
93602	Xanthinuria type II
158003	Xanthoma disseminatum
2882	Xanthomatosis with sisterolemia
79433	Xanthous oculocutaneous albinism
79155	Xanthurenic aciduria
93602	XDH and AOX dual deficiency
93601	XDH deficiency
293621	XECD
910	Xeroderma pigmentosum
276249	Xeroderma pigmentosum complementation group A
276252	Xeroderma pigmentosum complementation group B
276255	Xeroderma pigmentosum complementation group C
276258	Xeroderma pigmentosum complementation group D
276261	Xeroderma pigmentosum complementation group E
276264	Xeroderma pigmentosum complementation group F
276267	Xeroderma pigmentosum complementation group G
90342	Xeroderma pigmentosum variant
220295	Xeroderma pigmentosum/Cockayne syndrome complex
75496	XGPT deficiency
181	XHED
101088	XHIGM
3469	XK aprosencephaly
452	XLAG syndrome
596	XLCNM
79278	XLDPP
264580	XLG
89936	XLH
461	XLI
2182	X-linked acqueductal stenosis
43	X-linked adrenoleukodystrophy
47	X-linked agammaglobulinemia
43	X-linked ALD
88917	X-linked Alport syndrome
85278	X-linked Angelman-like syndrome
181	X-linked anhidrotic ectodermal dysplasia
85297	X-linked ataxia-deafness syndrome
85292	X-linked ataxia-dementia syndrome
139583	X-linked auditory neuropathy with peripheral sensory neuropathy type 1
1131	X-linked branchial arch syndrome
481	X-linked bulbospinal amyotrophy
111	X-linked cardioskeletal myopathy and neutropenia
329235	X-linked central congenital hypothyroidism with late-onset macroorchidism
329235	X-linked central congenital hypothyroidism with late-onset testicular enlargement
596	X-linked centronuclear myopathy
247765	X-linked cerebellar ataxia
163961	X-linked cerebral - cerebellar - coloboma syndrome
139396	X-linked cerebral adrenoleukodystrophy
64747	X-linked Charcot-Marie-Tooth disease
101075	X-linked Charcot-Marie-Tooth disease type 1
101076	X-linked Charcot-Marie-Tooth disease type 2
101077	X-linked Charcot-Marie-Tooth disease type 3
101078	X-linked Charcot-Marie-Tooth disease type 4
99014	X-linked Charcot-Marie-Tooth disease type 5
352675	X-linked Charcot-Marie-Tooth disease type 6
324601	X-linked cleft palate and ankyloglossia
98888	X-linked complex spastic paraplegia
1497	X-linked complicated corpus callosum dysgenesis
98888	X-linked complicated spastic paraplegia
306617	X-linked complicated spastic paraplegia type 1

ORPHA Number	Disease name
90001	X-linked cone dysfunction syndrome with myopia
95702	X-linked congenital adrenal hypoplasia
79495	X-linked congenital generalized hypertrichosis
565	X-linked copper deficiency
1661	X-linked corneal dermoid
52503	X-linked creatine transporter deficiency
85453	X-linked cutaneous amyloidosis
198	X-linked cutis laxa
85321	X-linked deafness - intellectual deficit syndrome
383	X-linked deafness type 2
139557	X-linked dHMN
1018	X-linked diffuse leiomyomatosis - Alport syndrome
1145	X-linked distal arthrogryposis multiplex congenita
139557	X-linked distal hereditary motor neuropathy
139557	X-linked distal spinal muscular atrophy
163966	X-linked dominant chondrodysplasia - hydrocephaly - microphthalmia
35173	X-linked dominant chondrodysplasia punctata
163966	X-linked dominant chondrodysplasia, Chassaing-Lacombe type
93951	X-linked dominant intellectual deficit - epilepsy
139557	X-linked dSMA
373	X-linked dysplasia gigantism syndrome
53351	X-linked dystonia-parkinsonism
75497	X-linked Ehlers-Danlos syndrome
98863	X-linked Emery-Dreifuss muscular dystrophy
293621	X-linked endothelial corneal dystrophy
85294	X-linked epilepsy - learning disabilities - behavior disorders
139583	X-linked hereditary sensory and autonomic neuropathy with deafness
139583	X-linked HSAN with deafness
2182	X-linked HSAS
2182	X-linked hydrocephalus with stenosis of aqueduct of Sylvius
101088	X-linked hyper-IgM syndrome
181	X-linked hypohidrotic ectodermal dysplasia
89936	X-linked hypophosphatemia
89936	X-linked hypophosphatemic rickets
461	X-linked ichthyosis
231692	X-linked IGHD
37042	X-linked immune dysregulation - polyendocrinopathy - enteropathy
317476	X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia
2571	X-linked immunoneurologic disorder
16	X-linked incomplete achromatopsia
1145	X-linked infantile spinal muscular atrophy
93952	X-linked intellectual deficiency, Hedera type
85338	X-linked intellectual deficit - ataxia - apraxia
324410	X-linked intellectual deficit - cardiomegaly - congestive heart failure
137831	X-linked intellectual deficit - cerebellar hypoplasia
85330	X-linked intellectual deficit - corpus callosum agenesis - spastic quadriparesis
85278	X-linked intellectual deficit - craniofacial dysmorphism - epilepsy - ophthalmoplegia - cerebellar atrophy
94083	X-linked intellectual deficit - dystonia - dysarthria
2076	X-linked intellectual deficit - epilepsy
85282	X-linked intellectual deficit - epileptic seizures - hypogenitalism - microcephaly - obesity
3459	X-linked intellectual deficit - gynecomastia - obesity
59	X-linked intellectual deficit - hypotonia
73220	X-linked intellectual deficit - hypotonic face
251383	X-linked intellectual deficit - microcephaly - cortical malformation - thin habitus
163937	X-linked intellectual deficit - microcephaly - pontocerebellar hypoplasia
163971	X-linked intellectual deficit - microcephaly - testicular failure
163956	X-linked intellectual deficit - nail dystrophy - seizures
52503	X-linked intellectual deficit - seizures - short stature - midface hypoplasia
163982	X-linked intellectual deficit - spastic quadriparesis
3242	X-linked intellectual deficit due to PQBP1 mutations
67045	X-linked intellectual deficit with isolated growth hormone deficiency
776	X-linked intellectual deficit with marfanoid habitus
85274	X-linked intellectual deficit, Ahmad type
1193	X-linked intellectual deficit, Atkin type
85293	X-linked intellectual deficit, Cabezas type
93947	X-linked intellectual deficit, Golabi-Ito-Hall type
163937	X-linked intellectual deficit, Najm type
93945	X-linked intellectual deficit, Porteous type
3242	X-linked intellectual deficit, Renpenning type
85278	X-linked intellectual deficit, South African type
85326	X-linked intellectual deficit, Stoll type
93950	X-linked intellectual deficit, Sutherland-Haan type
231692	X-linked isolated growth hormone deficiency
90625	X-linked isolated neurosensory deafness type DFN
90625	X-linked isolated neurosensory hearing loss type DFN
90625	X-linked isolated sensorineural deafness type DFN
90625	X-linked isolated sensorineural hearing loss type DFN
792	X-linked juvenile retinoschisis
79447	X-linked lethal multiple pterygium syndrome
452	X-linked lissencephaly - agenesis of the corpus callosum - genital anomalies
2148	X-linked lissencephaly type 1
452	X-linked lissencephaly with abnormal genitalia
452	X-linked lissencephaly with ambiguous genitalia
2442	X-linked lymphoproliferative disease
1131	X-linked mandibulofacial dysostosis
1131	X-linked mandibulofacial dysostosis with limb anomalies
59306	X-linked McLeod syndrome
319605	X-linked mendelian susceptibility to mycobacterial diseases
319623	X-linked mendelian susceptibility to mycobacterial diseases due to CYBB deficiency
319612	X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency
319612	X-linked mendelian susceptibility to mycobacterial diseases due to NEMO deficiency
776	X-linked mental retardation with marfanoid habitus
383	X-linked mixed conductive and neurosensory deafness
383	X-linked mixed conductive and neurosensory hearing loss
383	X-linked mixed conductive and sensorineural deafness
383	X-linked mixed conductive and sensorineural hearing loss
383	X-linked mixed deafness with perilymphatic gusher
319605	X-linked MSMD
319623	X-linked MSMD due to CYBB deficiency
319612	X-linked MSMD due to IKBKG deficiency
319612	X-linked MSMD due to NEMO deficiency
25980	X-linked myopathy with excessive autophagy
178461	X-linked myopathy with postural muscle atrophy

ORPHA Number	Disease name
85334	X-linked neurodegenerative syndrome, Bertini type
85336	X-linked neurodegenerative syndrome, Hamel type
314978	X-linked non progressive cerebellar ataxia
777	X-linked non-specific intellectual deficit
777	X-linked non-syndromic intellectual deficit
90625	X-linked nonsyndromic neurosensory deafness type DFN
90625	X-linked nonsyndromic neurosensory hearing loss type DFN
90625	X-linked nonsyndromic sensorineural deafness type DFN
90625	X-linked nonsyndromic sensorineural hearing loss type DFN
306597	X-linked Opitz BBB/G syndrome
306597	X-linked Opitz G/BBB syndrome
306597	X-linked Opitz syndrome
1175	X-linked progressive cerebellar ataxia
1652	X-linked recessive hypercalciuric hypophosphatemic rickets
93952	X-linked recessive intellectual deficit - epilepsy
83648	X-linked recessive intellectual deficit - macrocephaly - ciliary dysfunction
1652	X-linked recessive nephrolithiasis
54	X-linked recessive ocular albinism
85453	X-linked reticulate pigmentary disorder with systemic manifestations
1852	X-linked retinal dysplasia
792	X-linked retinoschisis
86788	X-linked severe congenital neutropenia
75563	X-linked sideroblastic anemia
2802	X-linked sideroblastic anemia - ataxia
100997	X-linked spastic paraplegia type 16
99015	X-linked spastic paraplegia type 2
171607	X-linked spastic paraplegia type 34
1145	X-linked spinal muscular atrophy type 2
85297	X-linked spinocerebellar ataxia type 3
85292	X-linked spinocerebellar ataxia type 4
93349	X-linked spondyloepimetaphyseal dysplasia
168544	X-linked spondylometaphyseal dysplasia
383	X-linked stapes gusher syndrome
852	X-linked thrombocytopenia with normal platelets
776	XLMR with marfanoid habitus

ORPHA Number	Disease name
596	XLMTM
54	XLOA
306597	XLOS
2442	XLP
85453	XLPDR
792	XLRS
231393	XLTT
25980	XMEA
317476	XMEN
93601	XO deficiency
93601	XOR deficiency
910	XP
220295	XP/CS complex
261476	Xp21 microdeletion syndrome
284180	Xp22.13p22.2 duplication syndrome
1643	Xp22.3 microdeletion syndrome
276249	XPA
276252	XPB
276255	XPC
276258	XPD
276261	XPE
276264	XPF
276267	XPG
90342	XPV
314389	Xq12-q13.3 duplication syndrome
1018	Xq22.3 microdeletion syndrome
261483	Xq27.3q28 duplication syndrome
261483	Xq27.3-q28 microduplication syndrome
243	XX female gonadal dysgenesis
2855	XX gonodal dysgenesis - deafness
393	XX, male syndrome
243	XX-GD
3375	XXX
983	XY gonadal agenesis
168558	XY sex reversal - adrenal failure
2843	Xylitol dehydrogenase deficiency
75496	Xylosylprotein 4-beta-galactosyltransferase deficiency
99829	Yellow fever
99829	Yellow Jack
662	Yellow nail syndrome
79434	Yellow oculocutaneous albinism
707	Yersiniosis
99829	YF

ORPHA Number	Disease name
876	Yolk sac tumor
252006	Yolk sac tumor of the central nervous system
2255	Yorifuji-Okuno syndrome
3240	Yoshimura-Takeshita syndrome
314485	Young adult-onset dHMN
314485	Young adult-onset distal hereditary motor neuropathy
2828	Young adult-onset Parkinsonism
3471	Young syndrome
3055	Young-Hugues syndrome
2828	Young-onset Parkinson disease
3472	Yunis-Varon syndrome
319213	Zambian hemorrhagic fever
98912	ZASPopathy
98912	ZASP-related myofibrillar myopathy
97240	Zebra body myopathy
217017	Zechi-Ceide syndrome
912	Zellweger syndrome
50812	Zellweger-like syndrome without peroxisomal anomalies
911	Zeta-associated-protein 70 deficiency
3301	Zimmer phocomelia
3473	Zimmermann-Laband syndrome
1775	Zinsser-Engman-Cole syndrome
3253	Zlotogora-Ogur syndrome
101036	Zlotogura-Martinez syndrome
913	Zollinger-Ellison syndrome
98995	Zonular cataract
2835	Zori-Stalker-Williams syndrome
912	ZS
3474	Zunich-Kaye syndrome
295187	Zygodactyly type 1
295189	Zygodactyly type 2
295191	Zygodactyly type 3
295193	Zygodactyly type 4
295193	Zygodactyly, Castilla type
295189	Zygodactyly, Lueken type
295191	Zygodactyly, Montagu type
295187	Zygodactyly, Weidenreich type
73263	Zygomycosis

For any questions or comments, please contact us: *contact.orphanet@inserm.fr*

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